UNIVERSITY OF CAPE COAST

MATERNAL AND PERINATAL CONTRIBUTING FACTORS TO CONGENITAL ABNORMALITIES IN CHILDREN BELOW FIVE YEARS

A STUDY AT THE KORLE BU TEACHING HOSPITAL

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DECLARATION

Candidates Declaration

I hereby declare that this thesis is the result of our own original research and that no part of it has been presented for another degree in this university or elsewhere.

Candidates Signature …… Date……

Name …….

Supervisors Declaration

We hereby declare that the preparation and presentation of the thesis were supervised in accordance with the guidelines on supervision of thesis laid down by University of Cape Coast.

Principal Supervisor's Signature…… Date……

Name:……

Co-Supervisor's Signature …… Date……

Name:……

ABSTRACT

The main purpose of the study was to examine the maternal and perinatal contributing factors of congenital abnormalities in children below five years. The study was carried out at the Korle bu Teaching Hospital in the Accra Metropolis. This study was an exploratory study which used quantitative method. The participants for the study were selected using convenience sampling. One hundred and eighty one questionnaires were distributed to mothers of children with congenital abnormalities who were below five years at the hospital. The data collected was coded and analyzed using the Statistical Package for Social Sciences (SPSS). Findings from the study suggested that, maternal and perinatal contributing factors including illicit drug use and herbal medication have a very significant and negative influence on the unborn child. Factors such as regular attendance to antennal clinic, healthy nutrition and taking of prescribed medication at the clinic also have significant and positive contribution to the good health of the baby. The study recommended that mothers should be encouraged to attend antenatal clinics regularly during pregnancy and health facilities should screen all pregnant women for congenital malformations for early detection and management. The psychological impact of congenital abnormalities on the family and educational programs can be organized to create awareness on the various types of congenital abnormalities and their management. It is anticipated that if these measures are taken into consideration, it will help to create awareness of congenital abnormalities, strengthen existing management procedures and help to reduce child morbidity and mortality in Ghana.

KEY WORDS

Congenital abnormality

Contributing factors

Maternal

Perinatal

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DEDICATION

To my family

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LIST OF ACRONYMS

AEDs	Antiepileptic Drugs
ARBD	Alcohol-Related Birth Defects
ARND	Alcohol-Related Neurodevelopmental Disorder
CBZ	Carbamazepine
DNA	Deoxyribonucleic Acid
DS	Down syndrome
G6PD	Glucose-6 Phosphate Dehydrogenase
GBS	Guillian- Barre Syndrome
IUGR	Intra Uterine Growth Retardation
MIM	Mendelian Inheritance in Man
NTD	Neural Tube Defects
OFC	Orofacial Disorders
OXC	Oxacabamazepine
PHT	Phenytoin
PRM	Primidone
RNA	Ribonucleic Acid
TPM	Topiramide
UK	United Kkingdom
USA	United States of America
VCFS	Velocardiofacial Syndrome
VPS	Valporate
ZNS	Zonisamide

CHAPTER ONE

INTRODUCTION

Background to the Study

Pregnancy is not just a matter of waiting to give birth (WHO, 2005), but often a defining phase in a woman's life to be happy and fulfilled as an individual living in the society (United Nations Development Programme [UNDP], 2010). According to the WHO (2005), the expectation and hope of every pregnant woman is to deliver a healthy baby without any complication or defects before, during and at the end of the pregnancy. Studies have shown that when a mother gives birth with complications or defects, it generates feelings of pain, despair, grief and anxiety (Taksande, Vilhekar, Chaturvedi & Jain, 2010; Christianson, Howson & Modell, 2009). Often than not, antenatal care is to detect, prevent and treat conditions or defects that threaten the health of the fetus (new born) and/or the mother, and to help women approach pregnancy and birth with positive experiences.

However, Taksande et al. (2010) opined that there were some hidden defects that may not surface during antenatal until later years of development. Congenital anomaly is any morphological, functional, biochemical or molecular defects that may develop in the embryo and fetus from conception until birth or at birth (WHO, 2000). The impact of defects on the fetus and newborn infants is great as they are responsible for 495,000 deaths worldwide (Eurocat Working Group, 2011 According to the WHO (2012), major causes of these defects are congenital in nature.). To the Eurocat, many nurse researchers and theorists were

in agreement that majority of these deaths occurred during the first year of life of these infants and thus, the main reason for the infant mortality rate in 2011(Alfaro-LeFevre, 2010, Berman & Snyder, 2012).

Congenital anomalies can be caused by single gene defects, chromosomal disorders, multi-factorial inheritance, environmental teratogens and micronutrient deficiencies. Congenital anomalies are important causes of infant and childhood deaths, chronic illness and disability. An estimated 303 000 newborns die within 4 weeks of birth every year, worldwide, due to congenital anomalies. Congenital anomalies can contribute to long-term disability, which may have significant impacts on individuals, families, health-care systems, and societies (WHO, 2016). The most common, severe congenital anomalies are heart defects, neural tube defects and Down syndrome. Although congenital anomalies may be the result of one or more genetic, infectious, nutritional or environmental factors, it is often difficult to identify the exact causes. Some congenital anomalies can be prevented. Vaccination, adequate intake of folic acid or iodine through fortification of staple foods or supplementation, and adequate antenatal care are just examples of prevention methods.

Ramagopalan et al. (2005); and Arnold and Christopher (2006) identified the most common body systems involved in congenital anomalies to include musculoskeletal, central nervous system, gastro intestinal system and cardiovascular system with the least affected system being the urogenital system. Congenital anomalies have emerged as the major childhood health problem, affecting approximately 1 in every 33 infants and are the fourth most common

cause of neonatal deaths in Africa. It is also the leading cause of about 3.2 million birth defect-related disabilities every year (WHO, 2012).

Many people who are ignorant about this condition tend to associate the phenomenon with several misconceptions. According to Nyasor (2012) most Africans believed that the possible causes of their children's condition have spiritual dimensions; gods on a visit. This intends influence most parents to shirk their responsibilities of providing the financial means to seek for the needed treatment for the child. Jowi (2013) had similar view with parents that congenital abnormality is a strain on child-parent relation, family routines and also other siblings in the family. Parents often fear to disclose their child's condition to friends and relatives because they have experienced a sense of shame, self-blame and rejection. They consequently withdraw from their relatives and social circle.

Statement of the Problem

The prevalence of congenital abnormities worldwide is estimated at 3-7%, but actual numbers vary widely between countries (Singh & Gupta, 2009; UNDP, 2010). For instance, in the United States and Canada where congenital anomalies are diagnosed intra uterine and aborted, the prevalence is estimated to be around 2-2.5% of all live births (Perinatal Health Report, 2002; Sekhobo & Druschel, 2001). However, in Asia, African and other developing countries the magnitude and incidence of congenital anomalies varies from 2.5% to 20% of infants at birth (Shawky and Sadik, 2011). For instance, in the studies of Shawky and Sadik (2011) it is reported that the prevalence of this anomaly is around 2.5% in India, about 11.4% in Pakistan, around 16% to 20% in African and West Africa. Ghana

is no exception to these findings as in 2010, the prevalence rate of neurological cases of 1.8% was recorded in the three northern regions, sidelining the unreported cases at health centers (Korle-Bu Hospital, 2013).

Some studies have shown that there are some predisposing factors to congenital anomaly, such that exposure to these factors makes this anomaly likely (Ekwere, Meneil, Agim & Jeminiwa, 2011; Taksande et al., 2010). According to UNDP (2010); WHO (2000); and Berman and Snyder (2012) predisposing factors include exposure to X-ray, alcohol and substance abuse, single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and micronutrient deficiencies. It has also been proved that this anomaly has caused the death of new born. Eurocat Working Group (2011) mentioned that the impact of defects on the fetus and newborn infants is great as they are responsible for 495,000 deaths worldwide. Knowledge and medical improvement is one way of minimizing or reducing the burden on the incidences of congenital abnormality.

Though, there is the recognition of the critical need to promote awareness of the diseases, very little has been done in terms of research work in Ghana and other developing countries (WHO, 2012). Additionally, literature on contributing and possible risk factors for congenital abnormality in Africa and for that matter, Ghana, is limited. Also, with the high index of suspicion based primarily on a careful family and maternal history, the understanding of the numerous causative factors are important in identifying congenital abnormalities, many of which can be detected only if one is searching for them. This study will therefore seek to investigate the maternal and perinatal risk factors of congenital abnormalities in

children under five years at the Child Health Department, Korle-Bu Teaching Hospital.

Purpose of the Study

The purpose of the study was to investigate the maternal and perinatal contributing factors of congenital abnormalities in children under five years at the Child Health Department of the Korle Bu Teaching Hospital.

Specific Objectives are:

1. To identify the types of congenital abnormalities seen at the korle bu teaching hospital
2. To find the maternal and perinatal characteristics that have contributed to congenital abnormalities in children below five years.
3. Assess the possible suggestions to minimize the risk factors of congenital abnormalities in children below five years

Significance of the Study

The findings of the study will help to create awareness of the maternal and perinatal risk factors of congenital abnormalities to reduce childhood mortalities and morbidities. The findings will also help the ministry of health and policy makers to devise strategies to reduce the factors and also strengthen existing ones. The outcome of the findings will give directions for further research studies on congenital abnormalities and also contribute to the body of knowledge on maternal and child health in general.

Definition of Terms

Congenital abnormality: is a disease condition that affects a baby from birth

Contributing factors: is anything that predisposes someone to a particular disease, injury or death

Maternal: is a relation obtained through ones mother

Perinatal: is any occurrence surrounding the period of pregnancy including antenatal, delivery and postnatal

Delimitations of the Study

There are multiple factors that contribute to congenital abnormalities. These include genetic factors; environmental factors and maternal factors. The study was focused only on the maternal and perinatal contributing factors of congenital abnormalities in children below five years at the Child Health Department of the Korle-Bu Teaching Hospital. It is also to conduct an exploratory study on only cases reported at the child health department.

Limitations of the Study

The limitations of the study included inability to have access to the medical records on time for confirmation of child's history. Also some of the information received from the mothers did not correspond with information in the child's medical record books. The researcher also needed to wait for particular days that mark clinic days in order to get research participants who fall within the inclusion criteria.

Organization of the Study

The study is organized into five chapters. The first chapter is the introduction and it covers the background to the study, statement of the problem, purpose of the study, research questions, and significance of the study, delimitations and organization of the study. Chapter two also reviews related literature relevant to the study. The third chapter presented the methodology of the study. It includes; the research design, the study area, population, sample and sampling procedure, the instrument for data collection, administration of instrument and procedures to be used for analyzing the data collected. Chapter four covers the results and discussion of the study while the last chapter. Chapter Five, presents the summary, conclusions and recommendations. The study would also provide areas for further studies.

CHAPTER TWO

REVIEW OF RELATED LITERATURE

Introduction

This chapter presents review of theories and models relating to the history of congenital abnormalities, and empirical studies related to maternal and perinatal characteristics of congenital abnormalities in children.

History of Teratology and Congenital Abnormality

Teratogens are substances that may produce physical or functional defects in the human embryo or fetus after the pregnant woman is exposed to the substance. Alcohol and cocaine are examples of such substances. Exposure to the teratogen affects the fetus or embryo in a variety of ways, such as the duration of exposure, the amount of teratogenic substance, and the stage of development of the embryo or fetus during the exposure. Teratogens may affect the embryo or fetus in a number of ways, causing physical malformations, problems in the behavioral or emotional development of the child, and decreased intellectual quotient (IQ) in the child. Additionally, teratogens may also affect pregnancies and cause complications such as preterm labors, spontaneous abortions, or miscarriages. Teratogens are classified into four types: physical agents, metabolic conditions, infection, and finally, drugs and chemicals (Saal et al., 1985).

The word teratogen originates from the Greek word for monster, teratos. Isidore, Geoffroy and Saint-Hilaire, defined it in 1832 in (General and Particular History of Structural Monstrosities in Man and Animals). People had sought explanations for abnormal human and animal development, however, for

centuries, and they had developed different theories about the causes for the abnormalities. In Babylon, many said that infants with congenital malformations, or structural abnormalities present at birth, were constellations in human forms as well as fortune-tellers. Many early Hebrews said that abnormal development resulted from the deformed person's association with the devil.

Aristotle, who lived in Athens, Greece in the fourth century, B.C., deemed birth defects as disturbances in reproduction rather than supernatural occurrences. Aristotle and Hippocrates claimed that a pregnant woman's experiences or emotions, which became maternal impressions, can affect the formation of the fetus. The theory of maternal impressions persisted until the early 1900s. At the beginning of the 19th century, Johann Friedrich Meckel, asserted that deviations from the normal developmental process caused malformations. Meckel his Journal of Anatomical Varieties, Finer and Pathological Anatomy examined anatomical defects and their causes. Meckel categorized abnormal development into four basic types: reduced or absent body parts (insufficient generative energy), enlarged or multiple body parts (excessive energy), aberration of form and of position, and hermaphroditism, which included deformities such as ambiguous genitalia (Saal et al., 1985).

Following Meckel, scientists in the nineteenth century began experimental studies to detect teratogens. Etienne Geoffroy Saint-Hilaire in Paris experimented on chick eggs by subjecting them to pricking, inversion, jarring, and abnormally high or low temperatures to study the resulting malformations. He believed that certain manipulations could invoke specific deformations. Although deformities

materialized, Saint-Hilaire did not identify their exact causes. Isidore in 1832 then reported the results of the experiments to his Treatise on Teratology (cited in Anat & Rodriguez, 1987). Other scientists Camille Dareste also experimented with teratogens and successfully produced abnormalities in chick embryos. Scientists in the twentieth century classified teratogens into four categories, physical, chemical, or infectious agents and maternal conditions (Anat & Rodriguez, 1987). Physical agents include ionizing radiation or other agents that contribute to hyperthermia, or elevated body temperature. Scientists gathered evidence in the early 1990s which supported their theories that there was an association between the high fevers of pregnant women and congenital abnormalities, such as cardiac defects, abdominal wall defects, or a disruption of the enervation of the large intestine.

Metabolic conditions affecting pregnant females such as malnutrition, diabetes, and thyroid disorders are a second category of teratogens. Metabolic conditions are abnormalities in the chemical process of producing energy from food, and thereby affect the development and function of the body. If a pregnant woman is malnourished, then her fetus likely lacks the nutrients essential for its development. In the case of diabetes, low blood sugar, or hypoglycemia, may cause fetal malformations. Thyroid disorders can also cause a number of teratogenic effects to a developing fetus, as well as adverse effects on pregnancy such as miscarriage, premature separation of the placenta from the uterine wall (placental abruption), preterm labour, and lower IQ scores in the children. Infections such as those caused by rubella virus, herpes simplex virus, and

syphilis, are a third kind of teratogen. Norman McAlister Gregg in 1941 (as cited in Singh & Gupta, 2009) made a connection between the virus and congenital malformations and contributed to discoveries of a teratogen that was not a manufactured chemical.

The fourth kind of teratogen includes drugs and chemicals the pregnant female ingests such as alcohol, cocaine, thalidomide, Agent Orange, and vitamin A and its derivatives, called retinorids. Fred Hale (1933) found that offspring had various congenital malformations such as anophthalmia, which is the absence of one or both eyes, and cleft palate. Hale's experiments established that an absence or deficiency of a nutrient could produce severe congenital malformations in mammalian embryos. Alcohol, which also falls under the fourth category of teratogen, can cause Fetal Alcohol Syndrome (FAS) in children born to women who drank too much alcohol while pregnant. FAS can cause defects such as minor facial abnormalities and damage to the brain, which consequently leads to learning, behavioral, and cognitive abnormalities (Singh & Gupta, 2009).

Theoretical Framework

A theory is a well-established principle that has been developed to explain some aspect of the natural world (Greener, 2008). A theory arises from repeated observations and testing and incorporates facts, laws, predictions, and tested hypotheses that are widely accepted (Cooper, 1989). A theoretical framework is used to limit the scope of the relevant data, by focusing on specific variables and defining the specific viewpoint that the researcher will take into account (Dawson, 2002).

Arrested Development Theory

Arrest embryonic development involves the cessation of active cell division and metabolic activity, and the capability of an animal to arrest embryonic developments results in temporal plasticity of the duration of embryonic period. Nineteenth-century teratologists contributed to the transcendentalists' pursuit of anatomy's general laws by describing and classifying examples of monstrous births as though they were representatives of distinct groups or species. According to Isidore-Geoffroy (1832 as cited in Tantibanchachai, 2014) was one of the proponents of this arrested foetal development theory. Geoffroy and Meckel described the theory as a law which was based on parallelism between embryological development and a linear taxonomy of adult organisms: monsters resembled the adult forms of lower species. According to Geoffroy, monsters caused by arrest of development were "des embryon spermanens," and attributed human monstrosities to arrests of development at one of the progressive stages through which the human embryo passes: the least recognizably human monsters (abnormalities) were due to arrests earliest in development (Tantibanchachai, 2014).

Meckel-Serres Law

Meckel and Serres in the 1830s were few authors who supported the idea of arrested development as a cause to human monstrosities (Pahomov & Porter, 2012). Meckel and Serres both argued that fetal deformities result when development prematurely stops, and they argued that these arrests characterized lower life forms, through which higher order organism's progress during normal

development. The concept that the embryo of higher order organisms progress through successive stages in which they resemble lower level forms is called recapitulation. The recapitulation ideas of Meckel and Serres became part of the debate about how to explain morphological similarities between species. The concept of scalanaturae, or the great chain of being, had an early proponent in Aristotle. Meckel and Serres took these ideas of a scalanaturae and recapitulation and applied them to their own embryological studies. Meckel outlines the idea that embryonic stages of higher forms recapitulate the forms of animals that reside lower on the great chain of being. Meckel used malformations, which he saw as the results of early terminations to development, to help support his theory of recapitulation (Tantibanchachai, 2014).

Serres, also argued that the developing human brain progressed through the hierarchy of nature as it developed; at first it looked like the brain of a fish, then a reptile, then a bird, and lastly a general mammalian brain before finally settling into the form of a human brain (Tantibanchachai, 2014). This order of brain development appeared to mirror the scalanaturae. He postulated that a formative force propelled the development of species, but the organisms in lower species had too little of the formative force to have their organs develop into the more complex organs found in higher species. For this theory, humans are the most complex life form because they have the greatest amount of what some later called Serres' force.

However Karl Ernst von Baer in 1827 criticized the recapitulation theories of Meckel and Serres "On the Developmental History of the Animals

Observations and Reflections" (Simpson, 1980; Shawky & Sadik, 2011). Von Baer disagreed with the theory that Meckel and Serres had constructed to explain similarities in embryonic development across the animal kingdom. Von Baer opposed the strict linearity that Meckel and Serres embraced, which saw all organisms placed on a single chain of life. Instead, von Baer embraced the separation of the animal kingdom into four distinct archetypes, or fundamental body plans: the radiate, like the starfish; the mollusca, like clams and octopus; the articulate, like insects and lobsters; and the vertebrata, like fish and humans. According to Shawky and Sadik (2011) Von Baer classified organisms into each of the four archetypes according to how those organisms developed from embryos. Von Baer reasoned that because animals could be divided into four archetypes, embryos could not recapitulate all lower forms throughout their development. Instead, von Baer argued that embryos appeared similar to their archetype at the beginning of development, and grew more specialized over time. Von Baer's account of the relationship between development and the natural hierarchy of animals, articulated in his 1828 text, formed the basis of what later scientists called von Baer's Laws.

Biogenetic Law

Following the dismissal by von Baer and Richard Owen, the recapitulation ideas of Meckel and Serres fell out of favor among scientists beginning in the late 1830s. In 1859, Charles Darwin, argued for the theory of common descent among species in his book, On the Origin of Species (Simpson, 1980). This prompted Ernst Haeckel to resurrect the main points of Meckel and Serres. Haeckel's new

iteration of Meckel's and Serres' ideas of recapitulation, for Haeckel called the biogenetic law, abandoned the explicit connection to the scalanaturae (Ramagopalan et al., 2005). Instead, Haeckel embraced Darwin's theory of common descent as the framework that unites all organisms. The biogenetic law connected the study of embryonic development, called ontogeny, with the study of the relationships of descent between species, called phylogeny.

The favourable reception of arrested development was largely due to its ability to supply aetiology for congenital malformations, in which respect it was seen as having filled the 'gap' left by the previous theory of congenital malformation "time-honoured theory of maternal impressions". According to North (cited in Pahomov & Porter, 2012), arrested development was a secondary cause that was implicated in the production of birth defects caused primarily by disease or other aetiologies. North noted that in cases of anencephaly the mother had often received a blow, or pressure to the abdomen, during pregnancy, which he thought could precipitate a developmental arrest. Simpson (1980) also in previous studies used Arrested Development Theory to explain hermaphroditism, an idea developed from his reading of William Harvey which asserts that in many cases, developmental arrest was secondary to intrauterine disease. In medicine, the principle was applied by analogy to tissues as well as embryos: morbid changes of bones associated with restricted growth were attributed to arrested development.

Retrogression Development Theory

Knox's key example of retrogression was one of the most celebrated specimens in his collection, the "tiger arm", which was on display in 1841 (Bates, 2005). The tiger arm – actually a human arm with an aberrant humeral foramen normally found in big cats – showed an anomaly that could not possibly be ascribed to developmental arrest, since no such foramen is observable in the human embryo at any stage. On the basis of this, Knox justified his rejection of arrested development in favour of Retrogressive Developments (Tubbs, 1995; Bates, 2005). Retrogressive development of the embryo produced offspring that showed characteristics of ancestral forms, but not necessarily forms that the embryo normally passed through during its development, thus it did not require acceptance of the theory of recapitulation. Whereas developmental arrest was ontogenesis stalled at an intermediate stage, retrogression represented a reawakening of latent developmental potential normally held in check by the tendency to heredity (Tubbs & Oakes, 1997).

Thomas Traill (1840) popularized retrogressive rather than arrested development by defining a monster as "a birth or production of a living being degenerating from the proper and usual disposition of the species to which it belongs" (Arztebl, 2006; p, 13). Traill speculated that degeneration could operate as a secondary cause in monsters that were primarily caused by forces external to the embryo. By the end of the nineteenth century, retrogression had replaced arrested development as the dominant theory of teratogeny (Christianson et al, 2009). Regression was also applied outside embryology, as arrested development

had been, as a pathogenetic mechanism of disease. In 1847, William Addison wrote his "law of the morphology or metamorphosis of the textures of the human body" which interpreted disease processes. One example of human "retrograde metamorphosis" was rickets. Addison also noted other conditions characterized by "replacement of a later or higher texture by one of an earlier or more primary type… as when the structure was evolving from its embryo state," a change reminiscent of the modern concept of differentiation (Christianson et al, 2009 p, 9).

In conclusion, it must be noted that contemporary theories of congenital abnormalities are abnormality-specific (heart, brain, instestinal, skeletal abnormalities). For instance a theory for congenital heart disease cannot be adopted in general to explain other causes congenital abnormality cases not related to the heart. However this was a gap which was non-existent when the early theories of Arrest Fetal Development and Retrogressive Theories were developed by the early scientist.

Definition of Congenital Anomalies

According to the World Health Organization (WHO) the term congenital anomaly includes any morphological, functional, biochemical or molecular defects that may develop in the embryo and foetus from conception until birth that is present at birth, whether detected at that time or not (Shawky & Sadik, 2011). Structural defects of prenatal origin are classified according to the cause, timing, and extent of the developmental disturbance. These include malformations (defective organogenesis), dysplasia (abnormal cell or tissue structure) and

deformations (mechanically induced changes of normal tissue) (Queiber & Spranger, 2006).

Major or severe anomalies impair function or greatly interfere with cosmetic value. They may be life threatening hence needing immediate management. If not corrected early major anomalies could have a negative impact on child's well-being and development. Minor anomalies occur in non-vital organs causing little or no functional effects. They do not cause distress in the newborn and hence there is no urgency for their correction in the neonatal period (Shamin, 2010).

Maternal and perinatal contributing factors of Congenital Abnormalities

The aim of this aspect of the paper is to provide a contextual to the evidence review process by examining the contribution of congenital abnormalities in children by other researchers. Congenital anomalies, congenital abnormalities, birth defects and congenital malformations are all terms used to describe developmental disorders of the *embryo* and *fetus*. Congenital anomalies are the second commonest cause of infant deaths in the world. Sant´e Canada (2002) mentions that some severe congenital anomalies are close to 100% lethal.

Malformations have multi-factorial aetiologies. There are several hundred separate anomalies which fall under these headings including structural, functional, metabolic and hereditary conditions. Winterbottom (2001) documented that every year an estimated 6 percent of total births worldwide are born with a serious birth defect of genetic or partially genetic origin. Additionally he said hundreds of thousands more are born with serious birth defects of post-

conception origin, including maternal exposure to environmental agents (teratogens) such as alcohol, rubella, syphilis and iodine deficiency that can harm a developing fetus (Winterbottom, 2001).

The March of Dimes foundation (2006) also in their research asserted that birth defects are of genetic origins which are congenital heart defects, neural tube defects, hemoglobin disorders, Down syndrome and glucose-6-phosphate dehydrogenase (G6PD) deficiency. Congenital malformations can be inherited or acquired. Congenital abnormalities can be attributed to known genetic conditions, teratogenic effects of chemicals and infection and finally a third cause which is unknown.

However, there is no single universally accepted system of classification of abnormalities or indeed a single agreed definition of what constitutes a congenital abnormality. It is clear from the above synopsis that congenital abnormalities originates from four main factors; Genetic factors, environmental factors, Multifactorial factor from both genetics and environment and an unknown factor. For the purpose of this study the researcher has narrowed review to these four main factors taking into consideration related theories and existing literature available.

Genetic Factors

From theoretical considerations there are a number of groups of genetic factors which might be expected to interfere with the development of the embryo. Most birth defects originate before conception and are due to abnormalities of the genetic material, that is, chromosomes and genes. There are quite a number of

individual malformations which are, for all practical purposes, entirely determined genetically. Genetic abnormalities can be inherited, in which case they are found in families. In some cases occur as an isolated event in a particular pregnancy. Perinatal health report of Canada (2010) share similar assertion when it documented that some genetic problems are new occurrences where the baby is the first to have the condition but he/she may pass it on to his/her children.

According to Sipek (2008) abnormality in the gene include chromosomal abnormalities, single gene defects and conditions known as multifactorial disorders, which are caused by the interaction of genes and the environment. He further reported that genetic factors are the most common causes of congenital malformations and account for approximately one-fourth of all congenital malformations. He further stated that genetically abnormalities occur as a result of Monogenic inheritance, Polygenic inheritance, chromosomal aberrations and others (epigenetics etc.)

Monogenic Inheritance/Single Gene Defects

Some congenital malformations are inherited as a monogenic trait. There are several genes, whose mutations are associated with selected congenital anomalies. The reference can be found in Mendelian Inheritance in Man (MIM) like Marfan syndrome, Ehlers – Dahnlos syndrome, Osteogenesis imperfect, Achondroplasia, Holoprosencephaly, Xerodermapigmentosu, etc. These disorders are the result of a single mutant gene and follow the Mendelian rules, either as autosomal dominant, autosomal recessive or X-linked traits. Many of the more

than 8,000 disorders identified are rare and others may not show morphological defects (Anderson, 1995).

Autosomal dominant gene defects give rise to recognizable effects in heterozygous individuals, usually with an equal sex distribution in about 50% of the offspring. Some of these disorders, such as Huntington disease and some of the autosomal dominant cerebellar ataxias, do not produce recognizable disease before adult life, whereas others, such as achondroplasia and atophoric dysplasia, are recognizable at birth and may be detected prenatally by ultrasound examination. When an autosomal disorder occurs with unaffected parents, a new mutation is not likely to recur in siblings. Gonadal mosaicism, reduced penetrance and variable expression may represent a small but real recurrence rate. Small deletions, responsible for contiguous gene syndromes, may segregate as dominant mutations. For example, velocardiofacial syndrome (VCFS) is due to deletion of 22q11, but with sufficient extensive deletion a more severe condition arises, including DiGeorge sequence (Anderson, 1995).

Recently, good evidence has come from America (Lowe, May & Reed, 1949 as cited in Ekwere et al., 2011) and independently from The Hospital for Sick Children, London, that the basic disturbance in fetus development or formation is due to a recessive genetic factor. Autosomal recessive gene defects occur equally in males and females, and are only clinically manifest in homozygotes with a recurrence risk of 25%. Therefore, affected individuals have healthy, heterozygous parents. Unless an autosomal recessive disorder is common in a certain population, such as Tay–Sachs disease in Ashkenazi Jews, there is

often a history of consanguineous marriage. An example of a recessive inherited disorder, affecting the CNS, is Meckel–Gruber syndrome (Ahdab-Barmada & Claassen 1990; Ekwere et al., 2011).

The X-linked recessive gene defects usually affect only males in 50% of cases if the mother is a carrier. The disorder is usually transmitted by healthy female carriers and their daughters have a similar chance of carrying the gene. Since the father, in general, does not pass an X chromosome to his sons, he will never pass the X-linked recessive trait to his male off spring. Examples are Duchenne muscular dystrophy and haemophilia (Gardnerand Sutherland 1996; Hamel 1999). Another study by Morch in 1941 also described the Achondroplasia (a type of dwarfism), as one which has shown near certainty due to a singledominant genetic factor. Again, many of the minor deformities of the skeleton such as the condition in which the fingers are short, only two phalanges instead of three, or that in which there are only two digits on the hand, giving the so-called lobster hand, are due to single dominant genes. (Gardnerand Sutherland 1996)

Some specific anomalies are also known to have a genetic origin where particular gene mutations or deletions have been identified. For example, most cases of Apert's syndrome are due to a spontaneous mutation affected one of two genes. The cause of the mutations is not known but when one or other mutation is present Apert's syndrome results. Apert's syndrome has an autosomal dominant inheritance, so that someone with Apert's syndrome has a 1 in 2 chance of passing the condition on to their children (Moore 2003). It is perhaps fair to say

that not many more diseases due to single genes are likely to be discovered unless they are very rare or unless a not uncommon disease is newly recognized and distinguished from other diseases causing similar signs and symptoms (Sipek, 2008).

Chromosomal Abnormalities

Chromosomal abnormalities are due to changes in the number or the structure of chromosomes from the normal state that result in a gain or loss of genetic material. Such abnormalities account for approximately 6 percent of birth defects in industrialized countries (Turnpenny & Ellard, 2005). Chromosomal abnormalities including numerical and structural abnormalities are a common cause of congenital malformations. Specific genetic syndromes are associated with the most common of these chromosomal defects. The most common live born example is Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13) (Kalousek et al., 1990). Down syndrome is generally caused by an extra chromosome 21 (trisomy 21), is the most common chromosomal abnormality in man.

Chromosome malformations are due to either excess or deficiency of chromosomal material including unbalanced rearrangements. Approximately 1 in 200 live newborns will have a chromosome abnormality. Excess or deficiency of chromosomal material can arise through a change in either chromosome 'Number or Structure'. Changes in chromosome number are of two types: Polyploidy and Aneuploidy, the loss or gain of a whole chromosome -monosomy and trisomy, respectively (Kalousek et al., 1990).

The Structural chromosome abnormalities may involve translocations (exchange of material between chromosomes), inversions, deletions or duplications (Gardner and Sutherland 1996). They may arise as a result of a parental chromosome rearrangement. Fusion at or near the centromere of the five acrocentric chromosomes, known as Robertsonian translocation, is one of the most common balanced structural rearrangements.

Environmental Factor

From theoretical considerations there are a number of groups of environmental factors which might be expected to interfere with the development of the embryo. Teratogenic factors have an adverse and disruptive effect on an embryo or a fetus between fertilization and birth. The term teratogen is usually limited to environmental agents, such as drugs, radiation and viruses. Those factors are commonly known as teratogens. The effect of teratogens is dependent on the genetics, that is, the genotype can modify the teratogenic effect. The disruptive effects include congenital abnormalities, embryonic and fetal death, intrauterine growth retardation (IUGR) and mental dysfunction. Sipek, (2008) classified teratogens into three main groups of Physical (x-rays, ionizing radiation, high temperature and mechanical factors), Chemical (exposure to alcohol, smoking, hard drugs, agrico-industrial industrial chemicals and cytostatic) and Biological (infectious agents, disease of the mother and other maternal conditions)

Physical Exposure to Radiations and High Temperature

Radiation effects on the developing brain were extensively studied after the atomic bombings of Hiroshima and NagasakiSchull et al., 1992; Arztebl, 2006). The most conspicuous effect on brain development is an increased occurrence of severe mental retardation, with or without microcephaly at specific gestational ages. The period between 8 and 15 weeks following fertilization appeared to be the most vulnerable. Schull et al. (1992) studied brain abnormalities in five of these mentally retarded individuals, using MRI. In the two patients exposed at the eighth or ninth week following fertilization, large areas of ectopic grey matter were seen, due to failure of neurons to migrate properly. The two individuals exposed in the 12th or 13th week showed no readily recognized ectopic grey areas but did show mild macrogyria, which implies some impairment in the development of the cortical zone. The one individual who was exposed in the 15th week did not show such changes. The brain was small with an apparently normal architecture.

Hyperthermia during pregnancy can cause embryonic death, abortion, growth retardation and developmental defects (Edwards et al. 1995, 2003). Cell proliferation, migration, differentiation and apoptosis are all adversely affected by elevated maternal temperature, showing some similarity to the effects of ionizing radiation. The development of the CNS is especially vulnerable: a 2.5 °C elevation for 1 h during early neural tube closure in rats resulted in an increased incidence of craniofacial defects, whereas 2–2.5 °C elevation for 1 h during early neurogenesis in guinea pigs caused an increase in the incidence of microcephaly

(Edwards et al. 1995). In general, thresholds and dose–response relationships vary between species. In humans, epidemiological studies suggest that an elevation of maternal body temperature by 2 °C for at least 24 h during fever can cause a range of developmental defects, however but there is little information on the threshold for shorter exposures (Chambers et al. 1998; Edwards et al. 2003).

Chemicals and Drugs

Drugs with a known teratogenic effect are relatively few (Shepard, 1998). Some drugs and known to cause congenital abnormality include tabacco, Aminopterin, Phentyoin, Valproic acid, Trimethadione, Lithium, Amphetamines, Tetracycline, organic mercury, lead and lithium. Retinoic acid syndrome malformations for instance appeared as a result of the use of Accutane (13-*cis*-retinoicacid) resulting in a surprising high incidence of severe craniofacial malformations (Gorlin et al. 2001).

Lack of Folic acid supplementation or using foods fortified with Folic acid during periconception period is associated with occurrence of congenital anomalies. Folic acid is known to be necessary for growth and function of human cells as it is crucial for biosynthesis and methylation of deoxyribonucleic acid (DNA) and ribonucleic acid (RNA) (Wisniewska & Wysocki, 2008). This is important for cell division, differentiation and regulation of gene expression especially when there is rapid cell division like during embryogenesis (Wisniewska & Wysocki, 2008). Folic acid is crucial for normal brain and spinal cord development during the first 4 weeks of gestation (Perinatal Health report-Canada, 2002). Preconception use of folic acid has been proved to cause

significant reduction of the risk for neural tube defects and other congenital anomalies like orofacial clefts, congenital heart diseases, urinary tract, limb and digestive system anomalies (Godwin, Sibbald, Bedard, Kuzeljevic, Lowry & Arbour, 2008).

Maternal chronic or excessive alcohol consumption, in particular during the first trimester of pregnancy, may lead to the fetal alcohol syndrome. Fetal alcohol spectrum disorder (FASD) encompasses a range of effects that can occur in an individual whose mother drank alcohol during pregnancy. These effects, which can range from mild to severe, include physical, behavioral, mental and learning disabilities with lifelong implications for affected individuals. The only cause of FASD is alcohol use during pregnancy and no threshold for safety is known. Major conditions in Fetal alcohol spectrum disorder (FASD) are Fetal alcohol syndrome (FAS), Partial FAS, Alcohol-related neuro developmental disorder (ARND), and Alcohol-related birth defects (ARBD) (Hoyme et al., 2005). Similar observation was also made by Clarren, Gilbert-Barness& Allen (1997).

Maternal Conditions

A variety of maternal diseases, either genetic or acquired, and deficiency states may affect the developing embryo. In other disorders, such as epilepsy, the therapy is most likely damaging. According to Scriver and Kaufman (2001) Maternal Phenylketonuria (PKU) is the best documented example of a genetic disorder in the mother affecting her offspring when her serum phenylalanine level is elevated during pregnancy. Without a strict diet throughout pregnancy, the

children of women with PKU have severe mental retardation, microcephaly and heart defects. Maternal diabetes mellitus type 1 is also identified as a risk factor for all sorts of congenital anomalies. Good control can prevent birth defects. However, a high incidence of Down syndrome and caudal regression syndrome have been noted (Godwin et al., 2008).

Pregnancy induced hypertension, vaginal bleeding early in pregnancy, twin pregnancy, oligohydramnios, polyhydramnios, breech presentation, period of gestation, antenatal care during pregnancy, history of previous abortions and still births have been observed to be maternal factors associated with congenital anomalies (Shawky & Sadik, 2011). Some maternal health conditions which have shown increased risks for congenital anomalies including obesity, insulin-dependent diabetes, various forms of folate deficiencies and phenylketonuria (Ramagopalan et al., 2010).

A number of infectious agents can affect the fetus, producing a range of effects from structural anomalies to mental retardation (Shawky & Sadik, 2011). Classically, the TORCH group of infections (toxoplasmosis, rubellavirus, cytomegalovirus and herpes/varicella virus) are screened for in the case of permanent cerebral impairment in the neonate (Sunderland, 1993; Ramagopalan et al., 2010). Also infections with human immunodeficiency virus (HIV) and other agents may lead to permanent fetal injury. Microcephaly, hydrocephalus, hydranencephaly and cerebral calcifications are most often found in the TORCH group of infections, and lead to developmental delay, psychomotor retardation and seizures. Microphthalmia is also often noted in toxoplasmosis, rubella and

HIV infection (Tominaga et al., 1996). Rubella virus is embryopathic but also has a recognizable fetopathic effect. Its features are cardiac defects, congenital cataract and deafness. Intracerebral calcifications, visible on ultrasound and CT examination, should raise suspicion for an intrauterine infection (Barth 2003; Ramagopalan et al., 2010).

Suspicion of a congenital malformation may arise on clinical grounds or because of an abnormal result from a routine prenatal investigation. Routine medical examination has been found to early detect some abnormalities. For instance high-frequency ultrasonography allows visualization of the normal and abnormal development of the embryo or fetus. Again Garel (2004) observed that second trimester serum test (triple test), first trimester serum test (double test) combined with nuchal translucency measurement on ultrasound examination, and the standard anomaly scan at 18–20 weeks of gestation was good to detect any neural tube defects. However, detailed knowledge about early development of the embryo and fetus is a prerequisite for evaluation of the pregnancy at risk for genetic diseases of the fetus, or when abnormal development of the embryo or fetus is suspected (Blaas et al. 1994; Amin et al. 1999; Blaas and Eik-Nes 1999).

According to Brock and Nicolaides (1992) a pregnancy may be at high risk of abnormality because of a particular family history or the advanced age of the mother. Higher-risk groups for chromosome abnormalities include older mothers, those with a previous chromosomally abnormal child, and when one parent is a translocation carrier. Associations between advanced paternal age and several congenital anomalies. Advance paternal age is known to be associated

with genetic changes in the sperm; this could lead to an increased risk for congenital anomalies in offspring.

Socio-Economic Factors

Socio-economic factors such as the level of education and employment status both mother and father has been identified to represent a selective risk factor for specific congenital malformations, confirming the role that social inequalities have effect on health, particularly reproductive health. Not all researchers have supported an automatic effect of teratogenic factors of drugs and chemicals on unborn children. Sipek (2008) noted that teratogenic effect is mother and dose-dependent. The same substance can be teratogenic only in a specific week of pregnancy. It can only affect the development of a specific organ / tissue. The effect is time-dependent and it is not easy to prove, that the congenital malformation was caused by the usage of a specific drug during pregnancy. During the time of blastogenesis, the damage caused by the teratogens cause no anomalies. The embryo is either able to repair all damage taken, or it stops to develop and dies (Sipek, 2008).

Barkovich and Linden (1994) also noted that in all instances the nature and the degree of congenital abnormality caused by infection is a function of the time of the infection. Early infections may lead to intrauterine death; lissencephaly may result from cytomegalovirus onset between 16 and 18 weeks of gestation, whereas polymicrogyria may be due to onset of infection between 18 and 24 weeks of gestation (Vries et al. 2004; Asante, 2015).

Measures to Minimize the maternal and perinatal risk factors of Congenital Abnormalities

The World Health Organisation in her sixty-third World Health Assembly in 2011 outlined a number of strategies to minimizing and treating congenital abnormality cases. Among the suggested strategies were screenings of newborn infants for congenital disorders to facilitate early detection, treatment and care. Neonatal screening programmes (physical examination of all neonates and screening for congenital hypothyroidism, phenylketonuria, sickle-cell disease and glucose-6-phosphate dehydrogenase deficiency) and training of primary health-care providers support the diagnosis and appropriate referral for treatment of infants with congenital disorders. According to WHO, physical examination of all newborn infants by trained primary health-care practitioners is feasible in most health systems and allows the identification of many birth defects, including cardiovascular defects that are associated with a high risk of early mortality and referral.

According to Queißer-Luft and Spranger (2006) perinatal and neonatal assessments used for quality control in gynecology and neonatology are surveillance systems for congenital defects. Such surveillance systems was investigated and captured about 50% of the major congenital malformations that are actually present for onward treatment (Lynberg & Edmonds 1992; Queisser-Luft, Stolz, Wiesel, Schlaefer & Spranger, 2002). The role of government in ensuring access to primary health care facilities with specialized trained practitioners in detecting congenital malformations in pregnant women have also

been identified. For instance Arztebl (2006) citing Lynberg and Edmonds (1992) observed that congenital malformations are a major public health issue and the quality of their treatment is an indicator of the quality of public health provision in general.

WHO (2012) documented that access to health and rehabilitation services is important to support the participation and inclusion of affected children. With appropriate training, primary health-care practitioners can recognize birth defects, diagnose common problems and identify associated disabilities, which in turn enables them to offer basic treatment and counseling, taking into account family and community circumstances and available medical services. Agrawal (2007) however noted that interventions for the control of genetic and congenital disorders at the national level, is not necessary but strategies and public health approaches can be incorporated into the existing reproductive health care system. He admitted that although some additional training and resources will be required, the potential benefit is considerable in terms of reduction of suffering as well as reduction of the health and economic burden related to the care of patients with genetic and congenital disorders.

Another strategy identified by researchers in the prevention, detection treatment of any form of congenital abnormality was the utilization of family planning services by couples intending to make families. According to Agrawal (2007) preconception information and services for family planning can help to reduce the number of high-risk pregnancies related to increased parental age. Agrawal in his conclusion noted that advice should be given to couples to

complete their intended family size preferably before the age of 35 years for women. Similar observation have been made by Modell et al., (1992) when he detected that chromosomal disorders and spontaneous abortion rises rapidly with maternal age after the age of 35 years.

Arztebl (2006) was a bit different in his submission in the measures in minimizing congenital abnormalities. He reported that access to health facilities, improvements in prenatal ultrasonography screening, induced abortions, intake of supplementary vitamins, etc. were not a suitable preventive measures but to reduce the number of congenital cases among number of live-born children but rather intensive reproductive public education campaigns were needed.

Empirical Literature Review

Maternal and Prenatal Risk Factors of Congenital Abnormalities

Rosano et al (2008) in their study compared parental socioeconomic status of 485 children affected by neural tube defect (NTD), orofacial clefts (OFC) and Down syndrome (DS). The socioeconomic measures included maternal and paternal level of education and employment. The study showed a higher risk for NTD and OFC for parents in low social class. As well, the estimated risk of DS was slightly higher, though not statistically significant. Clearly, it was seen among the socioeconomic variables taken into consideration, the mother's education level represent a significant risk factor associated with Orofacial cleft occurrence. The study further suggested that socioeconomic level may represent a selective risk factor for specific congenital malformation, confirming the role that social inequalities have on health, particularly reproductive health.

Vrijheid et al (2000) also investigated socioeconomic inequalities in the risk of congenital anomalies focusing on risk of specific anomaly subgroups. A total of 858 cases of congenital anomaly and 1764 non-malformed control births were collected between 1986 and 1993 from four UK congenital malformation registers. It was discovered that the risk of non-chromosomal anomalies increased with increasing socioeconomic deprivation. Some malformation subgroups showed increasing risk with increasing deprivation: all cardiac defects, malformation of the digestive system and multiple malformations. No evidence was found for socioeconomic variation for other non-chromosomal malformation groups, including neural tube defects and oral cleft. A decreasing risk with increasing deprivation found for all chromosomal malformation and Down's syndrome in unadjusted analyses, occurred mainly as a result of differences in the maternal age distribution between social classes. They study concluded on a note that the more deprived populations have a higher risk of congenital anomalies on non-chromosomal origin and some specific anomalies.

Dolk and Lancet (1998) study mothers who lived near hazardous waste landfill sites in Europe and found that there was increased risk of congenital anomaly in babies whose mothers love close to landfill sites that handle hazardous chemical wastes. In this study residence within 3km of landfill site was associated with a significantly raised risk of anomaly and there was a fairly consistent decrease in risk with distance away from the sites. The defects found were neural-tube, malformations of the cardiac septa and anomalies of great arteries and veins.

A study of 1151 cases of Down syndrome and 1572 cases of nine other severe anomalies revealed socioeconomic inequalities exist in the antenatal detection of Down Syndrome (DS) and subsequent termination rates are much higher for DS than the other anomalies. The overall antenatal detection was 57% for DS, which deceased with increasing deprivation. Termination rates for all anomalies are lower in more deprived areas leading to wide socioeconomic inequalities in live born infants with congenital anomaly, particularly DS and subsequent neonatal mortality (Budd et al., 2015).

Evidences of the presence of certain diseases during pregnancy have been documented as a risk factor in congenital abnormality. Janssen et al., (1996) studied the relationship between diabetes in pregnancy and development of congenital malformation in Washington from the year 1984 to 1991. A total of 1511 mothers with established and gestational diabetes were studied respectively. It was found that mothers with established diabetes were more likely to have congenital malformation than newborns of non-diabetic mothers. In contrast, there was only a slightly higher prevalence of congenital malformations among newborns of mothers with gestational diabetes. The association with maternal diabetes was greater for neonates with multiple malformations than for single malformations.

Four to seven fold association were observed with skeletal, cleft palate, neural tube and heart abnormalities. Similar observation was previously made by Chung et al., (1975) when they investigated 23,695 pregnancies from the prospective Collaborative Perinatal Project. From theoretical perspective

infections has been identified as a factor causing abnormalities in children congenitally. Zika Virus infection in pregnancy can cause congenital Brain abnormalities and Guillain-Barre Syndrome (GBS).

The study of the relationship between gross placental characteristics and prenatal outcome of low-risk singleton deliveries was done among 428 singleton deliveries in 2013 at the University of Ilorin, USA. It was concluded that there was an association between placenta parameters and foetal outcome at birth. Placental weight was positively correlated with birth weight, gestational age and occurrence of congenital abnormalities.

Another risk factor identified to be associated with congenital malformations in children born of mothers who got exposed to certain drugs has also been documented. Thalidomide embyropathy cases born between 1959 and 2010 in Brazil among 28 Brazilian individuals and reported progressive deafness and dental loss. Volkow, Compton and Wargo (2016) also reported of the risk associated with the use of marijuana during pregnancy. Toxic chemicals were cited by Talukder and Sharma (2006) as agents probably having serious effects on malformations, postnatal death, functional learning deficits, and premature aging.

There is also evidence that certain antiepileptic drugs (AEDs) were teratogenic and were associated with increased risk of congenital malformation. In an assessment of the effects of prenatal exposure to AEDs on the prevalence of congenital malformations in children, reported that children exposed to carbamazepine (CBZ), phenytoin (PHT), topiramate (TPM), and valproate (VPA) were at a higher risk of malformation than children born to women without

epilepsy. However, the study found no increased of malformation for lamotrigine (LTG), oxcarbazepine (OXC), primidone (PRM) or zonisamide (ZNS). Similar studies include that of Adab et al (2004).

The effects and human risks of environmental toxicants (drugs, chemical and physical agents) and reported an association between environmental toxicants and congenital abnormalities. Risk assessment of drug use in pregnancy. From the above empirical evidences, it was clear that several maternal and perinatal characteristics affect many variables leading to congenital abnormalities. Socioeconomic factor (level of education, employment), drugs, maternal diseases, exposure to certain environmental teratogens were few that has been captured in the review.

Conceptual Framework

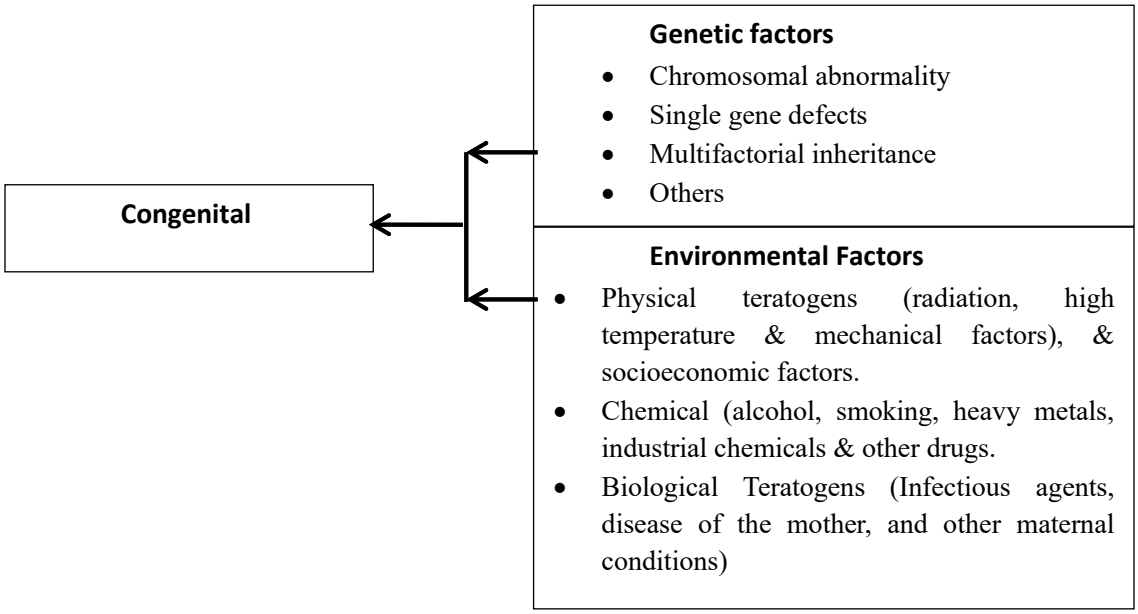

Figure 1: Conceptual framework of congenital abnormality

Source: Author's construct, (2017)

CHAPTER THREE

RESEARCH METHODS

Overview

This chapter provides detailed description of the methodology that was employed in the study. It include; the research design, population, sample and sampling procedures, research instrument, data collection procedure and the method of data analysis procedure.

Research Design

The study was an exploratory study which employed quantitative technique. Structured questionnaires were used to collect data from mothers on maternal and perinatal contributing factors of congenital abnormalities in children under five years at the Child Health Department of the Korle Bu Teaching. Data on the children was obtained from the children medical records.

Research Setting

The study site was Korle-Bu Teaching Hospital (KBTH). KBTH is a tertiary level referral hospital situated in the Western part of Accra, on the Guggisberg Avenue on the Harley Street. It is the oldest and largest hospital in Ghana. The hospital has 1800 bed capacity and clinical and diagnostic departments/units of which the Child Health Department (CHD) is one. The Child Health Department was used for the study because it is the most preferred child specialist hospital and most children with major congenital abnormalities are referred to this unit for specialized care from other hospitals in the country. The

mothers of these children would be easily assessed for the study at the child health unit.

Population

The population for the study included all mothers with children below 5 years old with congenital abnormalities, who were attending the Child Health Department between March, 2017 and April, 2017. From the Child Health Registry of the Korle Bu Teaching Hospital, there were 4292 cases of children below five years, who reported to the unit from January, 2017 and December, 2017. Out of the 4292 reported cases, 330 were associated with congenital abnormality. Hence, the population for the study was 330 mothers. These included mothers of children below 5 years of age diagnosed with congenital abnormality at the cardiac clinic, neurodevelopmental clinic, renal clinic, paediatric surgery clinic as well as NICU clinic and the ward.

Sample and Sampling Technique

The sample included mothers of children with congenital abnormalities who are below five years at the Child Health Department of the Korle Bu Teaching Hospital. Hence, the inclusion criteria for participants was mothers of children below 5 years old with congenital abnormality who are admitted or attending clinic at the Child Health Department. From the Child Health Registry, there were 330 cases of children diagnosed with congenital abnormalities between January, 2016 and December, 2016.

In this research, non-probability sampling method was employed in which the participants were selected using convenience sampling.

Sample Size Determination

The sample size was computed with the formula proposed by Yamane (1967 as cited in Annette & Meier, 2009). The formula is given as

$$n = \frac{N}{1 + N(e)^2}$$

Where n is the sample size; N is the study population size; and e is the level of precision desired. Thus, taking a 95% confidence level and a desired precision (e) of 5% with the population of 330, the sample size would be calculated as:

$$n = \frac{330}{1 + 330(0.05)^2}$$

$$n = \frac{330}{1 + 0.825}$$

$$n = 180.82$$

Hence, 181 mothers with children diagnosed as well as the children with congenital abnormalities constituted the sample for the study.

Inclusion and Exclusion Criteria

The inclusion criteria for participants were mothers of children with congenital abnormalities who are below five years. These included those admitted or attending clinic at the Child Health Department but were diagnosed to have congenital abnormality. All mothers of children who were referred or admitted and diagnosed with any other medical condition other than congenital abnormalities were excluded from the study.

Data Collection Instrument

After a careful review of appropriate literature, a structured questionnaire was designed to collect data about the participants and the information received on the children was confirmed from their children medical records. A structured questionnaire with three major sections was used to collect data from respondents (Appendix A).

Section A sought information on the types of congenital abnormalities seen at the child health department of the Korle Bu teaching hospital

Section B focused on maternal medical and perinatal contributing factors of congenital abnormalities in children below five years.

Section C sought information on the methods to minimize the maternal and perinatal contributing factors to congenital abnormalities

The main reason for using the questionnaire was because it is an efficient way of collecting information.

Data Collection Procedure

Ethical clearance was sought from University of Cape Coast Institutional Review Board. An introductory letter was collected from University of Cape Coast (Appendix D) to Korle Bu teaching hospital. Approval was sought from Korle Bu teaching hospital, child health department and the records unit of Korle Bu Teaching Hospital in order to have access to the patient's medical records. The researcher also liaised with the nurses on duty at the various clinics to help identify mothers with children with congenital abnormalities for the research. Detailed information about the research and its significance was given to the mothers before participation. Eligible participants were enrolled consequentially. Informed Consent was sought from mothers with children with congenital abnormalities by signing a voluntary participation sheet or by thumb printing. Mothers of these children were assisted to complete a questionnaire for the research whilst waiting to see the doctor. Participants were assured that the data gathered was used for academic purposes only. Parts of the questionnaire that mothers do not understand were explained to them in the language that they understand. Privacy was provided during data collection.

Data was collected on demographic characteristics of both mother and child, maternal medical, gynaecological, obstetric and perinatal history. A questionnaire constructed based on the objectives was used to help answer the research questions. The classification of malformations was based upon the International Classification of diseases, XVII classification of congenital abnormalities (Appendix B). Information on the children was obtained by asking

the mother and then confirmed by the child's medical record but information on the maternal medical, gyanaecological, obstetric and perinatal history would be obtained from the mother.

Data Processing and Analysis

The data collected from the registry and interview guide was first numbered serially, edited, coded and analyzed. Data observed critically was be summarized in tables and diagrams. A computer software known as Statistical Package for Social Sciences (SPSS) version 22 was used to analyse the data.

Univariate followed by multivariate logistic regression analyses was done to determine maternal factors associated with congenital anomalies. Factors with a p-value of less than 0.05 on univariate analysis were subjected to multivariate logistic regression analysis. Crude (unadjusted) and adjusted odds ratios will be calculated to quantify the strength of association between congenital anomalies and associated factors.

For quantitative data, after checking the completeness, missing values, and coding of questionnaires, data was entered into the computer, processed and analyzed using the version 22 of the SPSS software. The data was summarized and described using descriptive statistics (frequencies, means, standard deviations etc.). The descriptive statistics provide simple summaries about the sample, the measures and then form the basis of quantitative analysis of data since the main instrument used will be questionnaire.

Chi square statistical analysis was used to test objective one and two where the researcher sorted the types and characteristics of the congenital

abnormalities. Objective three was examined by using multiple regression analysis to determine how the various maternal factors lead to congenital abnormalities.

Data Management

The data collected was held confidential and only accessible to the researcher. The confidential data was destroyed after the study.

Pre-Testing

Pre-testing of the data collection instrument refers to testing the instrument prior to the actual collection of data (Polit & Beck 2008). A pre-test of the checklist was done among 10 children with congenital abnormalities at the Princess Marie Children Hospital in Accra because it had similar features as the child health department of the Korle-bu Teaching Hospital. The pre-test was done in order to assess the appropriateness and suitability of the questions and to find out whether the respondents understand the questions as intended, as well as their willingness to answer the questions.

Also the data was analyzed by using Statistical Package for Social Sciences (SPSS) and frequencies and cross tabulation was run on them.

Ethical Consideration

Ethical clearance was sought from the instructional review board of the University of Cape Coast (Appendix C). An introductory letter was collected from the school and presented to the Korle Bu Hospital for approval for the research. Ethical clearance was sought from the Korle Bu Teaching Hospital ethical review

board. All information retrieved from the patient's health records were kept confidential. For ethical reasons researcher used the following principles, confidentially, voluntary participation and anonymity in the conduct of the research.

In terms of confidentiality the respondents were assured that the information will not be disclosed to anyone who is not directly involved in the study. For voluntary participation the respondents were fully informed and volunteered to participate in the study after explaining the purpose of the study – for academic purpose only. For anonymity, privacy and confidentiality was ensured during the interview. The respondents were assured that they will remain anonymous throughout the study that is their names/address will not be written on the questionnaire also. Informed consent was sought from mothers of children with congenital abnormalities by signing or thumb printing an informed consent form before participation. They were informed that they can decline from the research at any point without any influence on the care of the child.

CHAPTER FOUR

RESULTS AND DISCUSSIONS

Introduction

This chapter presents the results and discussion of data obtained from the study. The presentation of the results conforms to the order of arrangement in which the objectives of the study are arranged. However the first part presents the demographic characteristics of respondents and the rest of the results and discussion are presented under themes in line with the objectives of the study.

Table 1: Demographic characteristics of respondents

Variables		Frequency	Percentage
Age of the child	< 1year	88	48.6
	1-3years	75	41.4
	>3-5years	18	9.9
Sex of the child	Male	143	79.0
	Female	38	21.0
Maternal age	<20 years	10	5.5
	21-40 years	164	90.0
	41-50 years	7	3.9
Educational level	No education	12	6.6
	Basic education	49	27.1
	Secondary education	85	47.0
	Tertiary	29	16.0
	Other	6	3.3
Religious denomination	Traditional	6	3.3
	Moslem	75	41.4
	Christian	100	55.2

Source: Field Survey (2017)

In table 1 the demographic characteristics of the respondents were presented and it was revealed in the age of the children that most (48.6%) of the children were less than 1 year and 41.4% were between 1-3 years. Moreover 18 representing 9.9% were over 3 years but less than the 5 years. Again it was realised that the majority of the children (79.0%) were males. A look at the maternal age also revealed that about 90.0% of the mothers were between age 21-40 years, with 3.9% and 5.5% for mothers between 41-50 years and less than 20 years respectively. For the educational attainment of the mothers, it was observed that most of them had secondary education and 27.1% had basic education. It was also noticed that 29 of the mothers representing 16.0% have had tertiary education and 3.3% of the mother had other certificates. In terms of the religious background of the mothers, more than half of the mothers (55.2%) were Christians and 41.4% were Moslems and just 3.3% of the mothers too were Traditionalist.

Types of Congenital Malformation Seen at the Child Health Department, Korle Bu Teaching Hospital

Table 2: Types of malformations

Malformations	Frequency	Percentage
Anorectal	28	15.5
Atresia	6	3.3
Bladder Extrophy	3	1.7
Cerebral palsy	4	2.2
Cleft Lip & Palate	9	5.0
Coiled penis	2	1.1
Down Syndrome	8	4.4
Duodenal Atresia	6	3.3

Table 2, continued

Ectopic Kidney	2	1.1
Encephalocele	4	2.2
Equinovarus	8	4.4
Exomphalus	2	1.1
Fistula	1	.6
Heart disease	56	31.0
Hydrocephalus	10	5.5
Hydronephrosis	2	1.1
Hypospadias	8	4.4
Jejunal Atresia	2	1.1
No vaginal opening	6	3.3
Omphalocele	2	1.1
Posterior Uretral valve	2	1.1
Sacro-Coccygeal Tumour	1	.6
Sepsis	1	.6
Spina Bifida	2	1.1
Undescended testes	2	1.1
Ventricular Septal Defect	4	2.2
Total	181	100.0

Source: Field Survey (2017)

Results on Table 2 presented the various types of congenital malformations that the hospital recorded over the study period. It was observed that most (31.0%) of the children diagnosed of congenital malformation had heart diseases. Also in the table is Anorectal malformations, about 16% of the children over the period were diagnosed of this kind of malformation. There were other malformations that were seen or diagnosed on the cohort of patients and they included the Cleft lip & palate (5.0%), Hydrocephalus (5.5%), Down syndrome (4.4%), and Equinoxarus (4.4%). The study also revealed that there were minor

cases of malformations such as No vaginal opening (3.3%), Undescended testes (1.1%), Coiled penis (1.1%), Cerebral palsy (2.2%), Bladder Extrophy (1.7%), Duodenal Atresia (3.3%), and Ventricular Septal Defect (2.2%).

Table 3: System of the child affected

System affected	Frequency	Percent
Cardiovascular	59	33.7
Digestive	44	24.3
Gastrointestinal	4	2.2
Musculoskeletal	8	4.4
Nervous system	37	20.5
Reproductive	18	9.9
Urinary	9	5.0
Total	181	100.0

Source: Field Survey (2017)

Table 3 presented the systems or areas most affected when these malformations were diagnosed. From the table 33.7% of the systems affected by these malformations are cardiovascular or the circulatory system. The digestive system of patient with congenital malformation formed 24% of the systems affected by children recorded at the hospital over the study period. It was also revealed that there were other parts or systems such as the nervous system (20.5%), Musculoskeletal (4.4%), Gastrointestinal (2.2%), Reproductive (9.9%) and Urinary (5.0%) that are affected by congenital abnormalities. Again,

Table 4: Presence of congenital abnormality and time of diagnoses

Statements		Frequency	Percentage
Does the child have any congenital abnormality	Yes	181	100
	No	0	0
	Total	181	100.0
When was it diagnosed	Before delivery	2	1.1
	At birth	73	40.3
	On admission	106	58.6
	Total	181	100.0

Source: Field Survey (2017)

Table 4 finds out the presence of congenital abnormality and when that anomaly was diagnosed. From the table, it is noticed that all the children had congenital abnormality. Again in the same table it was realized that majority (58.6%) of these abnormality were diagnosed on admission at the hospital. Moreover, 73 representing 40.3% of the children were diagnosed with congenital abnormality at birth and 1.1% of the children with congenital abnormality were also diagnosed before delivery. From the forgoing it could be inferred that given the facilities at the hospital over the period of the study, congenital abnormalities were only diagnosed either at birth or on admission. It may not be out of place to assume that when anomalies are diagnosed on time their effects are mostly reduced or minimized.

Table 5: Cross tabulation of number of children that died during the period of data collection and the time of diagnosis

	Number of children dead or alive		Total
When congenital abnormality was diagnosed	Alive F (%)	Dead F (%)	F (%)
Before delivery	5(2.8)	0(0.0)	5(2.8)
At birth	97(53.6)	10(5.5)	107(59.1)
On admission	53(28.3)	16(8.8)	69(37.1)
Total	155(85.7)	26(14.3)	100.0

$\chi 2$ (5, n = 26) = 37.60, p =0 .001, *phi* =0.564

Source: Field Survey (2017)

Table 5 assessed the relationship that existed between time of diagnosis and the number of children dead. The relationship was tested by the use of the chi-square test of association or goodness of fit. The results showed that there was significant relationship between the time of delivery and the number of children that died during the period of data collection (p=0.001). Again on the table, it was realized that all 5(2.8%) children diagnosed of congenital abnormality before delivery survived or are alive and none (0.0%) was dead. Moreover, comparing children diagnosed at birth and on admission at the hospital over the study period, relatively more of the children diagnosed on admission died (8.8%) relative to 5.5% children diagnosed with malformation at birth. On the other, in terms of the children alive, there were more children diagnosed at birth (53.6%) than children diagnosed on admission (28.3%). In addition to the above, the results showed that

on a whole, children with congenital abnormalities have about 56.4% survival if they were diagnosed on time compared to malformations that are seen or diagnosed late.

Table 6: Cross tabulation of mode of delivery and presence of congenital abnormality

Does the child have any congenital abnormality?		**Mode of delivery**		**Total**
		Spontaneous virginal delivery	Caesarean Section	
Yes	100	129 (71.3%)	47(26.0%)	176(97.2%)
No	0	2(1.1%)	3(1.7%)	5(2.8%)
Total	100%	131(72.4%)	50(27.6%)	181(100.0%)

$\chi 2$ (1, n = 181) = 1.28, p =0 .256, *phi* = 0.122

Results of table 6 also examined whether there was a relationship between the mode of delivering of a child and cause of congenital malformation. According to the chi-square test of goodness of fit (with p =0 .256), there was no relationship or fit between mode of delivery and the presence of congenital abnormality. Even though there were no association between the presence of malformation and the mode of delivery, Table 4 showed that majority (71.3%) of children born by spontaneous virginal delivery had congenital abnormality while 26.0% of the children with congenital abnormality were by way of a caesarean section.

Table 7: Cross tabulation of gestational age and presence of congenital abnormality

Gestational age	Does the child have any congenital abnormality?		Total
	Yes	No	
< 38weeks	5(2.8%)	0(0.0%)	5(2.8%)
39-41weeks	63(34.8%)	5(2.8%)	68(37.6%)
42 weeks and above	108 (59.7%)	0(0.0%)	108 (59.7%)
Total	176(97.2%)	5(2.8%)	181(100.0%)

$\chi 2$ (2, n = 181) = 8.545, p = 0.001, *phi* = 0.117

Source: Field Survey (2017)

Results on table 7 examine the association that existed between gestational age and the presence congenital abnormality. It was revealed that, there was a significant relationship between gestational age of a baby and the presence of congenital malformation in children under five years ($p<0.001$). The overall effect shows that babies born late or beyond their normal gestation period have more chances (11.7%) of having congenital abnormality compared to children born earlier or at their normal gestation periods. Tables 7 again showed that majority of the children (59.7%) with congenital malformation were born at age 42 weeks old and above. It was also observed that 63 representing 34.8% of the children born with congenital malformation were born at age between 39-41weeks. Only 2.8% of the children with congenital malformations were born at age below 38 weeks.

Maternal and perinatal risk factors of congenital abnormalities in children below five years

Table 8: Regression result of how maternal and perinatal characteristics have contributed to congenital abnormalities in children below five years

	Unstandardized Coefficients		Standardized Coefficients	t	Sig
	B	Std. Error	Beta		
Constant	2.504	0.230		10.873	0.001
Were you regular attendant at antenatal during pregnancy?	0.129	0.034	0.251	3.825	0.001
Any gynecological condition	0.149	0.052	0.209	2.854	0.005
Taking all medications given at the antenatal clinic	0.066	0.035	0.147	1.893	0.001
Folic acid intake	0.083	0.034	0.184	2.459	0.001
Oligohydramnios or polyhydramnios	-0.001	0.025	-0.003	-0.037	0.971
Breech presentation	0.018	0.020	0.067	0.900	0.370
History of previous abortions and still births	-0.037	0.027	-0.116	-1.379	0.170
Does maternal occupation exposes her to any radiation or chemicals	0.015	0.023	0.053	0.676	0.500
Alcoholism	-0.022	0.043	-0.039	-0.508	0.612
Herbal medication	-0.010	0.002	-0.037	-5.000	0.001
Illicit drug	-1.007	0.099	-0.585	-10.15	0.001

Dependent Variable: Does the child have any congenital abnormality?

$F(10, 181) = 13.556$, $p = 0.001$

Results in Table 8 examined how maternal and perinatal characteristics have contributed to congenital abnormalities in children less than five years. From

the table, variables such as regular attendant at antenatal during pregnancy, gynecological conditions, taking of medications given, Folic acid intake, intake of some herbal medication during pregnancy and illicit drug intake were all significant in influencing the presence of congenital malformation in a child or a baby. While intake of some herbal medication during pregnancy and illicit drug intake had negative effect on the child, the rest of the variables positively affected the child's chances of getting congenital malformations. The ANOVA statistics beneath table 8 show that the entire model or regression was good for the discussion. According to Pallant (2011) and Tabachnick and Fidell (2007) if the F-statistic in the ANOVA is $p > 0.05$ then the entire model falls short or is not good.

The results revealed that there is an association between regular attendance to antenatal and the presence of congenital abnormality (p=0.001). Moreover on the table it was seen that, regular attendant at antenatal during pregnancy improves the lives of children. That is, the regular attendance at the antenatal during pregnancy, the less likely is the child to get congenital abnormally. For every extra day of hospital attendance, the odds of the child getting any congenital malformation decreases by 0.129 all other things being equal. Again on the table, gynecological conditions of the mother have a positive effect on the child getting any congenital malformation, such that the more the gynecological conditions of the mother, the higher the chances of her baby to report of any congenital malformations. The results also indicate that a mother

with gynecological conditions have 0.149 chances of reporting of any congenital disorder compared to mothers without any gynecological conditions.

The taking of all medications given to a mother at the antenatal was found to be significant (p=001) and positively affected the chances of reporting any congenital anomaly. The result showed that the odds that a mother who takes all her medication to report of any anomaly is 0.066 less than a mother who does not take all her medication given at the antenatal clinic all other thing remaining constant. Moreover, the intake of folic acid was also seen to have a significant and positive effect on the presence of congenital anomaly. Any extra folic acid intake by a mother reduces the chance of reporting any malformation to the baby by 0.083 compared to those who skips or ignores folic acid intake.

The study revealed a significant association between application of herbal medication and intake of illicit drugs and the presence of congenital abnormalities in children below five years (p=001).The study again found the application of herbal medications and the use of illicit drugs during pregnancy to have a negative impact on the child, such that the higher the intake or application of those drugs, the lower the health of their children. Again on table 8, for instance, the chances of reporting congenital abnormality increases by 0.010 if the mother uses herbal medication compared to not applying herbal medications. On the other hand, if a mother takes in illicit drug during pregnancy, the odds of reporting a child with any congenital anomaly increases by 1.007.

Minimization of the Maternal and Perinatal Risk Factors of Congenital Abnormalities

Table 9: Suggestions to minimize the maternal and perinatal risk factors of congenital abnormalities in children

Strategies		Frequency	Percentage
Education to start reproduction	Yes	144	63.0
before 35yrs	No	67	37.0
Rubella tetanus and other	Yes	165	91.2
immunization	No	16	8.8
Periconceptional supplement of	Yes	94	51.9
folic acid	No	87	48.1
Nutritional counseling	Yes	177	97.8
	No	4	2.2
Control of maternal infection	Yes	113	62.4
after diagnosis	No	68	37.6
Education on the avoidance of	Yes	153	84.5
teratogens, chemicals and	No	28	15.5
radiation			
Provision of genetic services	Yes	12	6.6
for couples with history of	No	169	93.4
congenital anomaly			
New born examination to	Yes	72	39.8
screen for congenital	No	109	60.2
anomalies			

Source: Field Survey, Owusu-Takyi (2017)

Results on table 9 gives the possible ways to minimize the congenital abnormalities in children less than five years. From the table, over two thirds (84.5%) of the entire respondents maintained that education on the avoidance of teratogens, chemicals and radiation by pregnant mothers may be a way to

addressing the issue of congenital anomaly. Again, 144 respondents representing over 50% of the entire respondents (63.0%) also mentioned that couples should be educated to start reproduction before 35 years. Though it may not be presents in this study, studies elsewhere have proven that given birth after certain age may not be helpful since most of the systems in the body may be weak. About 98% of the respondents also emphasized that giving of nutritional counseling to mothers and pregnant women is a sure way to reducing some congenital malformations. This view is also wholly true because it was established by this study that children born to malnourished mothers or woman were more likely to lack essential nutrients for their development.

There were other suggestions by 91.2% of the respondents that mothers should be immunized against Rubella teteanus and other teratogenic chemicals. Others also maintained that notwithstanding, the above mentioned strategies, there must be periconceptional supplement of folic acid. Additionally, 62.4% of the respondents stated that, there must be control of maternal infections after diagnosis. This is because lack of folic acid, infections and exposure to teretogenes and other environmental factors (radiations, high temperatures, smoke, and agrico-industrial industrial chemicals) may make the fetus susceptible to other congenital malformations. Especially, studies by Arztebl (2006); Sipek (2008) made credence to the fact that excess exposure to ionizing radiations and other chemicals may have negative effect on unborn children.

Discussion of the Results

This section gives a detailed discussion of the results of the study. It moreover tries to relate the findings of the study to previous studies provided in the literature in order to justify the different results of the different models and provide interpretation of the results. Table 1 gives the demographic characteristics of respondents in the study. It was observed that majority of the children with malformations were males and close to half of the entire children (48.6%) were below 1 year. For the mothers, 90% of them were aged between 21-40 years while a significant 55% were aged less than 20 years. Again, while most of the mothers were Christians (55.2%), 47% had Secondary school education. The demographic characteristics of the study were provided because the belief of the study is that the demographic description of respondent provides a basis for differentiating between responses, since aggregated responses may exclude some pertinent isolated concerns. This view coincided with the studies by Winterbottom (2001); Gill, Broussard, Devine, Green, Rasmussen, and Reefhuis (2012) who mentioned that some birth defects or congenital abnormality may be influenced by the demographic characteristics of the mothers. For instance, religious faith or affiliation and level of education may to a greater extent affect the aggravation of defects in children.

For instance, some birth defects come as a results of deficiency in other supplements e.g. Vitamin A and Folic acid. In Ghana where level of education is relatively low, mothers are oblivious on how to care for themselves during pregnancy (eg where to go for treatment, what to eat, etc.) may be victims.

According to Gill et al (2012), mothers over 40 years are positively associated with several cardiac defects including perimembranous, ventricular septal defect, and atrial septal defect. Again, there were also other cardiac defects observed to be associated with maternal age over 40 years. Example esophageal atresia, hypospadias and craniosynostosis. Gill et al (2012) continued to state that mothers less 20 years were associated with few birth defects including lens defects, perimembranous ventricular septal defect, left ventricular outflow tract obstruction, coarctation of the aorta, hypospadias and craniosynostosis.

Types of Congenital Malformation

Here the study clearly looked at the types of malformation that were present in the children or were recorded at the hospital. From Table it was seen that Heart disease (31%) and Anorectal (15.5%) were the most suffered anomalies by children even though there were other pockets of case of other malformations such as Hydrocephalus, Cleft lip & Palate, and Down syndrome. The results conform to the study of WHO (2016) that the most common and severe congenital anomalies are heart defects or diseases, neural tube defects, Down syndrome and Equinoxarus. The study went further to assert that defects such as the Down syndrome and Equinoxarus are very particular in less developed or developing countries where mothers grapple with issues like lack of nutrition, infections, lack of folic acid and iodine intake and other environmental conditions. For instance, if a pregnant woman is malnourished, then the fetus is likely to lack the nutrients essential for its development (Greener, 2008).

Experts also revealed that Down syndrome occurs when an individual has a full or partial extra copy of chromosome 21 and this alters the course of development. Down's syndrome causes a distinct facial appearance, intellectual disability and developmental delays. The Equinoxarus on the other hand is a congenital deformity involving one foot or both and the affected foot appears to have been rotated internally at the ankle (WHO, 2000). Singh and Gupta (2009) and Schumacher et al (1987) also categorized abnormal development into four basic types: reduced or absent body parts (insufficient generative energy), enlarged or multiple body parts (excessive energy), aberration of form and of position, and hermaphroditism. This is evident of the fact that study mentioned Hydrocephalus and Encephalocele as some of the malformations recorded by the hospital over the study period. For instance the Encephalocele is a neural tube defect characterized by sac-like protrusions of the brain and the membranes that cover it through openings in the skull. The Hydrocephalus on the other hand is a condition in which there is an abnormal accumulation of cerebrospinal fluid (CSF) within the brain and this causes increased pressure and makes the brain to swell.

There was also the mention of Anorectal anomalies and cleft and palate which were mostly caused by deficiencies and the intake of teratogen. Studies revealed that they are also rampant globally however earlier detection and surgery may correct the system (WHO, 2000). However, in less developed countries, the lack of money for treatment makes the prevalence to increase. The same may be said for Anorectal anomalies which are medical problems affecting the structure

of the anus and rectum. Folic acid is crucial for normal brain and spinal cord development during the first 4 weeks of gestation (Perinatal Health report-Canada, 2002). Preconception use of folic acid has been proved to cause significant reduction of the risk for neural tube defects and other congenital anomalies like orofacial clefts, congenital heart diseases, urinary tract, limb and digestive system anomalies (Godwin, Sibbald, Bedard, Kuzeljevic, Lowry & Arbour, 2008).

Table 3 also outlined the systems in victims that were most affected by the malformations. The study showed that the Cardiovascular (33.7%), Digestive (24.3%) and Nervous system (20.5%) were the most affected systems in victims. However, the least affected were Gastrointestinal, Musculoskeletal, and Urinary. These revelations were in line with the normal systems affected when congenital malformation occurs. The results support the study by WHO (2012) that explained that, most affected parts of the people diagnosed with congenital anomalies were the nervous system and the cardiovascular systems. This view was buttressed with the point that in less developed and some developed countries, congenital malformations have appeared as the major childhood health problem, affecting approximately 1 in every 33 infants and are the fourth most common cause of neonatal deaths in especially Africa. The results however contradicts the order in the study of Arnold and Christopher (2006). For instance, Arnold and Christopher (2006) mentioned that the most common body systems involved in congenital anomalies included musculoskeletal, central nervous

system, gastro intestinal system and cardiovascular system with the least affected system being the urogenital system.

Again, the study revealed that most (58.6%) of the victims were diagnosed of the particular anomaly on admission. It is only fair to assume that when anomalies are diagnosed on time their effects are mostly reduced or minimized. Pahomov and Porter (2012) and Tantibanchachai (2014) made a strong case for this. For instance, Pahomov and Porter (2012) made the case in support of the idea or theory of arrested development as a cause to congenital abnormality that, when deformities were seen on time they could be averted or corrected. This idea also sync with the old cliché that "the earlier the better". Tantibanchachai (2014) also supported the idea of early conception of problem by asserting that the development of the human brain progresses through the hierarchy of nature. That is to say, as humans grow the brain also grows as such any deformity to the process may have a negative effect on the person. In the case of a baby born with any deformity, if that deformity is not seen or dealt with timely it will grow worse and may be out of hand. Therefore, the timely diagnosis of congenital deformity or abnormality is crucial to the survival of the child.

Moreover with a significant chi-square (0.001) and a phi of 0.564 the study revealed that there was an association between time of diagnoses and survival of the child. This was such that the early detection of these anomalies are able to help avert the challenges associated with the disease. This result goes a long way to accentuate the arguments raised by Tantibanchachai (2014) that the earlier conception of problem was better, because it could be eliminated or

managed. History about congenital abnormalities or malformations has shown that most of the children or patients die because of late diagnosis (Singh & Gupta, 2009), although sometimes nonfactual diagnosis, lack of equipment and skills may also be a cause to some death. However, this does not relegate the fact that early detection and diagnosis of a problem may be half way to solving the problem. Again, notwithstanding the fact that specific tests for all the abnormalities are clearly unfeasible in the average case, the mere possibility of their presence or recognition may bolster the chances of being corrected at an early age.

Again, the study showed that there was no relationship between the mode of delivery and the type of malformation incurred. That is whether the child if born by spontaneous virginal delivery or by caesarean section does not influence the presence or the type of malformation. This revelation was supported by Asante (2015); Kurinczuk et al (2010); Winterbottom (2001). For instance, Kurinczuk et al (2010) stated that several hundred of child defects were by structural, functional, metabolic and hereditary conditions. Again, Asante (2015); Vries et al. (2004) also linked some congenital abnormality to socio-economic factors and environmental factors and not mode of delivery of the child. Though Winterbottom (2001) had mentioned that 6 percent of total births with serious birth defect was of genetic or partially genetic origin, he continued to state that the mode delivery especially C-sections accounts for ≤ 2% of all the total birth defects. On Table 7, the result suggested that congenital abnormalities can occur

in a child irrespective of the fact that the child was born preterm (below 38 weeks) or at term or post term (beyond 42 weeks).

Maternal and perinatal risk factors of congenital abnormalities in children

Under this section, the main statistic used was the Logistic regression. Logistics regressions are used when the predicted or the dependent variable is a binary response or has only two responses. Again, according to Pallant (2011) before a regression model is accepted, the ANOVA statistic or the F-statistic must be significant. That is if the F-statistic in the ANOVA is $p > 0.05$ then the entire model falls short or is not good. Again the results revealed that all the variable apart from alcoholism, Oligohydramnios or polyhydramnios, exposure to radiation or chemicals, and breech presentation were statistically significant in affecting the presence of any malformation in children. These results affirm the studies of Kurinczuk et al (2010); Queiber and Spranger (2006) Singh and Gupta (2009).

For instance Smith and Jones (as cited in Singh & Gupta, 2009) mentioned that the use of alcohol, cocaine, thalidomide, Orange Agent, and vitamin A and its derivatives, called retionids or teratogen (drugs and chemicals) have a telling effect to an unborn child. The study went further to explain that alcohol, which is also a teratogen, can cause Fetal Alcohol Syndrome (FAS) in children born to women who drank too much alcohol while pregnant. FAS can cause defects such as minor facial abnormalities and damage to the brain, which consequently leads to learning and behavioural abnormalities. Kurinczuk et al (2010) also citing Fred Hale (1933) indicated that pregnant mothers who excessively use hard drugs like

cocaine and thalidomide are likely to have offspring with various congenital malformations such as anophthalmia (the absence of one or both eyes, and cleft palate). These studies above all showed that some maternal and perinatal characteristics or behaviours during pregnancy contribute to some congenital malformation in their offspring.

Additionally Queiber and Spranger (2006); and Winterbottom (2001) stated that hundreds of children were born with serious birth defects of post-conception origin, including maternal exposure to environmental agents (teratogens) such as alcohol, rubella, syphilis and iodine deficiency that can harm a developing fetus. This underscores the fact that not congenital malformations are not caused by deficiencies or intake of teratogens only but also exposure to radiations and other environmental agents. Parmer et al (2010) also noted that some congenital malformations can be inherited or acquired and this is attributed to known genetic conditions. That is to say if a mother has a malformation on any part of the body, it is possible to pass it to the offspring or for the child to inherit.

Wisniewska and Wysocki (2008) also supported the results on Table 8 by emphasizing that the lack of folic acid supplementation or foods fortified with Folic acid during periconception period is associated with occurrence of congenital anomalies. This is because folic acid is known to be necessary for growth and function of human cells. Godwin et al (2008) and the Perinatal Health report- Canada (2002) also confirmed this stance by expressing that folic acid is crucial for normal brain and spinal cord development during the first 4 weeks of gestation. And that the preconception use of folic acid has been proved to cause

substantial reduction of the risk for neural tube defects and other congenital anomalies like orofacial clefts, congenital heart diseases, urinary tract, limb and digestive system anomalies.

Suggestions to minimization of the Maternal and Perinatal Risk Factors

Here respondents gave suggestions to ways of minimizing the maternal and perinatal risk factors to congenital abnormalities. There were suggestions such as education on the need to start reproduction before 35yrs because maternal age may be a predisposing factor to birth defect. Other also mentioned, counseling on nutrition, education on the avoidance of teratogens, chemicals and radiation and intake of folic acid supplement. These suggestions were also in line with Arztebl (2006); Sipek (2008).

Sipek (2008) explained that exposure to Physical (x-rays, ionizing radiation, high temperature and mechanical factors), Chemical (exposure to alcohol, smoking, hard drugs, agrico-industrial industrial chemicals and cytostatic) and Biological (infectious agents, disease of the mother and other maternal conditions) poses a great danger for unborn children. The most telling effect is the increased occurrence of severe mental retardation, with or without microcephaly at specific gestational periods. Arztebl (2006) also cited the effects after the atomic bombings of Hiroshima and Nagasaki as critical example of what was likely to affect the fetus if they were exposed to radiations and other poisonous gases. Other studies have also pointed out that nutritional deficiencies and lack of folic acid intake may also give rise to malformations such as the clefts and palate, urinary tract, limb or club limbs and others (Parmer et al., 2010).

CHAPTER FIVE

SUMMARY, CONCLUSIONS AND RECOMMENDATION

Introduction

This chapter presents the summary of major findings of the study. The conclusions drawn from the study and recommendations made are also presented. The first section of this chapter summarizes the entire study and also presents the key findings. The subsequent sections cover the conclusions and recommendations drawn from the findings.

Summary of the Study

The purpose of the study was to investigate the maternal and perinatal contributing factors of congenital abnormalities in children below five years at the Child Health Department of the Korle Bu Teaching Hospital. An exploratory study was employed in determining the maternal and perinatal contributing factors of congenital abnormalities in children below five years. The study used set of questionnaires and medical history of patients and employed the convenience sampling technique to collect information from the respondents. Again, the study simply used the multivariate logistic regression to analyze how maternal and perinatal contributing factors were associated with congenital anomalies.

The study reviewed relevant literature on the subject both on the historical antecedent and theoretical. Literature was also reviewed on areas like the causes and definitions of congenital abnormality as well as other empirical literature on

the contributing factors associated with congenital malformations. For the general finding of the study;

Types of congenital abnormalities in children below five years seen at the Child Health Department, Korle Bu Teaching Hospital

- The study revealed that most of the children diagnosed of congenital anomaly suffered from heart disease (31.0%), anorectal (15.5%), Cleft lip & palate (5.0%), Hydrocephalus (5.5%), Down syndrome (4.4%), Equinoxarus (4.4%), and Cerebral palsy (2.2%) and other malformations.
- It was also found that most of these malformations affected the cardiovascular system (33.7%), digestive system (24%), and the nervous system (20.5%).
- Again, 26 children referred or admitted at the hospital representing 14.3% died from various forms of congenital anomalies.
- The study again found that most of the children diagnosed of with congenital malformations were diagnosed on admission at the hospital (58.6%), 40.3% were diagnosed at birth and just 1.1% diagnosed before delivery.
- The study moreover revealed that there were statistical relationship between time of diagnosis of congenital abnormality and the presence of the disorder such that the earlier the diagnosis the better it was for correction or management. On the whole children with congenital abnormalities have about 56.4% survival if they were diagnosed on time compared to malformations that were diagnosed late.

How maternal and perinatal characteristics contribute to congenital abnormalities in children less than five years.

- In addition to the above, the study found that various perinatal and maternal characteristics influence or contribute to the presence of various forms of malformation on the child.
- The use of illegal drugs and application herbal medication had very significant but negative influence on the health of an unborn child. Chances were that mothers who use some herbal medications and illegal drugs during pregnancy have 0.010 and 1.007 odds of reporting a child with congenital anomaly respectively.
- The study also found regular attendance to antenatal clinic, taking of medication giving at the clinic, and intake of folic acid to be significant contributors to good health of unborn children.

Possible suggestions to minimize the maternal and perinatal risk factors of congenital abnormalities

- The study revealed that education of mothers on avoidance of teratogens, chemicals and radiation by pregnant mothers may be a way to addressing the issue of congenital anomaly,
- Again 63.0% of the respondents mentioned that education of mothers or couples to start reproduction before 35 years was a strategy to reducing congenital malformations.
- There were other suggestions by respondents (98%) that giving of nutritional counseling to mothers and pregnant women is a possible

strategy to reducing congenital malformations

- Moreover, about 91% and 62% of respondents mentioned that avoidance of and immunized against Rubella, tetanus and other teratogenic chemicals and intake of folic acid were some importance methods towards minimizing congenital malformations.

Conclusions

The study sought to investigate the maternal and perinatal risk factors of congenital abnormalities in children under five years and it was evident from the study that there were lots of contributing factors to the formation of congenital malformations. It was concluded that the incidence of congenital malformation is very high given the number of cases diagnosed and the number of death over the study period. The study revealed that age at delivery of the baby was an issue that must not be joked with since; babies or children delivered beyond 38 weeks have 97.2% chance of reporting malformations. The study however reckoned that mothers seek medical attention on time because early diagnoses of the problem affect the chances of managing or even correction of the malformations.

Moreover, the study can conclude that indeed maternal and perinatal characteristics contributed to congenital malformation in children under five years. Characteristics such as regular attendance at the antenatal, intake of all medications given at the clinic, consistent intake of folic acid and good nutrition have significant and positive influence on the unborn children. These actions are able to minimize the incidence of congenital malformations significantly. However, direct exposure to radiation and high temperatures, excessive exposure

to smoke and agrico-industrial chemicals, intake of alcohol, smoking, hard drugs, and application of some herbal concoctions poses danger to children. They were able to affect the development of the fetus adversely.

Finally, the study concluded that there were possible ways of minimizing or controlling the emergence of congenital abnormalities in children. Ways such as educating and counseling of mothers to avoid at least the use of illegal drugs, application of herbal medications, avoid work that exposes them to direct radiations, high temperatures, and smoke during pregnancy. It was established that education on good dieting, minimal exercises, intake of folic acid, regular attendance to antenatal and taking all medication are ways of reducing the incidences of congenital malformations. There were other suggestions to mothers to if possible do well to start reproduction before 35 years.

Recommendation

Based on the findings and conclusions made, the researcher deemed it necessary to make the following recommendations:

1. Mothers should be advised to consider the intake of folic acid and all medication given at the antenatal serious since they have great impact on the cell development and protection against infections which in turn may make the fetus susceptible malformations.
2. Mothers also should be admonished not skip the attendance of antenatal clinics. This is because any malformations could easily be detected or diagnosed and dealt with on time.
3. The study also advocates that women regulate or minimize the application

of herbal concoctions, alcohol drinking, smoking, illegal drugs, and other teratogenic chemicals. This is because excessive usage of these may adverse consequences on the unborn child.

4. Again, the study recommends that the hospital facility must do well to possibly screen all pregnant women of any malformations when they attend hospital or antenatal. Doctor and midwives must also take the opportunity of the antenatal attendance to educate mothers on the good nutrition and avoidance of hard drugs. They may also educate couples on ways of planning their families.
5. The hospital can also take a stance such that it makes sure all medications for pregnant women are taken at the hospital before leaving. This will in a way reduce the issues of not taking all their medications.
6. Finally, care givers and doctors or midwives must pay particular attention to pregnant women whose pregnancies have gone beyond the normal gestation periods.

Suggestions for Further Research

The researcher recommends these areas for further research

- The psychological impact of congenital abnormalities on the family.
- How educational programs can be organized to create awareness on the various types of congenital abnormalities and their management.

Implication for Nursing Practice

- All newborn babies must be properly assessed for congenital abnormalities for prompt management.
- Mothers should encourage to state reproduction early in life.
- Health education of the public on congenital abnormalities must be interfered to create awareness.

Implication for Nursing Education

- Course on obstetric and pediatric care must create more emphasis on congenital abnormalities to equip students in identification and management to congenital abnormalities

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APPENDIX A

QUESTIONNAIRE

This questionnaire forms part of a thesis conducted by a final year Master of Nursing student from the School of Nursing, University of Cape Coast on CONGENITAL ABNORMALITIES. It is meant to solicit information on MATERNAL AND PERINATAL RISK FACTORS OF CONGENITAL ABNORMALITIES IN CHILDREN BELOW FIVE YEARS in Ghana. All information provided therefore will be used strictly for academic purposes and shall be treated with utmost confidentiality. Please tick the appropriate option(s) provided under each question and fill in the spaces provided where applicable.

Demographic data of the Child

1. Age of child

   0-1 year ( ) 1 -3 years ( ) 3 -5 years ( )

2. Sex: Male ( ) Female ( )
3. Mode of delivery: Spontaneous Vaginal Delivery ( ) Caesarean Section ( )
4. Birth weight: less than 1kg ( ) 1.1kg-2kg ( ) 2.1- 2.5kg ( ) 2.6 -3.9 kg ( ) 4kg and above ( )
5. Age at delivery: less than 38 weeks ( ) 39 – 41 weeks ( )

   42 and above ( )

SECTION A: TYPE OF CONGENITAL ABNORMALITY SEEN AT THE CHILD HEALTH DEPARTMENT, KORLE BU TEACHING HOSPITAL

6. Does the child have any congenital abnormality? yes ( ) No ( )
7. When was it diagnosed?

   Before delivery ( ) At birth ( ) On admission ( )
8. The type of malformation. ……..
9. System affected……..
10. Does other malformations exist: yes ( ) No ( )
11. Does the child has any other medical condition yes ( ) No ( )
12. If yes, please specify…….

SECTION B:

MATERNAL CHARACTERISTICS AND THE PERINATAL RISK FACTORS THAT RESULT IN CONGENITAL ABNORMALITIES

13. Maternal Age: Under 20 ( ) 21- 40 ( ) 41- 50 ( )
14. Religious denomination

    Traditional ( ) Moslem ( ) Christian ( ) Other ( )
15. Mothers occupation……
16. Does maternal occupation exposes her to any radiation or chemicals?

    Yes ( ) No ( )

17. PLEASE TICK WHEN APPROPRIATE

Maternal medical and surgical history

	Yes	no
Hypertension	( )	( )
Heart disease	( )	( )
Sickle Cell Disease	( )	( )
Diabetes	( )	( )
TB, Chronic cough, asthma	( )	( )
HIV disease	( )	( )
Epilepsy	( )	( )
Hepatitis infection	( )	( )
Sexually transmitted disease	( )	( )

Others (specify)……

Family history

Hypertension	( )	( )
Diabetes	( )	( )
Sickle cell disease	( )	( )
Multiple pregnancies	( )	( )
Birth defects	( )	( )

If yes (specify)

Drug history

Smoking	( )	( )
Alcohol intake	( )	( )

Herbal medication	( )	( )
Illicit drug	( )	( )

Obstetric and Gynecological History

18. Number of children: Dead …….. Alive……..
19. Any congenital abnormalities in any of the children? yes ( ) no ( )
20. Any gynecological condition yes ( ) No ( )
21. If yes, please specify……
22. Was she a regular attendant at antenatal clinics during pregnancy? Yes( ) No ( )
23. Was any diagnosis of congenital abnormalities made during pregnancy before delivery? Yes ( ) No ( )
24. If there any history of herbal medication during pregnancy? Yes ( ) No ( )
25. Did the mother take all medications given at the antenatal clinic? Yes ( ) No ( )

26. Folic acid intake	Yes ( )	No ( )
27. Pregnancy induced hypertension	Yes ( )	No ( )
28. vaginal bleeding early in pregnancy	Yes ( )	No ( )
29. twin pregnancy	Yes ( )	No ( )
30. history of previous abortions and still births	Yes ( )	No ( )

31. if yes, please state the possible cause

SECTION C: WAYS TO MINIMIZE CONGENITAL ABNORMALITIES IN CHILDREN BELOW FIVE YEARS

32. Education to start reproduction before 35 years Yes ( ) No ( )
33. Rubella tetanus and other immunization Yes ( ) No ( )
34. Peri-conceptional supplementation of folic acid Yes ( ) No ( )
35. Nutrition counseling Yes ( ) No ( )
36. Control of maternal infections after diagnosis Yes ( ) No ( )
37. Education on avoidance of teratogens including drugs, chemicals and Radiation

    Yes ( ) No ( )
38. The provision of genetic services for couples with family history of congenital anomalies Yes ( ) No ( )
39. Newborn examination to screen for congenital anomalies Yes ( ) No ( )

THANK YOU!!!

APPENDIX B

INTERNATIONAL CLASSIFICATION OF DISEASES - XVII CONGENITAL MALFORMATIONS

Embryology - 12 Jul 2017 **Expand to Translate**

Contents

- 6.1 Q35 Cleft palate
- 6.2 Q36 Cleft lip
- 6.3 Q37.0 Cleft hard palate with bilateral cleft lip
- 7 Other congenital malformations of the digestive system (Q38-Q45)
- 7.1 Q38 Other congenital malformations of tongue, mouth and pharynx
- 7.2 Q39 Congenital malformations of oesophagus
- 7.3 Q40 Other congenital malformations of upper alimentary tract
- 7.4 Q41 Congenital absence, atresia and stenosis of small intestine
- 7.5 Q42 Congenital absence, atresia and stenosis of large intestine
- 7.6 Q43 Other congenital malformations of intestine
- 7.7 Q44 Congenital malformations of gallbladder, bile ducts and liver
- 7.8 Q45 Other congenital malformations of digestive system
- 8 Congenital malformations of genital organs (Q50-Q56)
- 8.1 Q50 Congenital malformations of ovaries, fallopian tubes and broad ligaments
- 8.2 Q51 Congenital malformations of uterus and cervix
- 8.3 Q52 Other congenital malformations of female genitalia
- 8.4 Q53 Undescended testicle
- 8.5 Q54 Hypospadias
- 8.6 Q55 Other congenital malformations of male genital organs
- 8.7 Q56 Indeterminate sex and pseudohermaphroditism
- 9 Congenital malformations of the urinary system (Q60-Q64)
- 9.1 Q60 Renal agenesis and other reduction defects of kidney
- 9.2 Q61 Cystic kidney disease
- 9.3 Q62 Congenital obstructive defects of renal pelvis and congenital malformations of ureter
- 9.4 Q63 Other congenital malformations of kidney
- 9.5 Q64 Other congenital malformations of urinary system
- 10 Congenital malformations and deformations of the musculoskeletal system (Q65-Q79)
- 10.1 Q65 Congenital deformities of hip
- 10.2 Q66 Congenital deformities of feet
- 10.3 Q67 Congenital musculoskeletal deformities of head, face, spine and chest
- 10.4 Q68 Other congenital musculoskeletal deformities
- 10.5 Q69 Polydactyly
- 10.6 Q70 Syndactyly
- 10.7 Q71 Reduction defects of upper limb
- 10.8 Q72 Reduction defects of lower limb
- 10.9 Q73 Reduction defects of unspecified limb
- 10.10 Q74 Other congenital malformations of limb(s)
- 10.11 Q75 Other congenital malformations of skull and face bones
- 10.12 Q76 Congenital malformations of spine and bony thorax
- 10.13 Q77 Osteochondrodysplasia with defects of growth of tubular bones and spine
- 10.14 Q78 Other osteochondrodysplasias

Introduction

The International Classification of Diseases (ICD) World Health Organization's classification used worldwide as the standard diagnostic tool for epidemiology, health management and clinical purposes. This includes the analysis of the general health situation of population groups. It is used to monitor the incidence and prevalence of diseases and other health problems. Within this classification "congenital malformations, deformations and chromosomal abnormalities" are (Q00-Q99) but excludes "inborn errors of metabolism" (E70-E90).

(ICD) ICD-10 was endorsed by the Forty-third World Health Assembly in May 1990 and came into use in WHO Member States as from 1994. The classification is the latest in a series which has its origins in the 1850s. The first edition, known as the International List of Causes of Death, was adopted by the International Statistical Institute in 1893. WHO took over the responsibility for the ICD at its creation in 1948 when

the Sixth Revision, which included causes of morbidity for the first time, was published. The World Health Assembly adopted in 1967 the WHO Nomenclature Regulations that stipulate use of ICD in its most current revision for mortality and morbidity statistics by all Member States.

The ICD is the international standard diagnostic classification for all general epidemiological, many health management purposes and clinical use.

ICD-10 Links: **XVII Congenital Malformations** | System Tables | XVI Perinatal Period | XV Pregnancy Childbirth | Abnormal Development | Reports

International Statistical Classification of Diseases and Related Health Problems 10th Revision (ICD-10) Version for 2010

Chapter XVII Congenital malformations, deformations and chromosomal abnormalities (Q00-Q99)

Excludes inborn errors of metabolism (E70-E90)

Internal site links on related pages include content additional to the ICD codes.

Congenital malformations of the nervous system (Q00-Q07)

Links: Neural System - Abnormalities | Neural Crest Abnormalities | WHO Links

Q00 Anencephaly and similar malformations

- Q00.0 Anencephaly, Acephaly, Acrania, Amyelencephaly, Hemianencephaly, Hemicephaly
- Q00.1 Craniorachischisis
- Q00.2 Iniencephaly

Q01 Encephalocele

Incl.: encephalomyelocele, hydroencephalocele, hydromeningocele, cranial meningocele, cerebral meningoencephalocele

Excl.: Meckel-Gruber syndrome (Q61.9)

- Q01.0 Frontal encephalocele
- Q01.1 Nasofrontal encephalocele
- Q01.2 Occipital encephalocele
- Q01.8 Encephalocele of other sites
- Q01.9 Encephalocele, unspecified

Q02 Microcephaly

Incl.: Hydromicrocephaly Micrencephalon Excl.: Meckel-Gruber syndrome (Q61.9)

Q03 Congenital hydrocephalus

Q03 Congenital hydrocephalus Incl.: hydrocephalus in newborn Excl.: Arnold-Chiari syndrome (Q07.0) hydrocephalus: acquired (G91.-) due to congenital toxoplasmosis (P37.1) with spina bifida (Q05.0-Q05.4)

- Q03.0 Malformations of aqueduct of Sylvius Aqueduct of Sylvius: anomaly obstruction, congenital stenosis

- Q03.1 Atresia of foramina of Magendie and Luschka Dandy-Walker syndrome
- Q03.8 Other congenital hydrocephalus
- Q03.9 Congenital hydrocephalus, unspecified

Q04 Other congenital malformations of brain

Excl.: cyclopia (Q87.0) macrocephaly (Q75.3)

- Q04.0 Congenital malformations of corpus callosum, Agenesis of corpus callosum
- Q04.1 Arhinencephaly
- Q04.2 Holoprosencephaly
- Q04.3 Other reduction deformities of brain, Absence, Agenesis, Aplasia, Hypoplasia of part of brain, Agyria, Hydranencephaly, Lissencephaly, Microgyria, Pachygyria Excl.: congenital malformations of corpus callosum (Q04.0)
- Q04.4 Septo-optic dysplasia
- Q04.5 Megalencephaly
- Q04.6 Congenital cerebral cysts, Porencephaly, Schizencephaly, Excl.: acquired porencephalic cyst (G93.0)
- Q04.8 Other specified congenital malformations of brain, Macrogyria
- Q04.9 Congenital malformation of brain, unspecified Congenital: anomaly, deformity, disease or lesion, multiple anomalies NOS of brain

Q05 Spina bifida

Incl.: hydromeningocele (spinal), meningocele (spinal), meningomyelocele, myelocele, myelomeningocele, rachischisis, spina bifida (aperta)(cystica), syringomyelocele Excl.: Arnold-Chiari syndrome (Q07.0), spina bifida occulta (Q76.0)

- Q05.0 Cervical spina bifida with hydrocephalus
- Q05.1 Thoracic spina bifida with hydrocephalus Spina bifida: dorsal thoracolumbar with hydrocephalus
- Q05.2 Lumbar spina bifida with hydrocephalus, Lumbosacral spina bifida with hydrocephalus
- Q05.3 Sacral spina bifida with hydrocephalus
- Q05.4 Unspecified spina bifida with hydrocephalus
- Q05.5 Cervical spina bifida without hydrocephalus
- Q05.6 Thoracic spina bifida without hydrocephalus Spina bifida: dorsal NOS, thoracolumbar NOS
- Q05.7 Lumbar spina bifida without hydrocephalus, Lumbosacral spina bifida NOS
- Q05.8 Sacral spina bifida without hydrocephalus
- Q05.9 Spina bifida, unspecified

Q06 Other congenital malformations of spinal cord

- Q06.0 Amyelia

- Q06.1 Hypoplasia and dysplasia of spinal cord, Atelomyelia, Myelatelia, Myelodysplasia of spinal cord
- Q06.2 Diastematomyelia
- Q06.3 Other congenital cauda equina malformations
- Q06.4 Hydromyelia Hydrorachis
- Q06.8 Other specified congenital malformations of spinal cord
- Q06.9 Congenital malformation of spinal cord, unspecified Congenital: anomaly, deformity, disease or lesion, NOS of spinal cord or meninges

Q07 Other congenital malformations of nervous system

Excl.: familial dysautonomia [Riley-Day] (G90.1), neurofibromatosis (nonmalignant) (Q85.0)

- Q07.0 Arnold-Chiari syndrome
- Q07.8 Other specified congenital malformations of nervous system Agenesis of nerve, Displacement of brachial plexus, Jaw-winking syndrome, Marcus Gunn's syndrome
- Q07.9 Congenital malformation of nervous system, unspecified Congenital: anomaly, deformity, disease or lesion, NOS of nervous system

Links: Neural System - Abnormalities | Neural Crest Abnormalities

Congenital malformations of eye, ear, face and neck (Q10-Q18)

Excl.: cleft lip and cleft palate (Q35-Q37) congenital malformation of: cervical spine (Q05.0, Q05.5, Q67.5, Q76.0-Q76.4) larynx (Q31.-) lip NEC (Q38.0) nose (Q30.-) parathyroid gland (Q89.2) thyroid gland (Q89.2)

Q10 Congenital malformations of eyelid, lacrimal apparatus and orbit

Excl.: cryptophthalmos: NOS (Q11.2) syndrome (Q87.0)

- Q10.0 Congenital ptosis
- Q10.1 Congenital ectropion
- Q10.2 Congenital entropion
- Q10.3 Other congenital malformations of eyelid Ablepharon Absence or agenesis of: cilia eyelid Accessory: eye muscle Blepharophimosis, congenital Coloboma of eyelid Congenital malformation of eyelid NOS
- Q10.4 Absence and agenesis of lacrimal apparatus Absence of punctum lacrimale
- Q10.5 Congenital stenosis and stricture of lacrimal duct
- Q10.6 Other congenital malformations of lacrimal apparatus Congenital malformation of lacrimal apparatus NOS
- Q10.7 Congenital malformation of orbit

Q11 Anophthalmos, microphthalmos and macrophthalmos

- Q11.0 Cystic eyeball
- Q11.1 Other anophthalmos Agenesis Aplasia of eye

- Q11.2 Microphthalmos Cryptophthalmos NOS Dysplasia of eye Hypoplasia of eye Rudimentary eye Excl.: cryptophthalmos syndrome (Q87.0)
- Q11.3 Macrophthalmos Excl.: macrophthalmos in congenital glaucoma (Q15.0)

Q12 Congenital lens malformations

- Q12.0 Congenital cataract
- Q12.1 Congenital displaced lens
- Q12.2 Coloboma of lens
- Q12.3 Congenital aphakia
- Q12.4 Spherophakia
- Q12.8 Other congenital lens malformations
- Q12.9 Congenital lens malformation, unspecified

Q13 Congenital malformations of anterior segment of eye

- Q13.0 Coloboma of iris Coloboma NOS
- Q13.1 Absence of iris Aniridia
- Q13.2 Other congenital malformations of iris Anisocoria, congenital Atresia of pupil Congenital malformation of iris NOS Corectopia
- Q13.3 Congenital corneal opacity
- Q13.4 Other congenital corneal malformations Congenital malformation of cornea NOS Microcornea Peter's anomaly
- Q13.5 Blue sclera
- Q13.8 Other congenital malformations of anterior segment of eye Rieger's anomaly
- Q13.9 Congenital malformation of anterior segment of eye, unspecified

Q14 Congenital malformations of posterior segment of eye

- Q14.0 Congenital malformation of vitreous humour Congenital vitreous opacity
- Q14.1 Congenital malformation of retina Congenital retinal aneurysm
- Q14.2 Congenital malformation of optic disc Coloboma of optic disc
- Q14.3 Congenital malformation of choroid
- Q14.8 Other congenital malformations of posterior segment of eye Coloboma of the fundus
- Q14.9 Congenital malformation of posterior segment of eye, unspecified

Q15 Other congenital malformations of eye

Excl.: congenital nystagmus (H55) ocular albinism (E70.3) retinitis pigmentosa (H35.5)

- Q15.0 Congenital glaucoma Buphthalmos Glaucoma of newborn Hydrophthalmos Keratoglobus, congenital, with glaucoma Macrocornea with glaucoma Macrophthalmos in congenital glaucoma Megalocornea with glaucoma
- Q15.8 Other specified congenital malformations of eye
- Q15.9 Congenital malformation of eye, unspecified Congenital: anomaly deformity NOS of eye

Q16 Congenital malformations of ear causing impairment of hearing

Excl.: congenital deafness (H90.-)

- Q16.0 Congenital absence of (ear) auricle
- Q16.1 Congenital absence, atresia and stricture of auditory canal (external) Atresia or stricture of osseous meatus
- Q16.2 Absence of eustachian tube
- Q16.3 Congenital malformation of ear ossicles Fusion of ear ossicles
- Q16.4 Other congenital malformations of middle ear Congenital malformation of middle ear NOS
- Q16.5 Congenital malformation of inner ear Anomaly: membranous labyrinth organ of Corti
- Q16.9 Congenital malformation of ear causing impairment of hearing, unspecified Congenital absence of ear NOS

Q17 Other congenital malformations of ear Excl.: preauricular sinus (Q18.1)

- Q17.0 Accessory auricle Accessory tragus Polyotia Preauricular appendage or tag Supernumerary: ear lobule
- Q17.1 Macrotia
- Q17.2 Microtia
- Q17.3 Other misshapen ear Pointed ear
- Q17.4 Misplaced ear Low-set ears Excl.: cervical auricle (Q18.2)
- Q17.5 Prominent ear Bat ear
- Q17.8 Other specified congenital malformations of ear Congenital absence of lobe of ear
- Q17.9 Congenital malformation of ear, unspecified Congenital anomaly of ear NOS

Q18 Other congenital malformations of face and neck

Excl.: cleft lip and cleft palate (Q35-Q37) conditions classified to

- Q67.0-Q67.4 congenital malformations of skull and face bones (Q75.-) cyclopia (Q87.0) dentofacial anomalies [including malocclusion] (K07.-) malformation syndromes affecting facial appearance (Q87.0) persistent thyroglossal duct (Q89.2)
- Q18.0 Sinus, fistula and cyst of branchial cleft Branchial vestige
- Q18.1 Preauricular sinus and cyst Fistula (of): auricle, congenital cervicoaural Pretragal sinus and cyst
- Q18.2 Other branchial cleft malformations Branchial cleft malformation NOS Cervical auricle Otocephaly
- Q18.3 Webbing of neck Pterygium colli
- Q18.4 Macrostomia
- Q18.5 Microstomia
- Q18.6 Macrocheilia Hypertrophy of lip, congenital
- Q18.7 Microcheilia
- Q18.8 Other specified congenital malformations of face and neck Medial: cyst fistula sinus of face and neck
- Q18.9 Congenital malformation of face and neck, unspecified Congenital anomaly NOS of face and neck

Links: Head Abnormalities | Hearing Abnormalities | Vision Abnormalities | Smell Abnormalities

Congenital malformations of the circulatory system (Q20-Q28)

Cardiovascular System - Abnormalities

ICD10 Congenital malformations of the circulatory system (Q20-Q28)

ICD-11 Structural developmental anomalies of the circulatory system (draft)

Q20 Congenital malformations of cardiac chambers and connections

Excl.: dextrocardia with situs inversus (Q89.3) mirror-image atrial arrangement with situs inversus (Q89.3)

- Q20.0 Common arterial trunk Persistent truncus arteriosus
- Q20.1 Double outlet right ventricle Taussig-Bing syndrome
- Q20.2 Double outlet left ventricle
- Q20.3 Discordant ventriculoarterial connection Dextrotransposition of aorta Transposition of great vessels (complete)
- Q20.4 Double inlet ventricle Common ventricle Cor triloculare biatriatum Single ventricle
- Q20.5 Discordant atrioventricular connection Corrected transposition Laevotransposition Ventricular inversion
- Q20.6 Isomerism of atrial appendages Isomerism of atrial appendages with asplenia or polysplenia
- Q20.8 Other congenital malformations of cardiac chambers and connections
- Q20.9 Congenital malformation of cardiac chambers and connections, unspecified

Q21 Congenital malformations of cardiac septa

Excl.: acquired cardiac septal defect (I51.0)

- Q21.0 Ventricular septal defect
- Q21.1 Atrial septal defect Coronary sinus defect Patent or persistent: foramen ovale ostium secundum defect (type II) Sinus venosus defect
- Q21.2 Atrioventricular septal defect Common atrioventricular canal Endocardial cushion defect Ostium primum atrial septal defect (type I)
- Q21.3 Tetralogy of Fallot Ventricular septal defect with pulmonary stenosis or atresia, dextroposition of aorta and hypertrophy of right ventricle.
- Q21.4 Aortopulmonary septal defect Aortic septal defect Aortopulmonary window
- Q21.8 Other congenital malformations of cardiac septa Eisenmenger's defect Pentalogy of Fallot Excl.: Eisenmenger's complex (I27.8) syndrome (I27.8)
- Q21.9 Congenital malformation of cardiac septum, unspecified Septal (heart) defect NOS

Q22 Congenital malformations of pulmonary and tricuspid valves

- Q22.0 Pulmonary valve atresia
- Q22.1 Congenital pulmonary valve stenosis
- Q22.2 Congenital pulmonary valve insufficiency Congenital pulmonary valve regurgitation
- Q22.3 Other congenital malformations of pulmonary valve Congenital malformation of pulmonary valve NOS
- Q22.4 Congenital tricuspid stenosis Tricuspid atresia
- Q22.5 Ebstein's anomaly
- Q22.6 Hypoplastic right heart syndrome
- Q22.8 Other congenital malformations of tricuspid valve
- Q22.9 Congenital malformation of tricuspid valve, unspecified

Q23 Congenital malformations of aortic and mitral valves

- Q23.0 Congenital stenosis of aortic valve Congenital aortic: atresia stenosis Excl.: congenital subaortic stenosis (Q24.4) that in hypoplastic left heart syndrome (Q23.4)
- Q23.1 Congenital insufficiency of aortic valve Bicuspid aortic valve Congenital aortic insufficiency
- Q23.2 Congenital mitral stenosis Congenital mitral atresia
- Q23.3 Congenital mitral insufficiency
- Q23.4 Hypoplastic left heart syndrome Atresia, or marked hypoplasia of aortic orifice or valve, with hypoplasia of ascending aorta and defective develop-ment of left ventricle (with mitral valve stenosis or atresia).
- Q23.8 Other congenital malformations of aortic and mitral valves
- Q23.9 Congenital malformation of aortic and mitral valves, unspecified

Q24 Other congenital malformations of heart

Excl.: endocardial fibroelastosis (I42.4)

- Q24.0 Dextrocardia Excl.: dextrocardia with situs inversus (Q89.3) isomerism of atrial appendages (with asplenia or polysplenia) (Q20.6) mirror-image atrial arrangement with situs inversus (Q89.3)
- Q24.1 Laevocardia Location of heart in left hemithorax with apex pointing to the left, but with situs inversus of other viscera and defects of the heart, or corrected transposition of great vessels.
- Q24.2 Cor triatriatum
- Q24.3 Pulmonary infundibular stenosis
- Q24.4 Congenital subaortic stenosis
- Q24.5 Malformation of coronary vessels Congenital coronary (artery) aneurysm
- Q24.6 Congenital heart block
- Q24.8 Other specified congenital malformations of heart Congenital: diverticulum of left ventricle malformation of: myocardium pericardium Malposition of heart Uhl's disease

- Q24.9 Congenital malformation of heart, unspecified Congenital: anomaly disease NOS of heart

Q25 Congenital malformations of great arteries

- Q25.0 Patent ductus arteriosus Patent ductus Botallo Persistent ductus arteriosus
- Q25.1 Coarctation of aorta Coarctation of aorta (preductal)(postductal)
- Q25.2 Atresia of aorta
- Q25.3 Stenosis of aorta Supravalvular aortic stenosis Excl.: congenital aortic stenosis (Q23.0)
- Q25.4 Other congenital malformations of aorta Absence Aplasia Congenital: aneurysm dilatation of aorta Aneurysm of sinus of Valsalva (ruptured) Double aortic arch [vascular ring of aorta] Hypoplasia of aorta Persistent: convolutions of aortic arch right aortic arch Excl.: hypoplasia of aorta in hypoplastic left heart syndrome (Q23.4)
- Q25.5 Atresia of pulmonary artery
- Q25.6 Stenosis of pulmonary artery Supravalvular pulmonary stenosis
- Q25.7 Other congenital malformations of pulmonary artery Aberrant pulmonary artery Agenesis Aneurysm, congenital Anomaly Hypoplasia of pulmonary artery Pulmonary arteriovenous aneurysm
- Q25.8 Other congenital malformations of great arteries
- Q25.9 Congenital malformation of great arteries, unspecified

Q26 Congenital malformations of great veins

- Q26.0 Congenital stenosis of vena cava Congenital stenosis of vena cava (inferior)(superior)
- Q26.1 Persistent left superior vena cava
- Q26.2 Total anomalous pulmonary venous connection
- Q26.3 Partial anomalous pulmonary venous connection
- Q26.4 Anomalous pulmonary venous connection, unspecified
- Q26.5 Anomalous portal venous connection
- Q26.6 Portal vein-hepatic artery fistula
- Q26.8 Other congenital malformations of great veins Absence of vena cava (inferior)(superior) Azygos continuation of inferior vena cava Persistent left posterior cardinal vein Scimitar syndrome
- Q26.9 Congenital malformation of great vein, unspecified Anomaly of vena cava (inferior)(superior) NOS

Q27 Other congenital malformations of peripheral vascular system

Excl.: anomalies of: cerebral and precerebral vessels (Q28.0-Q28.3) coronary vessels (Q24.5) pulmonary artery (Q25.5-Q25.7) congenital retinal aneurysm (Q14.1) haemangioma and lymphangioma (D18.-)

- Q27.0 Congenital absence and hypoplasia of umbilical artery Single umbilical artery
- Q27.1 Congenital renal artery stenosis
- Q27.2 Other congenital malformations of renal artery Congenital malformation of renal artery NOS Multiple renal arteries

- Q27.3 Peripheral arteriovenous malformation Arteriovenous aneurysm Excl.: ac
- Quired arteriovenous aneurysm (I77.0)
- Q27.4 Congenital phlebectasia
- Q27.8 Other specified congenital malformations of peripheral vascular system Aberrant subclavian artery Absence Atresia of artery or vein NEC Congenital: aneurysm (peripheral) stricture, artery varix
- Q27.9 Congenital malformation of peripheral vascular system, unspecified Anomaly of artery or vein NOS

Q28 Other congenital malformations of circulatory system

Excl.: congenital aneurysm: NOS (Q27.8) coronary (Q24.5) peripheral (Q27.8) pulmonary (Q25.7) retinal (Q14.1) ruptured: cerebral arteriovenous malformation (I60.8) malformation of precerebral vessels (I72.-)

- Q28.0 Arteriovenous malformation of precerebral vessels Congenital arteriovenous precerebral aneurysm (nonruptured)
- Q28.1 Other malformations of precerebral vessels Congenital: malformation of precerebral vessels NOS precerebral aneurysm (nonruptured)
- Q28.2 Arteriovenous malformation of cerebral vessels Arteriovenous malformation of brain NOS Congenital arteriovenous cerebral aneurysm (nonruptured)
- Q28.3 Other malformations of cerebral vessels Congenital: cerebral aneurysm (nonruptured) malformation of cerebral vessels NOS
- Q28.8 Other specified congenital malformations of circulatory system Congenital aneurysm, specified site NEC
- Q28.9 Congenital malformation of circulatory system, unspecified

Links: Cardiovascular System - Abnormalities

Congenital malformations of the respiratory system (Q30-Q34)

Links: Respiratory Abnormalities

Q30 Congenital malformations of nose

Excl.: congenital deviation of nasal septum (Q67.4)

- Q30.0 Choanal atresia Atresia Congenital stenosis of nares (anterior)(posterior)
- Q30.1 Agenesis and underdevelopment of nose Congenital absence of nose
- Q30.2 Fissured, notched and cleft nose
- Q30.3 Congenital perforated nasal septum
- Q30.8 Other congenital malformations of nose Accessory nose Congenital anomaly of nasal sinus wall
- Q30.9 Congenital malformation of nose, unspecified

Q31 Congenital malformations of larynx

Excl.: congenital (laryngeal) stridor NOS (P28.8)

- Q31.0 Web of larynx Web of larynx: NOS glottic subglottic
- Q31.1 Congenital subglottic stenosis
- Q31.2 Laryngeal hypoplasia
- Q31.3 Laryngocele
- Q31.5 Congenital laryngomalacia
- Q31.8 Other congenital malformations of larynx Absence Agenesis Atresia of cricoid cartilage, epiglottis, glottis, larynx or thyroid cartilage Cleft thyroid cartilage Congenital stenosis of larynx NEC Fissure of epiglottis Posterior cleft of cricoid cartilage
- Q31.9 Congenital malformation of larynx, unspecified

Q32 Congenital malformations of trachea and bronchus

Excl.: congenital bronchiectasis (Q33.4)

- Q32.0 Congenital tracheomalacia
- Q32.1 Other congenital malformations of trachea Anomaly of tracheal cartilage Atresia of trachea Congenital: dilatation malformation stenosis of trachea Congenital tracheocele
- Q32.2 Congenital bronchomalacia
- Q32.3 Congenital stenosis of bronchus
- Q32.4 Other congenital malformations of bronchus Absence Agenesis Atresia Congenital malformation NOS Diverticulum of bronchus

Q33 Congenital malformations of lung

- Q33.0 Congenital cystic lung Congenital: honeycomb lung lung disease: cystic polycystic Excl.: cystic lung disease, acquired or unspecified (J98.4)
- Q33.1 Accessory lobe of lung
- Q33.2 Sequestration of lung
- Q33.3 Agenesis of lung Absence of lung (lobe)
- Q33.4 Congenital bronchiectasis
- Q33.5 Ectopic tissue in lung
- Q33.6 Hypoplasia and dysplasia of lung Excl.: pulmonary hypoplasia associated with short gestation (P28.0)
- Q33.8 Other congenital malformations of lung
- Q33.9 Congenital malformation of lung, unspecified

Q34 Other congenital malformations of respiratory system

- Q34.0 Anomaly of pleura
- Q34.1 Congenital cyst of mediastinum
- Q34.8 Other specified congenital malformations of respiratory system Atresia of nasopharynx
- Q34.9 Congenital malformation of respiratory system, unspecified Congenital: absence anomaly NOS of respiratory organ

Links: Respiratory Abnormalities

Cleft lip and cleft palate (Q35-Q37)

Use additional code (Q30.2), if desired, to identify associated malformations of the nose. Excl.: Robin's syndrome (Q87.0)

Links: Cleft lip and cleft palate

Q35 Cleft palate

Incl.: fissure of palate palatoschisis Excl.: cleft palate with cleft lip (Q37.-)

- Q35.1 Cleft hard palate
- Q35.3 Cleft soft palate
- Q35.5 Cleft hard palate with cleft soft palate
- Q35.7 Cleft uvula
- Q35.9 Cleft palate, unspecified

Q36 Cleft lip

Incl.: cheiloschisis congenital fissure of lip harelip labium leporinum Excl.: cleft lip with cleft palate (Q37.-)

- Q36.0 Cleft lip, bilateral
- Q36.1 Cleft lip, median
- Q36.9 Cleft lip, unilateral Cleft lip NOS
- Q37 Cleft palate with cleft lip

Q37.0 Cleft hard palate with bilateral cleft lip

- Q37.1 Cleft hard palate with unilateral cleft lip Cleft hard palate with cleft lip NOS
- Q37.2 Cleft soft palate with bilateral cleft lip
- Q37.3 Cleft soft palate with unilateral cleft lip Cleft soft palate with cleft lip NOS
- Q37.4 Cleft hard and soft palate with bilateral cleft lip
- Q37.5 Cleft hard and soft palate with unilateral cleft lip Cleft hard and soft palate with cleft lip NOS
- Q37.8 Unspecified cleft palate with bilateral cleft lip
- Q37.9 Unspecified cleft palate with unilateral cleft lip Cleft palate with cleft lip NOS

Links: Cleft lip and cleft palate

Other congenital malformations of the digestive system (Q38-Q45)

Links: Gastrointestinal Abnormalities

Q38 Other congenital malformations of tongue, mouth and pharynx

Excl.: macrostomia (Q18.4) microstomia (Q18.5)

- Q38.0 Congenital malformations of lips, not elsewhere classified Congenital: fistula of lip malformation of lip NOS Van der Woude's syndrome Excl.: cleft lip (Q36.-) cleft lip with cleft palate (Q37.-) macrocheilia (Q18.6) microcheilia (Q18.7)
- Q38.1 Ankyloglossia Tongue tie
- Q38.2 Macroglossia
- Q38.3 Other congenital malformations of tongue Aglossia Bifid tongue Congenital: adhesion fissure malformation NOS of tongue Hypoglossia Hypoplasia of tongue Microglossia

- Q38.4 Congenital malformations of salivary glands and ducts Absence Accessory Atresia (of) salivary gland or duct Congenital fistula of salivary gland
- Q38.5 Congenital malformations of palate, not elsewhere classified Absence of uvula Congenital malformation of palate NOS High arched palate Excl.: cleft palate (Q35.-) cleft palate with cleft lip (Q37.-)
- Q38.6 Other congenital malformations of mouth Congenital malformation of mouth NOS
- Q38.7 Pharyngeal pouch Diverticulum of pharynx Excl.: pharyngeal pouch syndrome (D82.1)
- Q38.8 Other congenital malformations of pharynx Congenital malformation of pharynx NOS

Q39 Congenital malformations of oesophagus

- Q39.0 Atresia of oesophagus without fistula Atresia of oesophagus NOS
- Q39.1 Atresia of oesophagus with tracheo-oesophageal fistula Atresia of oesophagus with broncho-oesophageal fistula
- Q39.2 Congenital tracheo-oesophageal fistula without atresia Congenital tracheo-oesophageal fistula NOS
- Q39.3 Congenital stenosis and stricture of oesophagus
- Q39.4 Oesophageal web
- Q39.5 Congenital dilatation of oesophagus
- Q39.6 Diverticulum of oesophagus Oesophageal pouch
- Q39.8 Other congenital malformations of oesophagus Absent Congenital displacement Duplication (of) oesophagus
- Q39.9 Congenital malformation of oesophagus, unspecified

Q40 Other congenital malformations of upper alimentary tract

- Q40.0 Congenital hypertrophic pyloric stenosis Congenital or infantile: constriction hypertrophy spasm stenosis stricture of pylorus
- Q40.1 Congenital hiatus hernia Displacement of cardia through oesophageal hiatus Excl.: congenital diaphragmatic hernia (Q79.0)
- Q40.2 Other specified congenital malformations of stomach Congenital: displacement of stomach diverticulum of stomach hourglass stomach Duplication of stomach Megalogastria Microgastria
- Q40.3 Congenital malformation of stomach, unspecified
- Q40.8 Other specified congenital malformations of upper alimentary tract
- Q40.9 Congenital malformation of upper alimentary tract, unspecified Congenital: anomaly deformity NOS of upper alimentary tract

Q41 Congenital absence, atresia and stenosis of small intestine

Incl.: congenital obstruction, occlusion and stricture of small intestine or intestine NOS Excl.: meconium ileus (E84.1)

- Q41.0 Congenital absence, atresia and stenosis of duodenum
- Q41.1 Congenital absence, atresia and stenosis of jejunum Apple peel syndrome Imperforate jejunum
- Q41.2 Congenital absence, atresia and stenosis of ileum

- Q41.8 Congenital absence, atresia and stenosis of other specified parts of small intestine
- Q41.9 Congenital absence, atresia and stenosis of small intestine, part unspecified Congenital absence, atresia and stenosis of intestine NOS

Q42 Congenital absence, atresia and stenosis of large intestine

Incl.: congenital obstruction, occlusion and stricture of large intestine

- Q42.0 Congenital absence, atresia and stenosis of rectum with fistula
- Q42.1 Congenital absence, atresia and stenosis of rectum without fistula Imperforate rectum
- Q42.2 Congenital absence, atresia and stenosis of anus with fistula
- Q42.3 Congenital absence, atresia and stenosis of anus without fistula Imperforate anus
- Q42.8 Congenital absence, atresia and stenosis of other parts of large intestine
- Q42.9 Congenital absence, atresia and stenosis of large intestine, part unspecified

Q43 Other congenital malformations of intestine

- Q43.0 Meckel's diverticulum Persistent: omphalomesenteric duct vitelline duct
- Q43.1 Hirschsprung's disease Aganglionosis Congenital (aganglionic) megacolon
- Q43.2 Other congenital functional disorders of colon Congenital dilatation of colon
- Q43.3 Congenital malformations of intestinal fixation Congenital adhesions [bands]: omental, anomalous peritoneal Jackson's membrane Malrotation of colon Rotation: failure of incomplete insufficient of caecum and colon Universal mesentery
- Q43.4 Duplication of intestine
- Q43.5 Ectopic anus
- Q43.6 Congenital fistula of rectum and anus Excl.: congenital fistula: rectovaginal (Q52.2) urethrorectal (Q64.7) pilonidal fistula or sinus (L05.-) with absence, atresia and stenosis (Q42.0,Q42.2)
- Q43.7 Persistent cloaca Cloaca NOS
- Q43.8 Other specified congenital malformations of intestine Congenital: blind loop syndrome diverticulitis, colon diverticulum, intestine Dolichocolon Megaloappendix Megaloduodenum Microcolon Transposition of: appendix colon intestine
- Q43.9 Congenital malformation of intestine, unspecified

Q44 Congenital malformations of gallbladder, bile ducts and liver

- Q44.0 Agenesis, aplasia and hypoplasia of gallbladder Congenital absence of gallbladder
- Q44.1 Other congenital malformations of gallbladder Congenital malformation of gallbladder NOS Intrahepatic gallbladder
- Q44.2 Atresia of bile ducts

- Q44.3 Congenital stenosis and stricture of bile ducts
- Q44.4 Choledochal cyst
- Q44.5 Other congenital malformations of bile ducts Accessory hepatic duct Congenital malformation of bile duct NOS Duplication: biliary duct cystic duct
- Q44.6 Cystic disease of liver Fibrocystic disease of liver
- Q44.7 Other congenital malformations of liver Accessory liver Alagille's syndrome Congenital: absence of liver hepatomegaly malformation of liver NOS

Q45 Other congenital malformations of digestive system

Excl.: congenital: diaphragmatic hernia (Q79.0) hiatus hernia (Q40.1)

- Q45.0 Agenesis, aplasia and hypoplasia of pancreas Congenital absence of pancreas
- Q45.1 Annular pancreas
- Q45.2 Congenital pancreatic cyst
- Q45.3 Other congenital malformations of pancreas and pancreatic duct Accessory pancreas Congenital malformation of pancreas or pancreatic duct NOS Excl.: diabetes mellitus: congenital (E10.-) neonatal (P70.2) fibrocystic disease of pancreas (E84.-)
- Q45.8 Other specified congenital malformations of digestive system Absence (complete)(partial) of alimentary tract NOS Duplication Malposition, congenital of digestive organs NOS
- Q45.9 Congenital malformation of digestive system, unspecified Congenital: anomaly deformity NOS of digestive system

Links: Gastrointestinal Abnormalities

Congenital malformations of genital organs (Q50-Q56)

Excl.: androgen resistance syndrome (E34.5) syndromes associated with anomalies in the number and form of chromosomes (90-99) testicular feminization syndrome (E34.5)

Links: Genital Abnormalities | Genital Development

Q50 Congenital malformations of ovaries, fallopian tubes and broad ligaments

- Q50.0 Congenital absence of ovary Excl.: Turner's syndrome (96.-)
- Q50.1 Developmental ovarian cyst
- Q50.2 Congenital torsion of ovary
- Q50.3 Other congenital malformations of ovary Accessory ovary Congenital malformation of ovary NOS Ovarian streak
- Q50.4 Embryonic cyst of fallopian tube Fimbrial cyst
- Q50.5 Embryonic cyst of broad ligament Cyst: epoophoron Gartner's duct parovarian
- Q50.6 Other congenital malformations of fallopian tube and broad ligament Absence Accessory Atresia (of) fallopian tube or broad ligament Congenital malformation of fallopian tube or broad ligament NOS

Q51 Congenital malformations of uterus and cervix

- Q51.0 Agenesis and aplasia of uterus Congenital absence of uterus
- Q51.1 Doubling of uterus with doubling of cervix and vagina
- Q51.2 Other doubling of uterus Doubling of uterus NOS
- Q51.3 Bicornate uterus
- Q51.4 Unicornate uterus
- Q51.5 Agenesis and aplasia of cervix Congenital absence of cervix
- Q51.6 Embryonic cyst of cervix
- Q51.7 Congenital fistulae between uterus and digestive and urinary tracts
- Q51.8 Other congenital malformations of uterus and cervix Hypoplasia of uterus and cervix
- Q51.9 Congenital malformation of uterus and cervix, unspecified

Q52 Other congenital malformations of female genitalia

- Q52.0 Congenital absence of vagina
- Q52.1 Doubling of vagina Septate vagina Excl.: doubling of vagina with doubling of uterus and cervix (51.1)
- Q52.2 Congenital rectovaginal fistula Excl.: cloaca (43.7)
- Q52.3 Imperforate hymen
- Q52.4 Other congenital malformations of vagina Congenital malformation of vagina NOS Cyst: canal of Nuck, congenital embryonic vaginal
- Q52.5 Fusion of labia
- Q52.6 Congenital malformation of clitoris
- Q52.7 Other congenital malformations of vulva Congenital: absence cyst malformation NOS of vulva
- Q52.8 Other specified congenital malformations of female genitalia
- Q52.9 Congenital malformation of female genitalia, unspecified

Q53 Undescended testicle

- Q53.0 Ectopic testis Unilateral or bilateral ectopic testes
- Q53.1 Undescended testicle, unilateral
- Q53.2 Undescended testicle, bilateral
- Q53.9 Undescended testicle, unspecified Cryptorchism NOS

Q54 Hypospadias

Excl.: epispadias (64.0)

- Q54.0 Hypospadias, balanic Hypospadias: coronal glandular
- Q54.1 Hypospadias, penile
- Q54.2 Hypospadias, penoscrotal
- Q54.3 Hypospadias, perineal
- Q54.4 Congenital chordee
- Q54.8 Other hypospadias
- Q54.9 Hypospadias, unspecified

Q55 Other congenital malformations of male genital organs

Excl.: congenital hydrocele (P83.5) hypospadias (54.-)

- Q55.0 Absence and aplasia of testis Monorchism
- Q55.1 Hypoplasia of testis and scrotum Fusion of testes

- Q55.2 Other congenital malformations of testis and scrotum Congenital malformation of testis or scrotum NOS Polyorchism Retractile testis Testis migrans
- Q55.3 Atresia of vas deferens
- Q55.4 Other congenital malformations of vas deferens, epididymis, seminal vesicles and prostate Absence or aplasia of: prostate spermatic cord Congenital malformation of vas deferens, epididymis, seminal vesicles or prostate NOS
- Q55.5 Congenital absence and aplasia of penis
- Q55.6 Other congenital malformations of penis Congenital malformation of penis NOS Curvature of penis (lateral) Hypoplasia of penis
- Q55.8 Other specified congenital malformations of male genital organs
- Q55.9 Congenital malformation of male genital organ, unspecified Congenital: anomaly deformity NOS of male genital organ

Q56 Indeterminate sex and pseudohermaphroditism

Excl.: pseudohermaphroditism: female, with adrenocortical disorder (E25.-) male, with androgen resistance (E34.5) with specified chromosomal anomaly (96-99)

- Q56.0 Hermaphroditism, not elsewhere classified Ovotestis
- Q56.1 Male pseudohermaphroditism, not elsewhere classified Male pseudohermaphroditism NOS
- Q56.2 Female pseudohermaphroditism, not elsewhere classified Female pseudohermaphroditism NOS
- Q56.3 Pseudohermaphroditism, unspecified
- Q56.4 Indeterminate sex, unspecified Ambiguous genitalia

Links: Genital Abnormalities

Congenital malformations of the urinary system (Q60-Q64)

Links: Renal Abnormalities | Renal Development

Q60 Renal agenesis and other reduction defects of kidney

Incl.: atrophy of kidney: congenital infantile congenital absence of kidney

- Q60.0 Renal agenesis, unilateral
- Q60.1 Renal agenesis, bilateral
- Q60.2 Renal agenesis, unspecified
- Q60.3 Renal hypoplasia, unilateral
- Q60.4 Renal hypoplasia, bilateral
- Q60.5 Renal hypoplasia, unspecified
- Q60.6 Potter's syndrome

Q61 Cystic kidney disease

Excl.: acquired cyst of kidney (N28.1) Potter's syndrome (60.6)

- Q61.0 Congenital single renal cyst Cyst of kidney (congenital)(single)
- Q61.1 Polycystic kidney, autosomal recessive Polycystic kidney, infantile type
- Q61.2 Polycystic kidney, autosomal dominant Polycystic kidney, adult type

- Q61.3 Polycystic kidney, unspecified
- Q61.4 Renal dysplasia Multicystic: dyplastic kidney kidney (developmental) kidney disease renal dysplasia Excl.: polycystic kidney disease (61.1-61.3)
- Q61.5 Medullary cystic kidney Sponge kidney NOS
- Q61.8 Other cystic kidney diseases Fibrocystic: kidney renal degeneration or disease
- Q61.9 Cystic kidney disease, unspecified Meckel-Gruber syndrome

Q62 Congenital obstructive defects of renal pelvis and congenital malformations of ureter

- Q62.0 Congenital hydronephrosis
- Q62.1 Atresia and stenosis of ureter Congenital occlusion of: ureter ureteropelvic junction ureterovesical orifice Impervious ureter
- Q62.2 Congenital megaloureter Congenital dilatation of ureter
- Q62.3 Other obstructive defects of renal pelvis and ureter Congenital ureterocele
- Q62.4 Agenesis of ureter Absent ureter
- Q62.5 Duplication of ureter Accessory Double ureter
- Q62.6 Malposition of ureter Deviation Displacement Ectopic Implantation, anomalous (of) ureter or ureteric orifice
- Q62.7 Congenital vesico-uretero-renal reflux
- Q62.8 Other congenital malformations of ureter Anomaly of ureter NOS

Q63 Other congenital malformations of kidney

Excl.: congenital nephrotic syndrome (N04.-)

- Q63.0 Accessory kidney
- Q63.1 Lobulated, fused and horseshoe kidney
- Q63.2 Ectopic kidney Congenital displaced kidney Malrotation of kidney
- Q63.3 Hyperplastic and giant kidney
- Q63.8 Other specified congenital malformations of kidney Congenital renal calculi
- Q63.9 Congenital malformation of kidney, unspecified

Q64 Other congenital malformations of urinary system

- Q64.0 Epispadias Excl.: hypospadias (54.-)
- Q64.1 Exstrophy of urinary bladder Ectopia vesicae Extroversion of bladder
- Q64.2 Congenital posterior urethral valves
- Q64.3 Other atresia and stenosis of urethra and bladder neck Congenital: bladder neck obstruction stricture of: urethra urinary meatus vesicourethral orifice Impervious urethra
- Q64.4 Malformation of urachus Cyst of urachus Patent urachus Prolapse of urachus
- Q64.5 Congenital absence of bladder and urethra
- Q64.6 Congenital diverticulum of bladder
- Q64.7 Other congenital malformations of bladder and urethra Accessory: bladder urethra Congenital: hernia of bladder malformation of bladder or

urethra NOS prolapse of: bladder (mucosa) urethra urinary meatus urethrorectal fistula Double: urethra urinary meatus

- Q64.8 Other specified congenital malformations of urinary system
- Q64.9 Congenital malformation of urinary system, unspecified Congenital: anomaly deformity NOS of urinary system

Links: Renal Abnormalities | Renal Development

Congenital malformations and deformations of the musculoskeletal system (Q65-Q79)

Links: Musculoskeletal Abnormalities | Limb Abnormalities

Q65 Congenital deformities of hip

Excl.: clicking hip (R29.4)

- Q65.0 Congenital dislocation of hip, unilateral
- Q65.1 Congenital dislocation of hip, bilateral
- Q65.2 Congenital dislocation of hip, unspecified
- Q65.3 Congenital subluxation of hip, unilateral
- Q65.4 Congenital subluxation of hip, bilateral
- Q65.5 Congenital subluxation of hip, unspecified
- Q65.6 Unstable hip Dislocatable hip Subluxatable hip
- Q65.8 Other congenital deformities of hip Anteversion of femoral neck Congenital acetabular dysplasia Congenital coxa: valga vara
- Q65.9 Congenital deformity of hip, unspecified

Q66 Congenital deformities of feet

Excl.: reduction defects of feet (Q72.-) valgus deformities (acquired) (M21.0) varus deformities (acquired) (M21.1)

- Q66.0 Talipes equinovarus
- Q66.1 Talipes calcaneovarus
- Q66.2 Metatarsus varus
- Q66.3 Other congenital varus deformities of feet Hallux varus, congenital
- Q66.4 Talipes calcaneovalgus
- Q66.5 Congenital pes planus Flat foot: congenital rigid spastic (everted)
- Q66.6 Other congenital valgus deformities of feet Metatarsus valgus
- Q66.7 Pes cavus
- Q66.8 Other congenital deformities of feet Clubfoot NOS Hammer toe, congenital Talipes: NOS asymmetric Tarsal coalition Vertical talus
- Q66.9 Congenital deformity of feet, unspecified

Q67 Congenital musculoskeletal deformities of head, face, spine and chest

Excl.: congenital malformation syndromes classified to Q87.- Potter's syndrome (Q60.6)

- Q67.0 Facial asymmetry
- Q67.1 Compression facies
- Q67.2 Dolichocephaly
- Q67.3 Plagiocephaly
- Q67.4 Other congenital deformities of skull, face and jaw Depressions in skull Deviation of nasal septum, congenital Hemifacial atrophy or

hypertrophy Squashed or bent nose, congenital Excl.: dentofacial anomalies [including malocclusion] (K07.-) syphilitic saddle nose (A50.5)

- Q67.5 Congenital deformity of spine Congenital scoliosis: NOS postural Excl.: infantile idiopathic scoliosis (M41.0) scoliosis due to congenital bony malformation (Q76.3)
- Q67.6 Pectus excavatum Congenital funnel chest
- Q67.7 Pectus carinatum Congenital pigeon chest
- Q67.8 Other congenital deformities of chest Congenital deformity of chest wall NOS

Q68 Other congenital musculoskeletal deformities

Excl.: reduction defects of limb(s) (Q71-Q73)

- Q68.0 Congenital deformity of sternocleidomastoid muscle Congenital (sternomastoid) torticollis Contracture of sternocleidomastoid (muscle) Sternomastoid tumour (congenital)
- Q68.1 Congenital deformity of hand Congenital clubfinger Spade-like hand (congenital)
- Q68.2 Congenital deformity of knee Congenital: dislocation of knee genu recurvatum
- Q68.3 Congenital bowing of femur Excl.: anteversion of femur (neck) (Q65.8)
- Q68.4 Congenital bowing of tibia and fibula
- Q68.5 Congenital bowing of long bones of leg, unspecified
- Q68.8 Other specified congenital musculoskeletal deformities Congenital: deformity of: clavicle elbow forearm scapula dislocation of: elbow shoulder

Q69 Polydactyly

- Q69.0 Accessory finger(s)
- Q69.1 Accessory thumb(s)
- Q69.2 Accessory toe(s) Accessory hallux
- Q69.9 Polydactyly, unspecified Supernumerary digit(s) NOS

Q70 Syndactyly

- Q70.0 Fused fingers Complex syndactyly of fingers with synostosis
- Q70.1 Webbed fingers Simple syndactyly of fingers without synostosis
- Q70.2 Fused toes Complex syndactyly of toes with synostosis
- Q70.3 Webbed toes Simple syndactyly of toes without synostosis
- Q70.4 Polysyndactyly
- Q70.9 Syndactyly, unspecified Symphalangy NOS

Q71 Reduction defects of upper limb

- Q71.0 Congenital complete absence of upper limb(s)
- Q71.1 Congenital absence of upper arm and forearm with hand present
- Q71.2 Congenital absence of both forearm and hand
- Q71.3 Congenital absence of hand and finger(s)
- Q71.4 Longitudinal reduction defect of radius Clubhand (congenital) Radial clubhand
- Q71.5 Longitudinal reduction defect of ulna

- Q71.6 Lobster-claw hand
- Q71.8 Other reduction defects of upper limb(s) Congenital shortening of upper limb(s)
- Q71.9 Reduction defect of upper limb, unspecified

Q72 Reduction defects of lower limb

- Q72.0 Congenital complete absence of lower limb(s)
- Q72.1 Congenital absence of thigh and lower leg with foot present
- Q72.2 Congenital absence of both lower leg and foot
- Q72.3 Congenital absence of foot and toe(s)
- Q72.4 Longitudinal reduction defect of femur Proximal femoral focal deficiency
- Q72.5 Longitudinal reduction defect of tibia
- Q72.6 Longitudinal reduction defect of fibula
- Q72.7 Split foot
- Q72.8 Other reduction defects of lower limb(s) Congenital shortening of lower limb(s)
- Q72.9 Reduction defect of lower limb, unspecified

Q73 Reduction defects of unspecified limb

- Q73.0 Congenital absence of unspecified limb(s) Amelia NOS
- Q73.1 Phocomelia, unspecified limb(s) Phocomelia NOS
- Q73.8 Other reduction defects of unspecified limb(s) Longitudinal reduction deformity of unspecified limb(s) Ectromelia NOS Hemimelia NOS Reduction defect of limb(s) NOS

Q74 Other congenital malformations of limb(s)

Excl.: polydactyly (Q69.-) reduction defect of limb (Q71-Q73) syndactyly (Q70.-)

- Q74.0 Other congenital malformations of upper limb(s), including shoulder girdle Accessory carpal bones Cleidocranial dysostosis Congenital pseudarthrosis of clavicle Macrodactylia (fingers) Madelung's deformity Radioulnar synostosis Sprengel's deformity Triphalangeal thumb
- Q74.1 Congenital malformation of knee Congenital: absence of patella dislocation of patella genu: valgum varum Rudimentary patella Excl.: congenital: dislocation of knee (Q68.2) genu recurvatum (Q68.2) nail patella syndrome (Q87.2)
- Q74.2 Other congenital malformations of lower limb(s), including pelvic girdle Congenital: fusion of sacroiliac joint malformation (of): ankle (joint) sacroiliac (joint) Excl.: anteversion of femur (neck) (Q65.8)
- Q74.3 Arthrogryposis multiplex congenita
- Q74.8 Other specified congenital malformations of limb(s)
- Q74.9 Unspecified congenital malformation of limb(s) Congenital anomaly of limb(s) NOS
-

Q75 Other congenital malformations of skull and face bones

Excl.: congenital malformation of face NOS (Q18.-) congenital malformation syndromes classified to Q87.- dentofacial anomalies

[including malocclusion] (K07.-) musculoskeletal deformities of head and face (Q67.0-Q67.4) skull defects associated with congenital anomalies of brain such as: anencephaly (Q00.0) encephalocele (Q01.-) hydrocephalus (Q03.-) microcephaly (Q02)

- Q75.0 Craniosynostosis Acrocephaly Imperfect fusion of skull Oxycephaly Trigonocephaly
- Q75.1 Craniofacial dysostosis Crouzon's disease
- Q75.2 Hypertelorism
- Q75.3 Macrocephaly
- Q75.4 Mandibulofacial dysostosis Syndrome: Franceschetti Treacher-Collins
- Q75.5 Oculomandibular dysostosis
- Q75.8 Other specified congenital malformations of skull and face bones Absence of skull bone, congenital Congenital deformity of forehead Platybasia
- Q75.9 Congenital malformation of skull and face bones, unspecified Congenital anomaly of: face bones NOS skull NOS

Q76 Congenital malformations of spine and bony thorax

Excl.: congenital musculoskeletal deformities of spine and chest (Q67.5-Q67.8)

- Q76.0 Spina bifida occulta Excl.: meningocele (spinal) (Q05.-) spina bifida (aperta)(cystica) (Q05.-)
- Q76.1 Klippel-Feil syndrome Cervical fusion syndrome
- Q76.2 Congenital spondylolisthesis Congenital spondylolysis Excl.: spondylolisthesis (acquired) (M43.1) spondylolysis (acquired) (M43.0)
- Q76.3 Congenital scoliosis due to congenital bony malformation Hemivertebra fusion or failure of segmentation with scoliosis
- Q76.4 Other congenital malformations of spine, not associated with scoliosis Congenital: absence of vertebra fusion of spine kyphosis lordosis malformation of lumbosacral (joint) (region) Hemivertebra Malformation of spine Platyspondylisis Supernumerary vertebra unspecified or not associated with scoliosis
- Q76.5 Cervical rib Supernumerary rib in cervical region
- Q76.6 Other congenital malformations of ribs Accessory rib Congenital: absence of rib fusion of ribs malformation of ribs NOS Excl.: short rib syndrome (Q77.2)
- Q76.7 Congenital malformation of sternum Congenital absence of sternum Sternum bifidum
- Q76.8 Other congenital malformations of bony thorax
- Q76.9 Congenital malformation of bony thorax, unspecified

Q77 Osteochondrodysplasia with defects of growth of tubular bones and spine

Excl.: mucopolysaccharidosis (E76.0-E76.3)

- Q77.0 Achondrogenesis Hypochondrogenesis
- Q77.1 Thanatophoric short stature
- Q77.2 Short rib syndrome Asphyxiating thoracic dysplasia [Jeune]

- Q77.3 Chondrodysplasia punctata
- Q77.4 Achondroplasia Hypochondroplasia Osteosclerosis congenita
- Q77.5 Dystrophic dysplasia
- Q77.6 Chondroectodermal dysplasia Ellis-van Creveld syndrome
- Q77.7 Spondyloepiphyseal dysplasia
- Q77.8 Other osteochondrodysplasia with defects of growth of tubular bones and spine
- Q77.9 Osteochondrodysplasia with defects of growth of tubular bones and spine, unspecified

Q78 Other osteochondrodysplasias

- Q78.0 Osteogenesis imperfecta Fragilitas ossium Osteopsathyrosis
- Q78.1 Polyostotic fibrous dysplasia Albright(-McCune)(-Sternberg) syndrome
- Q78.2 Osteopetrosis Albers-Schönberg syndrome
- Q78.3 Progressive diaphyseal dysplasia Camurati-Engelmann syndrome
- Q78.4 Enchondromatosis Maffucci's syndrome Ollier's disease
- Q78.5 Metaphyseal dysplasia Pyle's syndrome
- Q78.6 Multiple congenital exostoses Diaphyseal aclasis
- Q78.8 Other specified osteochondrodysplasias Osteopoikilosis
- Q78.9 Osteochondrodysplasia, unspecified Chondrodystrophy NOS Osteodystrophy NOS

Q79 Congenital malformations of the musculoskeletal system, not elsewhere classified

Excl.: congenital (sternomastoid) torticollis (Q68.0)

- Q79.0 Congenital diaphragmatic hernia Excl.: congenital hiatus hernia (Q40.1)
- Q79.1 Other congenital malformations of diaphragm Absence of diaphragm Congenital malformation of diaphragm NOS Eventration of diaphragm
- Q79.2 Exomphalos Omphalocele Excl.: umbilical hernia (K42.-)
- Q79.3 Gastroschisis
- Q79.4 Prune belly syndrome
- Q79.5 Other congenital malformations of abdominal wall Excl.: umbilical hernia (K42.-)
- Q79.6 Ehlers-Danlos syndrome
- Q79.8 Other congenital malformations of musculoskeletal system Absence of: muscle tendon Accessory muscle Amyotrophia congenita Congenital: constricting bands shortening of tendon Poland's syndrome
- Q79.9 Congenital malformation of musculoskeletal system, unspecified Congenital: anomaly NOS deformity NOS of musculoskeletal system NOS

Links: Musculoskeletal Abnormalities | Limb Abnormalities

Other congenital malformations (Q80-Q89)

Links: Integumentary Abnormalities

Q80 Congenital ichthyosis

Excl.: Refsum's disease (G60.1)

- Q80.0 Ichthyosis vulgaris
- Q80.1 X-linked ichthyosis
- Q80.2 Lamellar ichthyosis Collodion baby
- Q80.3 Congenital bullous ichthyosiform erythroderma
- Q80.4 Harlequin fetus
- Q80.8 Other congenital ichthyosis
- Q80.9 Congenital ichthyosis, unspecified

Q81 Epidermolysis bullosa

- Q81.0 Epidermolysis bullosa simplex Excl.: Cockayne's syndrome (Q87.1)
- Q81.1 Epidermolysis bullosa letalis Herlitz' syndrome
- Q81.2 Epidermolysis bullosa dystrophica
- Q81.8 Other epidermolysis bullosa
- Q81.9 Epidermolysis bullosa, unspecified
- Q82 Other congenital malformations of skin Excl.: acrodermatitis enteropathica (E83.2) congenital erythropoietic porphyria (E80.0) pilonidal cyst or sinus (L05.-) Sturge-Weber(-Dimitri) syndrome (Q85.8)
- Q82.0 Hereditary lymphoedema
- Q82.1 Xeroderma pigmentosum
- Q82.2 Mastocytosis Urticaria pigmentosa Excl.: malignant mastocytosis (C96.2)
- Q82.3 Incontinentia pigmenti
- Q82.4 Ectodermal dysplasia (anhidrotic) Excl.: Ellis-van Creveld syndrome (Q77.6)
- Q82.5 Congenital non-neoplastic naevus Birthmark NOS Naevus: flammeus portwine sanguineous strawberry vascular NOS verrucous Excl.: café au lait spots (L81.3) lentigo (L81.4) naevus: NOS (D22.-) araneus (I78.1) melanocytic (D22.-) pigmented (D22.-) spider (I78.1) stellar (I78.1)
- Q82.8 Other specified congenital malformations of skin Abnormal palmar creases Accessory skin tags Benign familial pemphigus [Hailey-Hailey] Cutis laxa (hyperelastica) Dermatoglyphic anomalies Inherited keratosis palmaris et plantaris Keratosis follicularis [Darier-White] Excl.: Ehlers-Danlos syndrome (Q79.6)
- Q82.9 Congenital malformation of skin, unspecified

Q83 Congenital malformations of breast

Excl.: absence of pectoral muscle (Q79.8)

- Q83.0 Congenital absence of breast with absent nipple
- Q83.1 Accessory breast Supernumerary breast
- Q83.2 Absent nipple
- Q83.3 Accessory nipple Supernumerary nipple
- Q83.8 Other congenital malformations of breast Hypoplasia of breast
- Q83.9 Congenital malformation of breast, unspecified

Q84 Other congenital malformations of integument

- Q84.0 Congenital alopecia Congenital atrichosis

- Q84.1 Congenital morphological disturbances of hair, not elsewhere classified Beaded hair Monilethrix Pili annulati Excl.: Menkes' kinky hair syndrome (E83.0)
- Q84.2 Other congenital malformations of hair Congenital: hypertrichosis malformation of hair NOS Persistent lanugo
- Q84.3 Anonychia Excl.: nail patella syndrome (Q87.2)
- Q84.4 Congenital leukonychia
- Q84.5 Enlarged and hypertrophic nails Congenital onychauxis Pachyonychia
- Q84.6 Other congenital malformations of nails Congenital: clubnail koilonychia malformation of nail NOS
- Q84.8 Other specified congenital malformations of integument Aplasia cutis congenita
- Q84.9 Congenital malformation of integument, unspecified Congenital: anomaly NOS deformity NOS of integument NOS

Q85 Phakomatoses, not elsewhere classified

Excl.: ataxia telangiectasia [Louis-Bar] (G11.3) familial dysautonomia [Riley-Day] (G90.1)

- Q85.0 Neurofibromatosis (nonmalignant) Von Recklinghausen's disease
- Q85.1 Tuberous sclerosis Bourneville's disease Epiloia
- Q85.8 Other phakomatoses, not elsewhere classified Syndrome: Peutz-Jeghers Sturge-Weber(-Dimitri) von Hippel-Lindau Excl.: Meckel-Gruber syndrome (Q61.9)
- Q85.9 Phakomatosis, unspecified Hamartosis NOS

Q86 Congenital malformation syndromes due to known exogenous causes, not elsewhere classified

Excl.: iodine-deficiency-related hypothyroidism (E00-E02) nonteratogenic effects of substances transmitted via placenta or breast milk (P04.-)

- Q86.0 Fetal alcohol syndrome (dysmorphic)
- Q86.1 Fetal hydantoin syndrome Meadow's syndrome
- Q86.2 Dysmorphism due to warfarin
- Q86.8 Other congenital malformation syndromes due to known exogenous causes

Q87 Other specified congenital malformation syndromes affecting multiple systems

- Q87.0 Congenital malformation syndromes predominantly affecting facial appearance Acrocephalopolysyndactyly Acrocephalosyndactyly [Apert] Cryptophthalmos syndrome Cyclopia Syndrome: Goldenhar Moebius oro-facial-digital Robin Whistling face
- Q87.1 Congenital malformation syndromes predominantly associated with short stature Syndrome: Aarskog Cockayne De Lange Dubowitz Noonan Prader-Willi Robinow-Silverman-Smith Russell-Silver Seckel Smith-Lemli-Opitz Excl.: Ellis-van Creveld syndrome (Q77.6)
- Q87.2 Congenital malformation syndromes predominantly involving limbs Syndrome: Holt-Oram Klippel-Trénaunay-Weber nail patella

Rubinstein-Taybi sirenomelia thrombocytopenia with absent radius [TAR] VATER

- Q87.3 Congenital malformation syndromes involving early overgrowth Syndrome: Beckwith-Wiedemann Sotos Weaver
- Q87.4 Marfan's syndrome
- Q87.5 Other congenital malformation syndromes with other skeletal changes
- Q87.8 Other specified congenital malformation syndromes, not elsewhere classified Syndrome: Alport Laurence-Moon(-Bardet)-Biedl Zellweger

Q89 Other congenital malformations, not elsewhere classified

- Q89.0 Congenital malformations of spleen Asplenia (congenital) Congenital splenomegaly Excl.: isomerism of atrial appendages (with asplenia or polysplenia) (Q20.6)
- Q89.1 Congenital malformations of adrenal gland Excl.: congenital adrenal hyperplasia (E25.0)
- Q89.2 Congenital malformations of other endocrine glands Congenital malformation of parathyroid or thyroid gland Persistent thyroglossal duct Thyroglossal cyst
- Q89.3 Situs inversus Dextrocardia with situs inversus Mirror-image atrial arrangement with situs inversus Situs inversus or transversus: abdominalis thoracis Transposition of viscera: abdominal thoracic Excl.: dextrocardia NOS (Q24.0) laevocardia (Q24.1)
- Q89.4 Conjoined twins Craniopagus Dicephaly Double monster Pygopagus Thoracopagus
- Q89.7 Multiple congenital malformations, not elsewhere classified Monster NOS Multiple congenital: anomalies NOS deformities NOS Excl.: congenital malformation syndromes affecting multiple systems (Q87.-)
- Q89.8 Other specified congenital malformations
- Q89.9 Congenital malformation, unspecified Congenital: anomaly NOS deformity NOS

Links: Integumentary Abnormalities

Chromosomal abnormalities, not elsewhere classified (Q90-Q99)

Links: Genetic Abnormalities | Trisomy 21 | Trisomy 18 | Trisomy 13 | Philadelphia chromosome | Disorders of Sex Development

Q90 Down's syndrome

- Q90.0 Trisomy 21, meiotic nondisjunction
- Q90.1 Trisomy 21, mosaicism (mitotic nondisjunction)
- Q90.2 Trisomy 21, translocation
- Q90.9 Down's syndrome, unspecified Trisomy 21 NOS

Q91 Edwards' syndrome and Patau's syndrome

- Q91.0 Trisomy 18, meiotic nondisjunction
- Q91.1 Trisomy 18, mosaicism (mitotic nondisjunction)
- Q91.2 Trisomy 18, translocation * Q91.3 Edwards' syndrome, unspecified
- Q91.4 Trisomy 13, meiotic nondisjunction

- Q91.5 Trisomy 13, mosaicism (mitotic nondisjunction)
- Q91.6 Trisomy 13, translocation
- Q91.7 Patau's syndrome, unspecified

Q92 Other trisomies and partial trisomies of the autosomes, not elsewhere classified

Incl.: unbalanced translocations and insertions Excl.: trisomies of chromosomes 13, 18, 21 (Q90-Q91)

- Q92.0 Whole chromosome trisomy, meiotic nondisjunction
- Q92.1 Whole chromosome trisomy, mosaicism (mitotic nondisjunction)
- Q92.2 Major partial trisomy Whole arm or more duplicated.
- Q92.3 Minor partial trisomy Less than whole arm duplicated.
- Q92.4 Duplications seen only at prometaphase
- Q92.5 Duplications with other complex rearrangements
- Q92.6 Extra marker chromosomes
- Q92.7 Triploidy and polyploidy
- Q92.8 Other specified trisomies and partial trisomies of autosomes
- Q92.9 Trisomy and partial trisomy of autosomes, unspecified

Q93 Monosomies and deletions from the autosomes, not elsewhere classified

- Q93.0 Whole chromosome monosomy, meiotic nondisjunction
- Q93.1 Whole chromosome monosomy, mosaicism (mitotic nondisjunction)
- Q93.2 Chromosome replaced with ring or dicentric
- Q93.3 Deletion of short arm of chromosome 4 Wolff-Hirschorn syndrome
- Q93.4 Deletion of short arm of chromosome 5 Cri-du-chat syndrome
- Q93.5 Other deletions of part of a chromosome Angelman syndrome
- Q93.6 Deletions seen only at prometaphase
- Q93.7 Deletions with other complex rearrangements
- Q93.8 Other deletions from the autosomes
- Q93.9 Deletion from autosomes, unspecified

Q95 Balanced rearrangements and structural markers, not elsewhere classified

Incl.: Robertsonian and balanced reciprocal translocations and insertions

- Q95.0 Balanced translocation and insertion in normal individual
- Q95.1 Chromosome inversion in normal individual
- Q95.2 Balanced autosomal rearrangement in abnormal individual
- Q95.3 Balanced sex/autosomal rearrangement in abnormal individual
- Q95.4 Individuals with marker heterochromatin
- Q95.5 Individuals with autosomal fragile site
- Q95.8 Other balanced rearrangements and structural markers
- Q95.9 Balanced rearrangement and structural marker, unspecified

Q96 Turner's syndrome

Excl.: Noonan's syndrome (Q87.1)

- Q96.0 Karyotype 45,X

- Q96.1 Karyotype 46,X iso (XQ)
- Q96.2 Karyotype 46,X with abnormal sex chromosome, except iso (XQ)
- Q96.3 Mosaicism, 45,X/46,XX or XY
- Q96.4 Mosaicism, 45,X/other cell line(s) with abnormal sex chromosome
- Q96.8 Other variants of Turner's syndrome
- Q96.9 Turner's syndrome, unspecified

Q97 Other sex chromosome abnormalities, female phenotype, not elsewhere classified

Excl.: Turner's syndrome (Q96.-)

- Q97.0 Karyotype 47,XXX
- Q97.1 Female with more than three X chromosomes
- Q97.2 Mosaicism, lines with various numbers of X chromosomes
- Q97.3 Female with 46,XY karyotype
- Q97.8 Other specified sex chromosome abnormalities, female phenotype
- Q97.9 Sex chromosome abnormality, female phenotype, unspecified

Q98 Other sex chromosome abnormalities, male phenotype, not elsewhere classified

- Q98.0 Klinefelter's syndrome karyotype 47,XXY
- Q98.1 Klinefelter's syndrome, male with more than two X chromosomes
- Q98.2 Klinefelter's syndrome, male with 46,XX karyotype
- Q98.3 Other male with 46,XX karyotype
- Q98.4 Klinefelter's syndrome, unspecified
- Q98.5 Karyotype 47,XYY
- Q98.6 Male with structurally abnormal sex chromosome
- Q98.7 Male with sex chromosome mosaicism
- Q98.8 Other specified sex chromosome abnormalities, male phenotype
- Q98.9 Sex chromosome abnormality, male phenotype, unspecified

Q99 Other chromosome abnormalities, not elsewhere classified

- Q99.0 Chimera 46,XX/46,XY Chimera 46,XX/46,XY true hermaphrodite
- Q99.1 46,XX true hermaphrodite 46,XX with streak gonads 46,XY with streak gonads Pure gonadal dysgenesis
- Q99.2 Fragile X chromosome Fragile X syndrome
- Q99.8 Other specified chromosome abnormalities
- Q99.9 Chromosomal abnormality, unspecified

Links: Genetic Abnormalities | Trisomy 21 | Trisomy 18 | Trisomy 13 | Philadelphia chromosome | Disorders of Sex Development

Reference World Health Organisation. **International Statistical Classification of Diseases and Related Health Problems**. (1992) 10th Revision (ICD-10). Geneva: WHO ICD-10 - 2016 Online (English)

APPENDIX C

ETHICAL CLEARANCE FROM UNIVERSITY OF CAPE COAST

UNIVERSITY OF CAPE COAST

INSTITUTIONAL REVIEW BOARD SECRETARIAT

TEL: 03321-33172/3 / 0207355653/ 0244207814
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OMB NO: 0990-0279
IORG #: IORG0009096

C/O Directorate of Research, Innovation and Consultancy

8TH FEBRUARY, 2017

Ms Josephine Owusu Takyi
School of Nursing and Midwifery
University of Cape Coast

Dear Ms Takyi,

ETHICAL CLEARANCE –ID :(UCCIRB/CHAS/2016/110)

The University of Cape Coast Institutional Review Board (UCCIRB) has granted **Provisional Approval** for the implementation of your research protocol titled **'Incidence and risk factors of congenital abnormalities in children under five years, a study at the Child Health Department of Korle Bu Teaching Hospital.'**

This approval requires that you submit periodic review of the protocol to the Board and a final full review to the UCCIRB on completion of the research. The UCCIRB may observe or cause to be observed procedures and records of the research during and after implementation.

Please note that any modification of the project must be submitted to the UCCIRB for review and approval before its implementation.

You are also required to report all serious adverse events related to this study to the UCCIRB within seven days verbally and fourteen days in writing.

Always quote the protocol identification number in all future correspondence with us in relation to this protocol.

Yours faithfully,

ADMINISTRATOR
INSTITUTIONAL REVIEW BOARD
UNIVERSITY OF CAPE COAST
Date:.....

Samuel Asiedu Owusu
Administrator

APPENDIX D

INTRODUCTORY LETTER TO KORLE BU TEACHING HOSPITAL

UNIVERSITY OF CAPE COAST
COLLEGE OF HEALTH AND ALLIED SCIENCES
SCHOOL OF NURSING AND MIDWIFERY
DEAN'S OFFICE

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UNIVERSITY POST OFFICE
CAPE COAST, GHANA.

Our Ref:
Your Ref: SNM/R/2/161

25th January, 2017

The Director
Korle-Bu Teaching Hospital
P. O. Box 77
Accra

Dear Sir,

INTRODUCTORY LETTER: JOSEPHINE OWUSU-TAKYI

The above named is a Level 850 Post Graduate Student at the School of Nursing and Midwifery, University of Cape Coast.

As Part of the school's requirement for graduation, she has to do a research and present a report on it. She intends to collect data from the Korle-Bu Teaching Hospital as her research topic depicts: **Incidence and Risk Factors of Congenital Abnormalities in Hospital Children Under 5 Years**

We would be grateful if you could accord her any assistance she may require from you to enable her collect her data successfully.

Counting on your usual cooperation.

Thank you

Yours faithfully,

Dr. Samuel Victor Nuvor
Vice-Dean

APPENDIX F

INFORMED CONSENT FORM

Title: [*MATERNAL AND PERINATAL CHARACTERISTICS OF CHILDREN BELOW FIVE YEARS WITH CONGENITAL ABNORMALITIES SEEN AT THE CHILD HEALTH DEPARTMENT, KORLE BU TEACHING HOSPITAL]*

Principal Investigator: [JOSEPHINE OWUSU-TAKYI]
Address: [UNIVERSITY OF CAPE COAST, SCHOOL OF NURSING, DEPARTMENT OF NURSING AND ALLIED HEALTH SCIENCE]

General Information about Research

The general purpose of the study will be to investigate the maternal and perinatal contributing factors of children below five years with congenital abnormalities seen at the child health department, Korle Bu Teaching Hospital.

Specific Objectives

4. To identify the types of congenital abnormalities seen at the korle bu teaching hospital
5. To find the maternal and perinatal characteristics that has contributed to congenital abnormalities in children below five years.
6. Assess the possible suggestions to minimize the risk factors of congenital abnormalities in children below five years

Significance of the Study

The findings of the study will help to create awareness of the maternal and perinatal risk factors of congenital abnormalities to reduce childhood mortalities and morbidities. The findings will also help the ministry of health and policy makers to devise strategies to reduce the factors and also strengthen existing ones. The outcome of the findings will give directions for further research studies on congenital abnormalities and also contribute to the body of knowledge on maternal and child health in general.

Procedures

To find answers to some of these questions, we invite you to take part in this research project. If you accept, you will be required to fill out a survey which will be provided by Josephine Owusu-Takyi and collected by Josephine Owusu-Takyi.

You are being invited to take part in this research because we feel that your experience as a mother caring for a child with congenital abnormalities can contribute much to this research by providing the researcher with information that can be used to minimize the incidence of

congenital abnormalities in the near future. Yow will be required to answer questions about your lifestyle before and during the pregnancy, details about your child including the type of congenital abnormalities and other comorbidities as well as the mode of delivery and antenatal care.

If you do not wish to answer any of the questions posed during the interview, you may say so and the interviewer will move on to the next question. The interview will take place on the ward, and no one else but the interviewer will be present. The information recorded is considered confidential, and no one else except the researcher will have access to the information documented during your interview.

If you do not wish to answer any of the questions included in the survey, you may skip them and move on to the next question. The questionnaire would be distributed and collected by the researcher. The information recorded is considered confidential, and no one else except the researcher will have access to your survey.

The expected duration of the interview will be 30 minutes.

Possible Risks and Discomforts

The possible risks and discomfort include answering questions about the child's delivery process and pregnancy which may be remind the mothers of maybe a painful delivery and also questions about the child's condition which they may like to keep confidential because of social misconception and discrimination.

Possible Benefits

The participant engagement in the research will help to provide vital information about the particular congenital abnormality and help us to devise and implement possible strategies to manage and prevent further incidence of congenital abnormalities in the near future.

Alternatives to Participation: NOT APPLICABLE

Confidentiality

All information obtained from the patients' medical records and mothers with children with congenital abnormalities will be held confidential and be used only for the purpose of academic purposes and also to improve health care. "We will protect information about you to the best of our ability. You will not be named in any reports.

Compensation

Because it is an academic research study, no compensation would be given to the participants.

Additional Cost: NOT APPLICABLE

Staying in the Research: NOT APPLICABLE

Voluntary Participation and Right to Leave the Research

You have the right to leave at any point in the research without the care of your child being affected.

Termination of Participation by the Researcher: NOT APPLICABLE

Notification of Significant New Findings: NOT APPLICABLE

Contacts for Additional Information

In case of any injury or abuse during the research, please do not hesitate to call my principal supervisor

Dr. S. V. NUVOR
Contact number: 0205853850
Senior Lecturer
University of Cape Coast Medical School

Your rights as a Participant

This research has been reviewed and approved by the Institutional Review Board of University of Cape Coast (UCCIRB). If you have any questions about your rights as a research participant you can contact the Administrator at the IRB Office between the hours of 8:00 am and 4:30 p.m. through the phones lines 0332133172 and 0244207814 or email address: irb@ucc.edu.gh.

VOLUNTEER AGREEMENT

The above document describing the benefits, risks and procedures for the research title **(**MATERNAL AND PERINATAL CONTRIBUTING FACTORS OF CONGENITAL ABNORMALITIES IN CHILDREN BELOW FIVE YEARS A STUDY AT THE KORLE BU TEACHING HOSPITAL**)** has been read and explained to me. I have been given an opportunity to have any questions about the research answered to my satisfaction. I agree to participate as a volunteer.

_____ _____

Date Name and signature or mark of volunteer

If volunteers cannot read the form themselves, a witness must sign here:

I was present while the benefits, risks and procedures were read to the volunteer. All questions were answered and the volunteer has agreed to take part in the research.

_____ _____

Date Name and signature of witness

I certify that the nature and purpose, the potential benefits, and possible risks associated with participating in this research have been explained to the above individual.

_____ _____

Date Name Signature of Person Who Obtained Consent