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# Fetal Congenital Anomalies in Africa: Diagnostic and Management Challenges

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## Abstract

There is paucity of knowledge on the causes, diagnosis, management and prevention of fetal congenital anomalies in Africa. The chapter will highlight on the general causes and specific factors concerning congenital anomalies in Africa. The problems of diagnosis and management of congenital anomalies will be extensively discussed. There is also going to be a discussion on how fetal anomalies contribute to maternal and perinatal mortality and morbidity. Screening of congenital anomalies is another black point and will be discussed emphasizing on simple strategies applicable in resource constrained environment. A section will be dedicated on prevention of fetal congenital anomalies, particularly prevention of specific factors that increase the risk of fetal anomalies in Africa. Finally, there will also be discussion on collaborative care as a panacea in the management of fetal congenital fetal anomalies, including my experience in this area. Specific examples will be given to illustrate the utility of collaborative in resource limited countries.

**Keywords:** congenital, anomalies, diagnosis, management, Africa

## 1. Introduction

Congenital anomalies can be defined as structural or functional anomalies [e.g. metabolic disorders] that occur during intrauterine life and can be identified prenatally, at birth or sometimes may only be detected later in infancy, such as hearing defect [1]. Every year, an estimated 7.9 million children are born with serious birth defects of genetic or partly genetic origin [2]. Over 1 million more infants are born with serious birth defects of post-conception origin including those that result from maternal exposure to environmental agents [teratogens] such as alcohol, rubella, syphilis, and iodine deficiency that can harm the developing fetus [3]. Thus, an estimated 9 million infants—representing approximately 7% of births—are born annually with defect that may kill them or results in lifelong disability [4]. An estimated 270,000 newborns die during the first 28 days of life every year from congenital anomalies [5]. Ninety-four percent of children with birth defects are born in low-income countries and 95% of those who die as a result of birth defect also born there [6]. Accurate data of prevalence of congenital fetal anomalies rare. Data is usually obtained from registries of congenital anomalies and this indicates that congenital anomalies are seen in 2–3% of newborns which is similar to what is seen in the industrialized world [7]. Congenital anomalies account for 8–15% of

perinatal deaths and 13–16% of neonatal deaths in India [8]. Reliable data from low-income countries on fetal anomalies like data from other health indices is difficult to come by and even where available it is generated from institutional studies rather than from population based studies. Yet most congenital anomalies and their severe consequences are seen in developing low-income. In developed countries common causes of perinatal and neonatal have been dealt with and congenital anomalies are now seen as causes of perinatal and neonatal death. In developing countries, the reverse is the case and this may be the reason why the contributions of congenital anomalies in perinatal and neonatal mortality is well appreciated. So the augment goes in view of the presence of commoner causes of perinatal and neonatal mortality it will not be appropriate to allocate resources trying to reduce mortality from congenital anomalies. The is question now is, can we wait to achieve health transition in which common causes of perinatal and neonatal mortality are eliminated before addressing the issue of congenital anomalies and their contribution to perinatal and neonatal mortality? The answer is no. What we fail to realize is in developing countries congenital anomalies indirectly contribute to maternal mortality. Imagine a situation where a pregnant woman goes in to labor with a fetus with an undiagnosed congenital anomaly that preclude vaginal delivery and as we know more than half of pregnant women in developing countries labor and deliver at home. In this scenario the labor be prolonged and with time obstructed, membranes would have ruptured, chorio-amnionitis would have set in and as consequence develop postpartum hemorrhage or puerperal sepsis and die or she develop ruptured uterus and die. Congenital fetal anomalies can lead to both perinatal and maternal mortality and morbidity.

In this chapter, I will discuss congenital anomalies, their causes, prenatal diagnosis, treatment and prevention with the peculiarities of the African environment in view.

## **2. Background**

Multitudes of factors determine the overall quality of health and pregnancy safety and outcome in Africa.

Illiteracy and poverty are factors that directly or indirectly influence health and pregnancy outcome in Africa. In Sub-Saharan Africa proportion of workers living in extreme poverty is 57% [9]. Other factors are ignorance, superstitious beliefs, bad cultural practices and poorly developed health infrastructure. The rate malnutrition in Sub-Saharan Africa is 23% [10]. In many countries in Africa utilization of antenatal care services is low and it is at this critical time pregnant women are screened for various diseases and ultrasound screening is done for fetal anomalies. In Nigeria only 2/3 of pregnant women attend antenatal care and only 40% deliver under the care of skilled birth attendants [10]. Utilization of the antenatal and delivery services in other African countries is much lower. Sub-Saharan Africa has the lowest contraceptive prevalence 13% and highest unmet need for family planning 28% [10]. Infections and infestations, lack of immunization against diseases that may be harmful to pregnancy and exposure to various potentially harmful substances increase the risk for fetal anomalies in Sub-Saharan African women. Lack of reliable data collection and recording means that the prevalence of congenital anomalies in the region is based on institutional estimates not the actual numbers. This is the background to understanding the causes, management and prevention of congenital anomalies in Africa. It is with this background in mind that this chapter will discuss the causes, management and prevention of congenital anomalies in Africa.

### 3. Causes of congenital anomalies

Fifty percent of birth defects have no clear identifiable cause and in the other 50% there are factors that considered as the cause. The causes can be broadly classified in to 3:

- Pre-conception
- Post-conception
- Birth defects of unknown cause (**Table 1**)

The pre-conception causes of birth defects are those causes that have their origin before conception and are genetic or partly genetic in origin [Genes and Chromosomes]. They found in families and can be inherited e.g. Sickle cell disease. They can also be seen as isolated incident in a particular pregnancy. The post-conception causes of birth defects are those anomalies that arise after conception or but before parturition. The last category of birth defects are those whose cause is unknown. The prevalence of birth defects based on the cause as shown in the above table is a broad division based on what is found in developed countries. If population based studies are conducted the findings differ from those above. This is because in developed countries concerted efforts were made to reduce environmental exposure to teratogens in pregnancy, institute preconception care to optimize medical conditions before pregnancy, immunize against infections that may affect the fetus in utero, offer pregnancy termination to where anomalies are identified etc. These measures reduce the prevalence of congenital anomalies of genetic origin or environmentally induced. In Africa environmental factors may play a role in causing birth defects. These may be from diseases [Viral, Bacterial and Protozoan/parasitic] or from exposure to teratogens [Alcohol, Cigarette, Pesticides and traditional medications who chemical constituents are unknown]. There is a difference in the annual numbers of birth defects, annual deaths from birth defects and annual under-5 deaths between Low-income, Middle-income and High-income countries.

Cause	%
<b>Pre-conception</b>	
Chromosome disorders	6
Single gene disorders	7.5
Multifactorial	20–30
<b>Subtotal</b>	40
<b>Post-conception</b>	
Teratogens	7–8%
Intrauterine abnormalities	2
<b>Subtotal</b>	10
Unknown cause	50
<b>Total</b>	100

*Ternpenny and Ellard [11].*

**Table 1.**  
*Percentage of birth defects by cause in high income countries.*

	Low-income countries	Middle-income countries	High-income countries	Total
Annual total birth defects (millions)	4.75 60%	2.64 34%	0.49 6%	7.9
Annual early deaths of birth defects (millions)	2.38 72%	0.79 24%	0.14 4%	3.3
Annual under-5 deaths (millions)	8.8 80%	1.8 16%	0.6 4%	11.2

MOD [3]; UNICEF 2001.

**Table 2.**  
*Estimated numbers and percentage of annual total birth defects, early deaths due to birth defects, and under-5 deaths for low-, middle-, and high- income countries.*

This is a demonstration of the impact of strategies put in place to control birth defects based on the level of economic development and investments made in health care infrastructure and health care provision and prevalence of modifiable risk factors for birth defects (**Table 2**).

4. Chromosomal abnormalities as cause of birth defects

These account for 6% of birth defects in developed countries in industrialized countries [11]. The most common example of is Down syndrome which is characterized by an extra chromosome and is also called trisomy 21. This is condition is now diagnosed early [Thickened Nuchal translucency, absent or hypoplastic nasal born etc.] and pregnancy can be terminated. Other defects in this category include Edward’s syndrome and patau syndrome. Many infants are born with Down syndrome in Africa because early diagnosis and termination is not possible. Lots of resources are expended by families and communities on caring for the affected infants which constitute a burden for the family.

Single gene defects an estimated 7.5% of birth defects [11]. They are caused by alteration gene structure and more than 6000 single gene defects were described.

Environmental factors as cause of birth defects.

**Intrauterine infections;**

**Bacterial infections:** Example, Syphilis which in Sub-Saharan Africa is seen 6–16% of pregnant women [12]. Currently most countries in Africa screen for Syphilis during antenatal care and provide treatment for those affected. Those affected present with features of the disease in the first 3 months of life which include; Vesiculobullous eruptions or macular copper-colored rash on the palms and soles and popular lesions around the nose and mouth as well as petechial lesions.

**Protozoan infection:** Congenital toxoplasmosis occurs as a result of maternal vertical transmission to the fetus. It is a cause of severe fetal complications that may manifest in the early neonatal period but may manifest later and lead to life-long complications. Diagnosis is through laboratory tests, however ultrasound is helpful and can be used to assess prognosis. The classical triad of congenital Toxoplasmosis are; chorioretinitis, hydrocephalus and intracranial calcifications.

**Viral infections:** Many viral infections are implicated as cause of several birth defects. Some viral diseases present with non-specific clinical features and many other infections have similar features. In Africa with poor health infrastructure screening and diagnosis for most diseases is challenging. Thus viral diseases may affect pregnant women and cause fetal congenital anomalies which may not in the



long run be linked to the actual cause. Some common viral infections associated with congenital birth defects include; **Zika virus**, **Cytomegalovirus**, **Rubella virus**.

**Zika virus:** Infection Zika virus was first recorded in East Africa in 50s [Uganda and Tanzania]. It has recently caused epidemic in the Americas and travel advisory was issued with regard to this infection. The virus causes microcephaly and other congenital abnormalities known as Zika syndrome. It is also associated with other pregnancy complications including preterm birth and miscarriages.

**Cytomegalovirus [CMV]:** Women with cytomegalovirus infection have 1 in 3 chance of transmitting the infection to their fetus through the placenta. Not all exposed babies present with disease or its complications. There some ultrasound features detectable during prenatal screening which include; microcephaly, intracranial calcifications, ventriculomegaly, ascites, hepato-splenomegaly, intestinal, periventricular or hepatic echo densities and fetal hydrops. One or more of these may indicate congenital CMV.

**Rubella virus:** Rubella is one the most dangerous viral infection that lead to serious complications in the fetus. Approximately 25% of infant born to mothers who contract rubella in the first trimester of pregnancy have congenital rubella syndrome [CRS] [4]. In countries with successful rubella immunization programs, CRS has been eliminated. In the remaining 50% of countries, more than 100,000 infants are born with CRS annually [13]. The common birth defects from CRS include; cataract, heart defects, low birth weight, skin rash at birth, deafness, and intellectual disabilities. Others include glaucoma, brain damage, thyroid and other hormone problems.

## 5. Drugs and birth defects

Drugs of various types are known to cause congenital birth defects. In Africa most drugs are bought over the counter as there are strict regulations. Because of poverty low medications are preferred by patients and such drugs may have potential side effects including embryo toxicity. As most pregnancies are unplanned women on treatment for some medical conditions may become pregnant and continue taking treatment with drugs that are teratogenic to the fetus. As antenatal care patronage is low women on such drugs may not be discovered until damage has been. Diseases requiring drug treatment either singly or in combination are common in Africa, ranging from infections, endocrine diseases such as diabetes and thyroid diseases, haemoglobinopathies, Epilepsy, leprosy, etc.

**Misoprostol:** This is common drug used to induce abortion and is cheap, readily available and is sold over the counter in most African countries. Girls who have unwanted resort to its use without recourse to doctor's prescription. In approximately 80% of cases it fails to induce abortion and the pregnancy continue to term [14]. Misoprostol even though of low teratogenicity it is known to cause facial nerve paralysis, with or without limb defects, probably due to vascular disruption of the subclavian artery and an ischemia in the embryonic brain stem [15–17].

## 6. Anti-epileptics

Drugs such as Phenytoin and sodium valproate are known to cause birth defects.

**Phenytoin:** This is known to cause fetal hydantoin syndrome; IUGR, Microcephaly, Limb defects, Hypoplastic nails and distal phalanges, Heart defects and cleft lip [18]. Up to 1 in 10 of babies whose mothers take sodium valproate are at risk of having a birth defect and up to 1 in 40 have developmental problems as they

grow. Sodium valproate use in pregnancy can cause; Spina bifida or a cleft palate, Atrial septal defect, Hypospadias, polydactyl and craniosynostosis [19].

**Hydroxyurea:** This drug is used in the management of Sickle cell disease and in acute myeloid leukemia. Sickle cell disease is common disease in Africa and has devastating consequences and Hydroxyurea is used to reduce crises and decrease anemia. This drug can cause fetal malformations including; Partial ossification of the cranial bones, absence of eye sockets, hydrocephalus, bipartite sternbrae and missing lumbar vertebrae.

**Warfarin:** Warfarin is used for anticoagulation and the fetuses of pregnant women on treatment with the drug may develop Fetal Warfarin syndrome, Hypoplasia of nasal bridge, Laryngomalacia, Pectus carinatus, Atrial septal defect, Patent Ductus arteriosus, Ventriculomegaly, stippled epiphyses, telebrachydactyly and IUGR [20].

**Alcohol:** Alcohol consumption is very common in African communities and both local and bottled brews are consumed. This is considered as traditional. Alcohol consumption in pregnancy is associated with Fetal alcohol syndrome [FAS] characterized by; intellectual disability, behavior problems, IUGR and congenital heart defect can occur in an individual whose mother drank alcohol during pregnancy [21]. In western Cape province of South Africa, more than 4% of 6- to 7-year-old school children had FAS. Comparable studies in Johannesburg found 2.7% of children with FAS [22]. This raises concern about prevalence of FAS in middle and low-income countries where alcohol is available and used by women of reproductive age [23].

**Traditional medicine consumption:** Consumption of traditional medicine is common in Africa. These medicines are prepared from different herbs and other substances whose chemical composition is unknown. These preparations may contain chemical agents with teratogenic effects and cause congenital birth defects. Some birth defect which may be considered idiopathic may perhaps be caused by these traditional medications. Research is needed to determine the chemical constituents of some traditional medications to determine their teratogenic potential.

## **7. Deficiencies of essential elements and vitamins**

Malnutrition as stated earlier is an important problem in African communities. Most of the essential elements are needed for normal fetal development. Malnourished women of reproductive age may lack these essential substances and could be at risk of having babies with various forms of birth defects. An important vitamin whose deficiency leads to birth defect is folic acid and has been implicated as a cause of neural tube defect. Its use as a supplement in the preconception period and early pregnancy has been shown to protect development of neural tube defects. Iodine deficiency is also implicated as a cause of birth defects [Iodine Deficiency Disorder characterized by intellectual disability, motor and auditory disabilities]. The severity depends on the level of deficiency in the mother. In 1998, an estimated 60,000 babies were born worldwide with severe iodine deficiency disorder (Cretinism), and an estimated 28 million pregnancies were still at risk of less severe Iodine-deficiency disorder from maternal iodine deficiency [24].

**Exposure to pesticides/Herbicides:** In Africa subsistence farming is a source of livelihood and many women of reproductive age are engaged in it. Today there is a plethora of chemical agents used as pesticides and herbicides on the farm without wearing protective gear. As women are of the farming populations they are exposed to these potentially teratogenic chemicals and may thus be at risk of having their fetuses affected resulting in birth defects. There is the need for authorities to look into the importance and use of these agents by instituting strict controls and regulations to catastrophic effects on the population.

**Metabolic disease:** Metabolic diseases such as diabetes have for been known to be associated poor pregnancy outcomes. Diabetic women are at risk of recurrent miscarriages, unexplained intrauterine fetal death, intrauterine growth restriction, fetal macrosomia and congenital birth defects. IDDM affects 0.5% of pregnancies in industrialized countries [25], and is becoming more prevalent in Africa especially among the elites and opulent segment of the population who are increasingly adopting western life styles. Infants born of mothers with insulin-dependent diabetes [IDDM] have up to threefold risk of having a serious birth defect [4]. Birth defect prevalence in infants of diabetic mothers is related to the level of control of the disease and in middle and low income countries diabetic control is sub-optimal because of low quality health infrastructure. Common birth defects seen infants whose mothers were diabetics include; Heart defects, Spinal and brain defects, Renal, Gastrointestinal tract defects and limb deficiencies.

**How culture, beliefs and other factors influence prevalence of birth defects:** Low-income countries particularly those in Africa are in special a position regarding the prevalence of certain diseases and conditions. In Africa women get pregnant at the extremes of age. Pregnancy at the age of 35 years and above is associated with an increased birth prevalence of chromosomal trisomies, particularly Down syndrome [4]. In middle and low-income countries, a high percentage of women give birth over the age of 35 years without the availability of community education and universally available and accessible family planning services, medical genetic screening, prenatal diagnosis, or associated services [4]. The birth prevalence of chromosomal aneuploidies is therefore high in these countries [26]. The percentage of births in women over 35 years ranges from 11 to 15% in developing regions of the world, compared to 5–9% in industrialized countries [27]. Another important factor affecting birth defect prevalence in Africa is the practice of consanguineous marriage. Consanguineous marriage is accepted by 20% of world's population [4]. This culture increases the birth prevalence of autosomal recessive birth defects, almost doubling the risk of neonatal and childhood death from birth defects [26, 28, 29]. Poverty is an important element in disease causation. Poor people are exposed to various deprivations leading to poor nutrition and susceptibility to infections and their attendant impact on general wellbeing. Mothers in poverty are more likely to be malnourished before and during pregnancy, and are at greater risk of exposure to environmental teratogens such as alcohol and maternal infections [30]. This will lead to congenital anomalies in the fetuses carried by those affected. In Africa malaria is endemic. Healthy carriers of recessive hemoglobin disorders (Sickle cell anemia and Thalassemia) glucose-6-phosphate dehydrogenase (G6PD) deficiency have a well-documented survival advantage against lethal effects of malaria compared to non-carriers of these conditions. As a result of this, carriers are more likely to survive to reproductive age. Over the years this has led to an increase in the population prevalence of these in tropical Africa [4]. Consequently, the birth prevalence of thalassemia, sickle cell disease and G6PD deficiency is high in malaria endemic regions of the world such as Sub-Saharan Africa, Eastern Mediterranean and North Africa, South East Asia and Western Pacific [31, 32].

**Prenatal diagnosis its benefits and its challenges in Africa:** The benefits of prenatal diagnosis can be viewed from different angles.

1. It provides an opportunity to classify the anomalies into lethal [e.g. Anencephaly, Bilateral renal agenesis] and non-lethal [e.g. Cleft lip, Polydactyl]
2. Surgically correctable and non-surgically correctable



- 3. It helps to identify those defects that are amenable to intra-uterine surgery and those that can be managed postnatally.
- 4. It helps decide when best to deliver, how to deliver and where to deliver
- 5. It provides us with a window to counsel the parents on the nature of the defect, treatment options and prognosis and thus assist them make an informed choice.
- 6. Where management is not available at the facility where diagnosis made, appropriate and timely referral can be made.

Prenatal diagnosis is testing for disease or condition in a fetus before it is born [1]. The aim of prenatal diagnosis is to detect birth defects which can morphological, genetic or biochemical. It involves different processes and it can be broadly classified in two, invasive and noninvasive. The invasive test requires taking fetal tissue which could be blood [Cordocentesis], placental tissue [Chorionic villus sampling] and amniotic fluid [Amniocentesis]. The non-invasive tests involve the use of ultrasound to image the various structures of the fetus to identify the normal or the abnormal. Ultrasound thus detects morphological aberrations [e.g. gastroschisis, omphalocele, anencephaly], or serve as a means of getting access to fetal tissues for further testing [CVS, Fetal amnio and Cordocentesis]. With further advancement in scientific techniques non-invasive test can now be done on maternal blood [Harvesting fetal cells in maternal blood and subjecting them to genetic testing]. In Africa prenatal diagnosis and screening for congenital defects is at the stage of infancy as the personnel and facilities are few and in most places non-existent, where available accessibility and affordability becomes an issue (Table 3).

Urban Area Rural Area				
Geographical area	One examination	≥ 3 examinations	One examination	≥ 3 examinations
North Africa	88%	53%	20%	5%
Sub-Saharan Africa	32%	14%	6%	1%
Southern Africa	68%	38%	18%	6%
Source: Matres Mundi International Africa.				

Table 3.  
Prenatal ultrasonography in Africa.

Geographical area	North Africa	Sub-Saharan Africa	Southern Africa
Ultrasonographic PD	<50%	<10%	<25%
Biochemical PD	<5%	—	<5%
Invasive PD	<2%	—	<2%
Financial arrangement	Private	—	Private
Source: Foulkese Set al. [10].			

Table 4.  
Percentage of prenatal diagnostic techniques.

Prenatal diagnosis has positively impacted on our knowledge of congenital anomalies that is why it is essential even in low-income countries. Prenatal ultrasound diagnosis of congenital defects is one of the black points of African ultrasonography [33]. Ultrasound as an instrument for prenatal diagnosis is now available in many African countries but its application in prenatal diagnosis still faces a lot of challenges. The number of congenital defects diagnosed is very low and nearly always late in pregnancy [33]. This does not provide opportunity for meaningful and timely interventions. As there are no established screening programs in Africa, diagnosis of congenital defects is opportunistic and happens by chance. Malformation detection rates do not exceed 20% [34–36] (**Table 4**).

## **8. Reasons for the challenges**

1. Poorly trained/untrained service providers
2. Inappropriate/obsolete equipment
3. Lack of ultrasound facilities in rural Africa where the bulk of the population reside
4. Absence of dedicated screening programs for congenital anomalies
5. Absence of laboratories for genetic and chromosomal analysis
6. Accessibility and affordability are important issues even where the services are available

In general, prenatal diagnosis for congenital anomalies is opportunistic and most often it happens by chance. Because of wide spread poverty the largest proportion of pregnant women are excluded from the opportunity.

## **9. Management of prenatally diagnosed congenital anomalies in Africa**

Management/Treatment of any clinical condition including congenital anomalies is hinged on accurate and reliable diagnosis. Accurate diagnosis requires well trained personnel and appropriate equipment. A comprehensive management will also require the services of different specialists [Obstetricians, Neonatologists, Pediatric surgeons, special care nurses trained in care of infants with congenital anomalies]. These are all hard to come by in Africa. People generally recognize gross physical anomalies, hidden anomalies are not appreciated before birth [e.g. Cardiac anomalies]. It is only when the child is born and start manifesting with clinical symptoms that the parents will appreciate the problem. When a child is born with gross anomalies such anomalies may be associated with some syndrome. When the gross anomaly is corrected the genetic syndrome problem will remain and will manifest itself. Parents will attribute the manifestations of the genetic syndrome to metaphysical causes. It is thus difficult to make them understand the real cause and the possible remedy. As facilities for genetic/chromosomal analysis are few and in most cases non-existent it becomes difficult to make

comprehensive evaluation and diagnosis. This makes final decision on management extremely difficult for the physician in Africa. Such is the environment perinatologists practice in Africa.

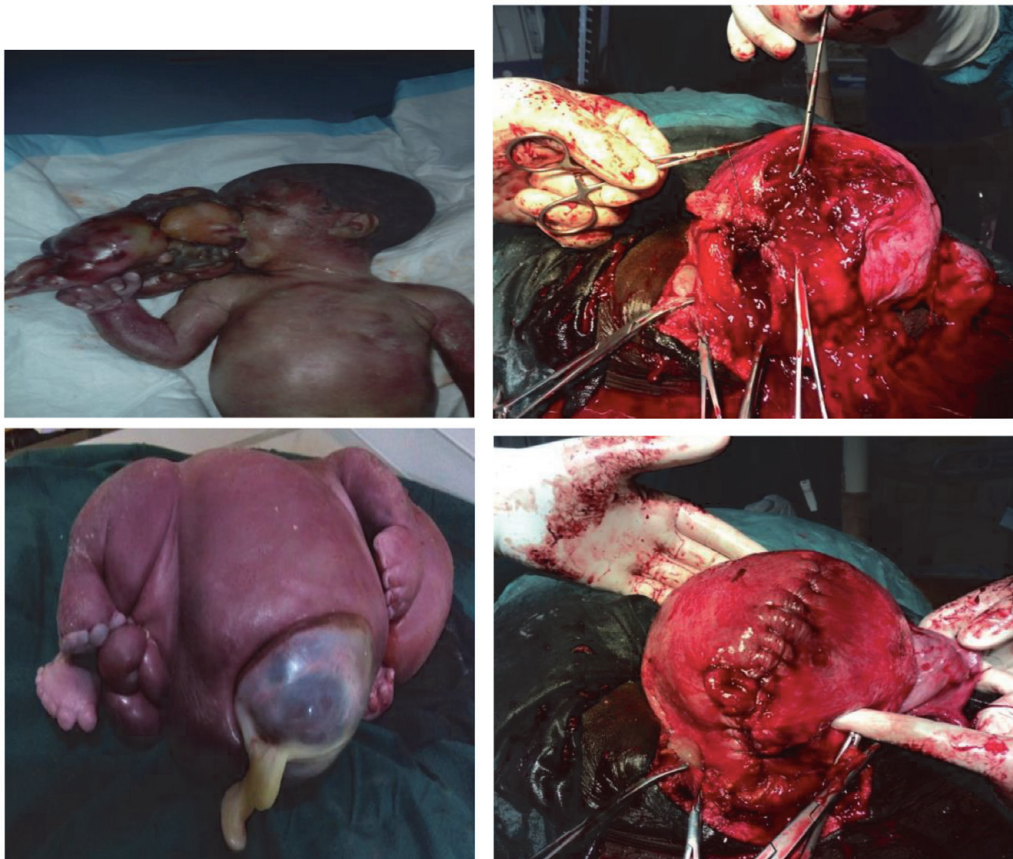
In this circumstance management of congenital anomalies cannot comprehensive and will be provided in a scattered manner. Often times when an anomaly is diagnosed treatment is limited to pregnancy termination where the laws allow. In one hospital 65% of pregnant women request for pregnancy termination when an anomaly is found in their fetus. In those with distressing polyhydramnios intermittent aspiration of the amniotic fluid is done to relieve the distress. Women who present with obstructed labor and a dead congenitally malformed fetus with hydrocephalus, delivery can be effected by craniotomy. For those that present with ruptured uterus, laparotomy is done and further management will depend on the extent of the rent and the clinical state of the fetus. Few countries [e.g. Egypt and South Africa] have centers that offer prenatal screening, diagnosis, treatment and follow up services, however such centers are not within the reach of the poor who carry most of the burden of congenital anomaly.

**10. Illustrative cases**

The cases below illustrate how pregnant women with undiagnosed fetuses with congenital anomaly will labor at home and developed ruptured uterus and present for treatment. All these cases presented with ruptured uterus following various interventions at home.







### 10.1 The way forward: collaborative care

This approach will require pooling of resources [Manpower, Equipment and other resources] to create referral hospitals in different countries and regions to serve as one stop shop able to provide care in all aspect of management of fetal congenital anomalies. Government, the private sector, philanthropists and other non-governmental organization can come together to establish such centers. Examples abound where similar collaboration has provided opportunity for treatment of some medical diseases requiring specialized care. The cardiothoracic center in Accra Ghana is now a regional center for referral that offer treatment for patients from different countries in West Africa as well as provide training for resident doctors from the whole sub-region. The first renal transplant at Aminu Kano Teaching Hospital was sponsored by philanthropist who also invites specialists from Britain and today thank to that effort the hospital is a referral and training center in renal transplant. In the management of congenital fetal anomalies similar approach can be adopted. The collaborative care group that I established at Abubakar Tafawa Balewa University Teaching Hospital Bauchi while I was there had achieved some success. We educated the community, Counsel parents, managed some pregnancies complicated by congenital anomalies and surgically treated a few cases despite the challenges we had. This can be replicated in other teaching hospitals.

## 11. Preventing birth defects: which approach?

The prevention of birth defects in Africa should be modified from the traditional approach that is adopted in other regions of the world. This is because of the



peculiarities of the African environment. As much as possible all preventive strategies should be simple, low cost and innovative. All stakeholders must be involved, governments, communities, professional societies dealing with the issue, and non-governmental organizations. First stage should involve educating policy makers with emphasis on the burden of the problem on families and society at large. Educating policy makers on the need to understand how congenital anomalies contribute to both maternal and perinatal mortality. Areas that will require government intervention especially in community education, training of personnel and provision of equipment. Involving traditional and religious leaders who are gate keepers, educating them on the causes and prevention of birth defects. Involving the media in community enlightenment through discussion programs, talk shows and jingles.

Even as we want evolve our own model of prevention based on our peculiar social, environmental and economic circumstances, we must learn lessons from the experiences of other countries such as Cuba, China, India and Brazil. We must extract some of elements in the model they used and input them in to our own model. As we develop our preventive strategies we need to have the following at the background for us to succeed:

1. Endemic poverty and illiteracy
2. Uncontrolled birth rates and poor uptake of family planning services
3. Malnutrition and micronutrient deficiencies
4. Poor uptake of immunization services
5. Loose control of drugs and substances which are known to be teratogenic
6. Customs and traditions which are harmful to health
7. Our goal is to eliminate causative factors of congenital defect
8. Emphasis should be on primary prevention as we prepare to introduce secondary and tertiary prevention.

This is background to discussing the strategies for the prevention of congenital anomalies in Africa.

First strategy is education which is the backbone of development and progress in all spheres of life. It is known that the higher the level of literacy of a community the better are its economic indices, social status and health seeking behavior of its members. Education may be crucial in understanding the causes, treatment options and methods of prevention of congenital anomalies. It will also go along way in eliminating traditional and cultural practices that put communities at risk for congenital anomalies.

## **12. Specific measures**

**Immunization** is an important strategy that has proven its efficacy in preventing other diseases. Congenital Rubella syndrome has virtually been eliminated in the United States and this is achieved through universal vaccination as a component of childhood immunization. In contrast the burden of congenital rubella in developing countries has been estimated to be about 100,000 per year [37], but only 28% of all developing countries have rubella immunization in place, as compared to 92% of

industrialized countries [38]. Africa is the continent worse off compared to other regions as only one of 47 countries in Africa immunize against rubella. Currently there is an effort to develop a vaccine against Zika. For immunization program to succeed political will is required and thus policy makers should be encouraged by all to make commitment in this regard.

**Folic acid and other micronutrients supplementation:** Folate supplementation has been effective in preventing neural tube defect. Although available in various food substances in common use but one sure way of getting adequate levels in women before conception is through supplementation. Folate is chief and can be afforded by women in Africa. Different countries have used different approaches to ensure that women of reproductive age get adequate levels prior to conception. The United States Public Health Service recommended that women capable of becoming pregnant should consume 400 µg of folic acid daily [39], and in 1996 the Food and Drug Administration mandated the fortification of all enriched grain products, like flour and pastas, with 140 µg per 100 g of grain. The efficacy of folic acid in preventing neural tube defects has been proven in a community-based intervention study in China. In this study 400 µg of folic acid pills alone given before conception was found to be effective in reducing neural tube defects by 85% in an area of high prevalence and by 41% in an area of low incidence [40]. South American countries have implemented various successful folic acid supplementation programs [Chile and Cuba], supported by various organizations including the Pan American Health Organization, the March of Dimes and the CDC. There are plans being implemented to monitor the effect of the policy on the prevalence of neural tube defects, taking advantage of a preexisting and ongoing register of congenital malformations [41]. Africa can take queue to implement a similar program and follow up to determine its effectiveness in preventing neural tube defects in the African environment.

**Family planning:** Africa has one of the highest fertility rates in the world and this is creating concerns as it is overstretching resources in the continent. High fertility rates go hand in hand with prevalence of various pregnancy complications including birth prevalence of congenital malformation. To address this, Africa has to make effort to reduce its population growth. Uncontrolled population growth is a precursor to poverty, disease and malnutrition all of which contribute to the prevalence of congenital malformation. Family planning reduces the prevalence of congenital malformations by reducing birth rates and fertility. It has been estimated that in many countries with high fertility reducing the number of children per family to 2–3 could reduce the prevalence of genetic disorders by 40–50%. Further, combined with encouragement to complete reproduction before the age of 35 years, family planning can contribute to a 50% reduction of Down syndrome [42].

**Avoidance of teratogens:** Exposure to teratogens has multiple angles. In Africa use of traditional remedies is common in many communities. The chemical constituents of such remedies are not known and could contain teratogenic substances. Another factor is behavior of pharmaceutical companies selling drugs to consumers without following the regulatory procedures. People can also buy drugs over the counter without recourse to physician's prescription and use it anyhow with the potential of harm. Again counterfeit drugs are everywhere and people buy them because they are cheaper but their potential for harm is much greater compared to the original. Compounding these factors are lax environmental quality regulations and unhealthy working conditions which expose pregnant women to environmental pollutants [43]. Community education on what teratogens are, teratogens in the vicinity of communities and how they can cause harm can go along way in reducing exposure.

**Other measures:** Premarital counseling and testing can go along way in reducing genetic/hereditary disorders. A case in point is sickle cell disease in which

premarital counseling and testing can reduce its prevalence. Avoiding consanguineous marriage has the potential of reducing propagation of hereditary diseases within population groups with hereditary diseases.

**Secondary/tertiary prevention:** This aims to reduce the number of children delivered with congenital malformation, whereas tertiary prevention is aimed at cure and amelioration of problems once a child with a congenital birth defect is born. Postnatal neonatal examination and screening of newborn children is a strategy in tertiary prevention, because once an anomaly is detected ameliorative measures can be instituted.

**Ultrasound screening of congenital anomalies with an option of pregnancy termination:** This has the potential of reducing the birth prevalence of congenital malformation but the issue of termination of pregnancy is the difficult part, as in many countries in Africa pregnancy termination can only be done when the life of the woman is at risk. Congenital malformations are not in themselves life threatening to the woman and therefore pregnancy cannot be terminated on account of congenital malformation in countries with restrictive abortion laws.

**A new concept [Targeted screening]:** The ideal thing is to screen all pregnant women for structural anomalies and test them for hereditary disorders and infections but the ideal is not always possible because the health care system in Africa is constrained by limited resource allocation. In view of this a transitional concept can be adopted pending acceptance of the whole population to prenatal screening and improve resource allocation to health care.

Targeted screening can be offered to the following category of pregnant women;

Women with; Previous history of babies with congenital anomalies, history of congenital anomaly in the family, index pregnancy with polyhydramnious, age more than 35 years, multiple gestation, consanguineous marriage, diabetes and those with sickle cell disease/Thalassemia.

### **13. Conclusion**

Birth defects or congenital anomalies are important cause of perinatal mortality and morbidity. In developed countries successes were achieved in screening, treatment and prevention over the years. In Africa the picture is different as many factors play a role in causing congenital anomalies different from those seen in developed countries. In Africa factors such as poverty, illiteracy, malnutrition, exposure to teratogens and poor environmental control play an important role. Screening, treatment and preventive services for congenital anomalies are poorly developed. To achieve control primary prevention should be established and strengthened and when this is achieved, then secondary and tertiary control should follow. Innovative strategies should be employed in this endeavor.

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