Proceedings of the Anatomical Society of Southsouth and Southeast Nigeria (ASSEN)

The seventh scientific conference of the Anatomical society of Southsouth and Southeast Nigeria was held between the 29th of November and 1st of December 2007, at the Department of Anatomy, University of Calabar, Calabar Nigeria. The following are some presentations during the opening ceremony.

Guest lecture delivered by Prof Asindi A Asindi at the 7th Scientific Conference of the Anatomical Society of South-South and SouthEastern Nigeria (ASSEN) in Calabar on Nov 30, 2007.

TOPIC: Congenital Malformation. Record Keeping in Nigeria: problems and prospects.

I greet and congratulate the Anatomical Society of South-South and South-Eastern Nigeria on her 7 Scientific Conference. I thank the local organizing Committee of this Conference for identifying and nominating my humble self to present a guest lecture on this occasion.

I studied my Anatomy as a medical student in the University of Ibadan in the late 1960s. In those days, we were taught gross anatomy just as demonstrated on the cadaver, pots, textbook pictures and :the atlases. The issues were that of gross regional and systemic anatomy. I used the Cunningham's Textbook of Anatomy and the famous textbook of Grant. By then, the recordings or contents in these volumes were on pure undiluted human parts; no issues were mentioned on variations or abnormalities either acquired or congenital. Indeed clinical anatomy which emphasizes structures and functions as they relate to the practice of medicine and other health sciences was not communicated to us. Nevertheless, I want to pay tribute to the renowned Prof Ade Grillo and Prof Adesalu who were my anatomy teachers in Ibadan. Though the two were medically qualified, they did not reflect the clinical aspect when imparting the subject. I suppose the curriculum in those days was not so clinically designed. From the early 1980s there has been a heavy presence of clinical medicine in the teaching of anatomy, embryology and histology.

My area of interest and subspecialty is Paediatric neurology hence I very often refer to textbooks of anatomy so as to remain fresh on my neuroanatomy, regarding craniospinal axis, dermatomes, myotomes, relationships. and courses and actions of both cranial and spinal nerves etc. One is excited about the progressively increasing inputs of applied and clinical anatomy in these volumes over the past two decades. The Clinically-oriented ANATOMY text co-authored by KIETH MOORE & AUTHUR DALLEY carries the foot and side notes labelled "clinical blue boxes," clinical pictures, plain radiographs and diagnostic radioimaging scans such as CTs and MRIs. Science students of medicine, dentistry, physiotherapy and other allied health sciences in the 21st century are therefore at a greater advantage and stand at a higher pedestal than we were in our pre-clinical years. Indeed, the current perspective in the teaching of anatomy can be regarded as student-friendly. Health science students can therefore step into the clinical years fully armed and with great confidence to face rigors ahead of them.

I can assume that it is this new concept and modern design in the delivery of Anatomy subject that has informed the choice of the theme of the present 2007 7th Annual General meeting and Scientific Conference of your ,Society: The theme which is Congenital malformation with the sub-theme Record keeping in Nigeria-problems and prospects, is therefore elegant. The statistics or auditing prevails is an essential element for progress in medical treatment, rehabilitation and prevention. The ASSEN is therefore moving with the times and is walking in tandem to meet the Millennium Development Goals. Congenital malformation belongs to the field of dysmorphology related to embryogenesis which has expanded dramatically as the number of recognizable patterns of malformation has more than tripled during the last 30 years. With the advent of high resolution neuroimaging technology and gene engineering capabilities, new insights have been gained in the prenatal and postnatal diagnosis and in

the pathogenesis of congenital defects.

Congenital malformation is defined as a permanent change produced by intrinsic abnormality of development on a body structure during prenatal life. The true incidence of congenital malformation in a geographical area can only be determined if all livebirths, fetal deaths and spontaneous and induced abortions are examined. It is estimated that congenital anomalies occur in 3% of all infants globally.

Congenital anomalies including structural malformations, chromosomal abnormalities and metabolic disorders are becoming the most important cause of perinatal mortality (about a quarter of all perinatal deaths) in the countries of Europe, and, after prematurity, the second cause of infant morbidity. Beyond the direct impact on the affected children and their families, they impose a tremendous financial burden on medical treatment, and on educational and support services.

The pattern and prevalence of birth defects may vary over time or geographical location thereby reflecting a complex interaction of known and unknown genetic and environmental factors including sociocultural, racial and ethnic variables. For instance the incidence of neural tube defect is higher in the Great Britain, Ireland and lowest in Asia and Africa. Consanguineous marriage is an important correlate of congenital malformation. Congenital malformation, being largely genetically related, is more prevalent in area with a high incidence of consanguineous marriages (first and second cousin marriages). In a study conducted on 3103 Saudi females, the consanguinity rate in different provinces was range from 52%-68% with the highest prevalence in first-cousin marriages. In my study (Asindi et al 2001) on neural tube defect in Saudi Arabia, 50% of cases were derived from consanguineous marriages. High rates of inbreeding which is inherent in Middle East and some North African and Southeast Asian countries influence, the frequency of genetic and congenital malformation. This has to do with culture and religion.

The terms MALFORMATION, DEFORMITY, DYSRUPTION, and DYSPLASIA are sometimes used interchangeably but they have different connotations regarding embryogenesis and embryopathy. Each involves single primary defects in development and categorized according to the nature of error in morphogenesis that has produced the observed structural defect. There are other definitions such as SEQUENCE and ASSOCIATION.

I intend to deliver this lecture, not as an anatomist or a clinical geneticist, but simply as a clinician hence I will not get into the details of these taxonomical varieties of dysmorphogenesis.
Basically, a malformation is a primary structural defect arising from a localized error in morphogenesis. A deformity is an alteration in shape or structure of a part that has differentiated normally. The term disruption is used for structural defect resulting from destruction of a previously normally formed part. And the term dysplasia refers to an abnormal organization of a cell and the structural consequences.

In contrast to the concept of the single primary defect in development, the designation, multiple malformation SYNDROME is used when several observed structural defects all have same or presumed aetiology. The defects usually include a number of anatomically unrelated errors in morphogenesis. Multiple malformation syndromes are caused by chromosomal abnormalities, teratogens and by single gene defects inherited in Mendelian pattern.

A congenital anomaly can be classified as minor or major following guidelines set out by the European Registers of Congenital Anomalies and Twins (EUROCAT).

A minor defect is one that has unusual morphologic features that are of no serious medical or cosmetic consequences to the patient. Minor external malformations are more common on the face, auricles, hands and feet. Extradigit and syndactyly are examples. Of special interest is the finding that about 0.5% of babies can have two or more minor anomalies. The finding of several minor anomalies in the same individual is unusual and often indicates that a more serious problem in morphogenesis has occurred.

A major malformation is defined as a one that results in fetal mortality, requires major surgical intervention or has a significant long-term effect on the newborn's physical and or mental functions. It

can involve a single organ, a whole system and can also be multisystemic.

What are the aetiological correlates of CM

Chromosomal disorders, prenatal effects of infection, drugs, chemicals,. Malnutrition, maternal diseases and radiation are aetiologically related to CM. The infectious agents, drugs, chemicals and radiation constitute the teratogens. For most defects the aetiology remains unknown. The so-called unknown or idiopathic cases may be regarded as cryptogenic in that, at very frequent intervals, Genomic and cytogenetic studies are linking these conditions to genetic aberrations. diseases and radiation are aetiologically related to CM. The infectious agents, drugs, chemicals and radiation constitute the teratogens. For most defects the aetiology remains unknown. The so-called unknown or idiopathic cases may be regarded as cryptogenic in that, at very frequent intervals, Genomic and cytogenetic studies are linking these conditions to genetic aberrations.

Chromosomal syndromes are by far the most common multiple congenital syndrome diagnosed in the neonatal period. It has been identified that 40%-50% of spontaneous abortions are due to chromosomally defective fetus. The chromosomal defect can be due to the presence of extra or absence of chromosome.

The most common of the chromosomally related condition in Nigeria are the Trisomies with Down Syndrome (Trisomy 21) being the most prevalent. Advanced maternal age of 40years and above and paternal age of 70 years and above are associated with CM, for example, the trisomies. The Apert syndrome which involves craniofacial deformity and gross syndactyly is closely associated with advanced paternal age.

The aetiology of neural tube defect has been proven to be closely related to folic acid deficiency in the mother during pregnancy. Folic acid deficiency can be the most obvious cause if there is a sustained high freequency of NTD in a population. Folic acid supplementation of diet is a well recognized preventive measure, with a dramatic decline in the incidence of this defect noted in many parts of the world. In the Western countries, NTD forms a large but diminishing proportion of all major CM. It needs to be given before and during early pregnancy for it to be effective.

Maternal infections in pregnancy have a tendency of causing a malformation in the first trimester but a disruption if the fetus is reached in the second and third trimester. Known infections include Toxoplasmosis, Rubella, cytomegalovirus, Herpes, syphilis and others. These have been given the mnemonic TORCHS syndrome.

Record keeping/documentation of CM in Nigeria

The true incidence of congenital malformation can only be determined if all livebirths, fetal deaths and spontaneous, and induced abortions are examined. Accurate quantification of congenital anomalies within a given population is essential for estimating their burden and documenting the need for prevention. The data collected from the monitoring system may then be used for identifying the prevalence trends, for conducting research on potential risk factors, for public health policy development, for planning and implementation of services needed by children with malformation and for evaluating the effects of preventive measures and treatment services.

A comprehensive documentation of the pattern of CM in Nigeria is virtually non-existent. Only the publications on isolated organs or the systems are available. For instance Adeloye and Odeku in UCH Ibadan had published repeatedly in the 1970s and 1980s on malformations of the nervous system in Western Nigeria. The reason for this is very obvious. For optimum documentation and statistics to be obtained, all pregnancies, deliveries and deaths must be recorded in an orthodox health facility. Problems associated with record keeping in Nigeria will emanate fi-om the following experience:

1. Only about 60% of Nigerian women attend antenatal care in government health facilities.
2. Of the 60% , only 50% deliver in hospital hence

3. Only 30% of Nigerian women deliver in orthodox health centres hence
4. About 70% of Nigerian women deliver at home and in the church or outside hospital.
5. Autopsy rate in most Nigerian hospital is known to be very low, and in some areas, non-existent.

The diagnosis of congenital malformation requires a detailed physical examination by a clinical Geneticist, dysmorphologist and a paediatrician. Where more than 70% of Nigerians are born at home and also die at home, how can one obtain anything near reasonable statistics?

Laboratory support for diagnosis

1. Appropriate investigations such as chromosomal analysis and karyotyping will require high tech methods of tissue preparation and electron microscopy.
2. Echocardiography, metabolic screening, skeletal surveys, radiography and clinical photography are necessary
3. Ultrasonography and high resolution computerized imaging involving MRI and CT are essential.

A review of standard textbook with photographs on dysmorphology databases would assist in making accurate diagnosis. This is done by studying the text and physically comparing photograph of the index case with that in the book.

There is a dearth of laboratory facilities for screening malformed infants in most of the health facilities including even the tertiary centres of care. Accurate health record keeping regarding CM will therefore remain a mirage for a very long time in Nigeria. Records on CM available in Nigeria and other developing countries are derived from newborn death and they are basically hospital-derived figures. It is quite obvious also that without a full compliment of the diagnostic tools some components of syndromic anomalies, especially those involving internal organs, can be missed.

Prospects or chances or possibility of something successful occurring in future:

The prospect of keeping accurate statistics of CM in Nigeria appears rather blunt. Currently most poor world countries are diverting attention toward combating infections, malnutrition and in fighting unnecessary wars. In Nigeria the situation is not different. Attention towards research on CM would not appear be a forefront priority even in the far future in Nigeria.

The MILLENIUM DEVELOPMENT GOALS were established and signed into action at the United Nations Millennium Summit of September 2000.This strategy endorsed by 190 countries including Nigeria, is an ambitious effort to reduce global poverty and improve the lives of people. There are 8 goals:

1. Eradication of extreme poverty and hunger
2. Achieve universal primary education
3. Promote gender equality and empowerment of women 4. Reduce child mortality
5. Improve maternal health
6. Combat HIV/AIDS, malaria and other diseases 7. Ensure environmental sustainability
8. Develop global partnership for development

The millennium Development Goals were to address the devastating problems of the world's poor, especially in the developing countries where hunger, malnutrition and poverty abound. To these goals are tagged 18 targets and it is hoped that by the year 2015 nearly all the goals would have been achieved by all world counties. Indeed, it is expected that child mortality rate would have been reduced by two-thirds before the year 2015.

In developed world countries of Europe ad America, there are hospitals established and dedicated for reproductive health and delivery in almost every city. Nigeria is long overdue in establishing health institutions dedicated to maternal/child care. This will encourage more of Nigerian women to deliver in hospital. It will cut down the tendency for overseas treatment at high cost. It could also attract highly

specialised Nigerian doctors in Diaspora to return home to serve. There are more than 10,000 Nigerian doctors in the USA alone.

CM constitutes 5-10 % of neonatal deaths in our hospitals. This is a gross underestimation since not all infants with CM die during the neonatal period. A majority are likely to linger on only to die at an older age, and most probably at home. One can therefore assert that CM is a cause for concern among Nigerians.

A case can be made for funding studies on the premise that:

1. A good number of Nigerian women are still reproductive beyond 40 years of age hence the incidence of the Down syndrome remains the risk. This should therefore attract intensive genetic counseling, if proven.
2. There is the culture of indiscriminate use of off-the-counter drugs and herbal concoctions for medication by Nigerians including the women. These are potentially teratogenic.
3. A huge consumption of higWy concentrated alcohol beverages has been observed among the people/women living in the creek areas of South South Nigeria. This can also be teratogenic.

A visit to Homes for the mentally and physically handicapped children will make one appreciate the array and enormous dimension of congenital problems that can afflict Nigerian children.
Other congenitally deformed children and adults are on our streets begging for alms.

I want to assume that most of the deformed children are hidden in their homes to avoid embarrassment to the families. Population/community surveys on CM can therefore produce a more realistic statistics/ figure.

For the Anatomical Society of Nigeria and also the Paediatric Association of Nigeria it is indeed a challenge to rise to the occasion by making a case that can justify government
and academic institutions making funds available for a national, regional or local survey on CM. There will surely be a cost-benefit to be realised and appreciated.

In Britain there are numerous NGOs formed in relation to some congenital diseases whereby both the parents, the affected children and medical scientists are members e.g. Down syndrome Society, Retts Syndrome, Spina Bifida etc. Part of their aims is to solicit for funds to support research on these diseases. There are a few such health-related societies in Nigeria e.g. Achondroplasia, Albinism. Unfortunately, I am not aware of their objectives. This is an avenue to be exploited for research.

Finally, it is expected that the on-going V AMED supplies of hitech equipment by the Federal Ministry of Health to the Nigerian tertiary health institutions will go a long way to enhance research on congenital defects. Medical scientists cannot afford to give up. Thank you.

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