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Down Syndrome in Nigeria Sub Saharan Africa

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1. Introduction

1.1 Burden

In his publication in 1866 titled 'Observation on an ethnic classification of idiots', John Langdon Down an English physician at the London Hospital, was the first to describe the external appearance of the genetic condition that was later to bear his name (Down, 1866). Following his description, scientists have conducted researches aimed at identifying the presence of the condition in various populations, race and ethnic groups as well as its incidence at birth and prevalence in the population. Most of the widely circulated reports on the incidence and prevalence of Down syndrome in Nigeria and other countries in sub Saharan Africa are estimates extrapolated from statistics obtained in developed countries such as the United Kingdom, United States of America, Canada or Australia. The studies did not consider the influence of factors such as socio cultural, genetic, racial and environmental characteristics on the prevalence of the condition, thus affecting the reliability of the data.

The earliest reported study on the incidence of Down syndrome by Adeyokunnu, in Ibadan, South Western Nigeria, reported an incidence of 1 in 865 live births (Adeyokunu, 1982). Prior to this time, it was believed that Down syndrome is rare or non-existent among Africans (Tompkins, 1964). This is corroborated by the reports of other clinicians that found no case of Down syndrome in their clinical practice over several years in Nigeria (Jelliffe, 1954a, Tooth, 1950). Rather, few cases reported were among Jamaicans and were postulated to be derived genetically from non African sources (Jelliffe, 1954b). While there may be other reports from Nigeria, the case report by Tompkins in 1964 was the first to draw attention to the occurrence of the condition among Nigerian children (Tompkins, 1964). After the report, it was now agreed that the condition is not as rare among Nigerians and Africans as was once believed. In spite of this however, there was still difficulty in achieving accurate data collection in Nigeria and other developing countries attributable to many factors. First, a large number of deliveries take place in non orthodox centres such as churches and traditional birth homes, most of which do not keep records. The government registered private maternity centres also have difficulty keeping accurate statistics (Oloyede, et.al, 2006). Secondly, within the community, cases of congenital malformations such as Down syndrome are not reported for record purpose because of the traditional belief that still associates them with witchcraft and witches. Consequently, true population based data

are difficult to generate and most of the data reported are hospital based. In spite of this however, health planning are still based on these data with the assumption that they represent the actual situation.

Since the study by Adeyokunnu, there has been no other published report about the incidence in Nigeria till date. However, in South Africa, the Down Syndrome South Africa gave an incidence of about 1 in 500 live births in the country (DSSA). In same country, a study between January, 1974 and December, 1993, reported an overall prevalence rate of 1.49 per 1000, with a gradual decline to 1.3 per 1000 in the last 5 years of the study period, among the 3 study populations. The higher prevalence (1.88) among the white population compared with the prevalence in coloured (1.54) and blacks (1.29) could be attributed to the possibility that fewer blacks than whites undergo prenatal screened and diagnosis of the condition. This conclusion arose from the relative distribution of the number of terminations following prenatal diagnosis, being higher among whites (18.3%), intermediate in coloured (5.8%) and lowest in blacks (1.4%). The same study also confirms that the decline in the overall prevalence occur among the white population, while the blacks maintain their prevalence rate (Molteno, et, al., 1997). It should be noted that there is a fundamental difference in the statistical inferences from incidence and prevalence. Incidence is based on total birth in a year, while prevalence is based on actual population. While the difference is significant in developed countries because there is a better uptake of prenatal diagnosis services that influences the total birth incidence, same may not be true in Nigeria, where the uptake of such service is still poor. The incidence of Down syndrome could be higher in developing countries, with two factors as possible reasons. First the proportion of women that that conceive after 35 years is gradually rising in Nigeria compared to developed countries. Second, there is a higher mortality from complications of Down syndrome such as congenital heart defects in developing countries.

1.2 Down syndrome, maternal age and reproductive health

The commonest genetic mechanism for Down syndrome is maternal meiotic non disjunction and this has a very strong link with advance maternal age. Shuttleworth in 1909 was the first to observe the association between increased maternal age and Down syndrome after examining 350 cases. (Shuttleworth, 1909). Although no study from Nigeria has confirmed this trend, but a study on 'Down's syndrome in South Africa-incidence, maternal age and utilisation of prenatal diagnoses', reported that the prevalence of Down syndrome has been found to increase with advancing maternal age (Op't Hof J, et al., 1991). There are enough reasons to justify the expectation that the prevalence would continue to rise in future. The main factor that would explain this conclusion is the increasing maternal age at which more women in Nigeria now conceive. This follows the increasing strong desire to complete formal education before conception and the rising incidence of infertility (Oloyede and Osagie, 2003).

Although there are few studies on the epidemiology of Down syndrome in Nigeria and other sub Saharan African countries, its impact on reproductive health and quality of life is well documented. It is an important contributor to pregnancy wastage especially in early trimester. This is however difficult to ascertain because of the poor culture of follow-up diagnosis especially after spontaneous first trimester abortion in Nigeria. Various congenital malformations are associated with Down syndrome. Oloyede reported the occurrence of

multiple congenital malformations in cases of Down syndrome in Sagamu, South Western Nigeria (Oloyede, et al., 2006). The pattern of congenital malformation seen in Ghana conforms to the well established pattern of malformations in Down syndrome. The main defect was atrioventricular defects and ventricular septal defects (Arthur, 1995). Other frequently reported anomalies are anorectal anomalies and speech disorders (Osei-Bagyina, 2000, Hesse, 2006). Majority present in early infancy, with features of congestive cardiac failure and cyanosis (Arthur, 1995).

Down syndrome has a lot of sociocultural colouration. Many local communities in Nigeria believed that all observable defects or early deaths which may occur in children with Down syndrome is traceable to parental misdeeds or links between the child and the evil world. In Ghana, it is believed that the Down child is given by the river gods and hence is called 'Nsuoba', meaning water children (Avoke, 1997). The pattern of response is similar in many other developing countries including Nigeria. Children that are affected by this syndrome are often times stigmatized by the society, treated with neglect and patronized. Lack of understanding and appreciation of the condition, makes the management of the individual with the condition difficult. The special educational and family support needed to optimize the well being of affected children are not usually available in many developing countries. The first coordinated effort at addressing the challenges of children with Down syndrome was provided through the National Down Syndrome Association of Nigeria and till date remains the only well coordinated group in West Africa. A similar organization is in South Africa called the Down Syndrome South Africa (DSSA). These bodies are non profit making. Like most other supportive organizations, the absence of community and governmental support is a strong limiting factor to their effectiveness. Overall, it is difficult for the child with Down syndrome child to realize the full potential in Nigeria and many other sub Saharan countries.

2. Strategies to reduce the incidence and impact of Down syndrome

The strategies to reduce the incidence and impact of Down syndrome should focus on 2 major areas

1. Implementation of acceptable and effective prevention programme in the prenatal period (Prevention)
2. Implementation of supportive health and non health initiatives for people affected by Down syndrome (Support)

2.1 Prevention programme

The seemingly rational way to prevent the birth of children with Down syndrome would be to prevent or reduce pregnancies in women above the cut off maternal age. There is however two issues involved in this policy. First, there is no clearly defined study from Nigeria that specifies the maternal age at which there is an exponential rise in the incidence of Down syndrome. Above this maternal age, the policy of no childbearing could be advocated based on the high incidence and also the risk of other complications of pregnancy above this age. Second, there are some considerations that influences the maternal age of conception such as the desire to complete educational pursuit, socioeconomic factors and increasing rate of infertility. These may altogether, increase the maternal age of conception in recent times. Having children is sometimes viewed from a completely different

perspective in many rural communities in Nigeria, where it is a taboo for women to stop further childbearing till she either dies in the process or gets too old to continue, especially in communities where children are involved in economic activities such as farming. In the final analysis, this approach to preventing Down syndrome is not feasible in Nigeria and many other sub-Saharan countries. A more acceptable compromise would be to advocate for the screening of women above the cut off maternal age.

Prevention strategy through screening and diagnosis is gradually becoming the hall mark of modern approach to the management and eradication of genetic condition in developed countries. It provides every woman an opportunity to determine the status of the fetus and takes decision on the future of affected pregnancies. This option of addressing the problem of Down syndrome is more relevant to developing countries where there is dearth of facilities to manage the physically challenged. In Nigeria, prenatal screening was considered the most feasible approach for the control of Sickle cell disorder (WHO, 1997). Genetic screening refers to the procedure to identify from within the population, individuals that are more likely to be produce offsprings with Down syndrome. Screening is not fool proof method as it merely suggests the propensity of occurrence of the condition. Its efficacy therefore improves as the number of tools used also increases.

The first and traditional screening method for the prevention of Down syndrome based on maternal age was introduced in the early 1970's, based on the observation by Shuttleworth (Shuttleworth, 1909). Although many physicians in Nigeria utilize maternal age in counselling women for the risk of Down syndrome, few utilize it for the purpose of screening. Op't Hof in South Africa shows that maternal age has influence on the risk of occurrence of Down syndrome in the country (Op't, 1991). The challenge is to arrive at an acceptable maternal age that can be used for screening in Nigeria, if it must differ from that used in Caucasians. This is because of the few reasons discussed earlier that contribute to increasing maternal age of conception. The decision about cut off maternal age also has economic and logistic implications. Because more women now achieve pregnancy at advance maternal age, it implies that the number that screens positive obtained using the traditional cut off maternal age of 35 years would also rise. This means additional logistics to cope with screening services. The logical step would be to further increase the cut off maternal age in other to maintain the recommended 5% screen positive rate. An important issue to contend with is the fact that the bulk of those that will end up having Down syndrome are in women below 35 years. Therefore maternal age would not suffice for effective screening in Nigeria.

A more common practice in Nigeria is the use of maternal serum hormones in the second trimester. Often times, it is not primarily intended for Down syndrome screening. The disadvantage of the present practice is that, screening is done at a gestational age when strong bonding has occurred between the mother and the fetus and the risk associated with extreme decisions such as termination of pregnancy is higher. Studies have indicated that, between 79.9 and 86% of women present either in the second trimester or third trimester for the first time in the antenatal period rather than in the first trimester, often times out of ignorance or misconceptions of the purpose and right time to commence antenatal care (Okunolola et al, 2008, Adekanle and Isawunmi, 2008, Ebeigbe and Igberase, 2005). The mean booking gestation in Nigerian is 20 weeks (Oladokun, et al., 2010). This observation is similar to reports from many sub-Saharan African countries but in sharp contrast to the

practice in United Kingdom and United States of America, where women are advised to register within the first twelve weeks (National Institute for Health and clinical excellence, 2003, American Academy of Paediatrics, 2002). The late registration is believed from a study in Nigeria to the fact that most women do not perceive any advantage in early booking and viewed antenatal as curative rather than preventive (Ndidi, 2010). The physicians are therefore constrained by this reason. Standardized laboratory support that is peculiar for measuring hormones in pregnancy and calculating their deviations from median is also not available. There are no available statistics on the screen positive rate using these methods because there are no well defined criteria for their utilization.

Recently, another advance tool for screening of Down syndrome known as the nuchal translucency was introduced by Nicolaides and his team in the United Kingdom (Nicolaides, et al, 1992). The nuchal translucency refers to the fluid at the back of the neck of the fetus at 11 - 13 + 6 weeks of pregnancy. It is confirmed to have improved the detection rate for DS significantly for a much lower false positive rate (Nicolaides, et al, 1992). Generally, innovations in medicine are not rapidly incorporated into practice in Nigeria, especially public institutions, where the majority of women patronize (Oloyede, 2008). The Nuchal translucency scan screening was recently introduced into clinical practice in Nigeria in 2006. The benefits of the Nuchal translucency scan are in many folds. Its use was also reported in South Africa (Naidoo, et al., 2008)

1. It is a first trimester procedure
2. It does not rely on too many personnel or logistics to conduct and interpret unlike the MSB
3. Other congenital structural abnormalities can be ruled out at same time

More recent innovations in Down syndrome screening such as the nasal bone, and fronto maxillary facial (FMF) angle are yet to be incorporated into practice in Nigeria.

2.1.1 Factors affecting uptake and utilization of prenatal screening

Prenatal screening for Down syndrome has been shown to be an effective tool to reduce the prevalence of Down syndrome. In countries such as South Africa, where it is fairly available, studies have shown a reduction in the prevalence of Down syndrome especially among the white population that utilize the screening service (Molteno,1997). Prenatal screening for Down syndrome is not routine in many antenatal clinics in Nigeria. Therefore, most women do not benefit from this service (Oloyede, 2008). This is unlike the practice in many developed countries, where screening is widely incorporated into antenatal programs (Rosch,et al., 2000)

Several studies on the Nigerian population about the uptake of prenatal screening and attitude of women to prenatal screening were reviewed. The factors are broadly classified as

- a. Health Service Factor
 - Skilled Manpower
 - Laboratory and Radiological Support
- b. Non Health Service Factors
 - Awareness
 - Cost
 - Legal status of abortion
 - Religion
 - Sociocultural

Health Service Factor

Skilled Manpower

Several factors are known to affect the utilization of antenatal screening service in Nigeria. The first and perhaps the most important is that of physician appreciation of the condition at risk. Oloyede in 2008 showed that the bulk of obstetrician and antenatal care givers in Nigeria underwent training curriculum that places less emphasis on knowledge and skill to undertake screening and diagnosis of genetic conditions such as Down syndrome (Oloyede, 2008). The same study shows that most (66.1%) obstetricians in Nigeria consider training in prenatal screening and diagnosis for Down syndrome either below average or just average, majority (89.3%) do not conduct routine antenatal counseling and almost all (92.9%) do not have a standard screening protocol. It was also showed that maternal serum biochemistry in the second trimester was favoured against the nuchal translucency screening in the first trimester of pregnancy. It is known that the ability to know what to do after screening is an encouraging factor to undertake screening procedures. The skill to undertake nuchal translucency screening can be acquired within a reasonable time by most physicians and is therefore a method that should be encouraged in Nigeria.

Laboratory and Radiological Support

Laboratory analysis of maternal serum hormones for screening for Down syndrome has its peculiarities. A major influencing factor is the race. One of the challenges involved in the laboratory support is the absence of reference standards for blacks. Laboratory analyses are done without standard references that are acceptable and reproducible. There are automated machines that are designed to generate values, compare with multiples of median and report immediately. Maternal samples from Nigeria and many other countries are analysed in South Africa or Europe until very recently. This has to do with the huge cost of analysis machines that is beyond the capacity of many centres in Nigeria. The implication of analysing in a distant centre is that it takes longer days to transport samples and delay the time of decision making. This delay could be more tolerable if there are local regional centres in the country that could as well conduct first trimester analysis.

Another important factor that could affect the utilization is that of availability of ultrasound scan. In Nigeria, ultrasound scan is available in many obstetric units and mostly utilized by pregnant women (Lamina, et al., 2004).

Non Health Service Factors

Awareness

Awareness about prenatal screening and diagnosis in Nigeria among both the health practitioners and public has been shown to be poor (Oloyede, et al., 2003, Oloyede, 2008). The role of health workers in the dissemination of appropriate information about prenatal diagnosis is very important (Oloyede, et al., 2003). This is what informs appropriate referral for utilization of screening service. Until after 2006, many health workers would refer women outside the country for prenatal diagnosis services or do nothing. Suggestions to improve awareness among the health workers include, incorporating education about prenatal screening and diagnosis into undergraduate education, wider dissemination of information in journals and scientific conferences and discussion in academic symposia (Oloyede, 2003).

Among pregnant women, there is a surprising good awareness of prenatal screening and diagnosis generally (Oloyede and Oyedele, 2008). Majority however, think only in terms of obvious structural defects that can be diagnosed through ultrasound scan. The traditional folk are also aware that advance maternal age is a risk factor to many fetal abnormalities and would therefore encourage early completion of procreation. Improvement in the awareness of Down syndrome screening could be achieved through information dissemination in local mass media in a manner that can be easily understood and appreciated. Information should address widely known preformed ideas and conceptions about the origin of conditions as well as the options of early diagnosis.

Cost

In Nigeria, cost is a major determinant of utilization of healthcare services. The majority of people live below poverty line and are struggling to cope with basic necessities of survival. Studies have demonstrated that the bulk (98.9%) of women that utilize prenatal diagnosis services are in the high socioeconomic group (Oloyede, 2005, Oloyede, 2008). Low utilization of prenatal screening was attributed to cost in 39.7% among other causes (Oloyede, 2008). Issues of subsidy from both governmental and non governmental agencies, bulk purchase of reagents for laboratory analysis are few suggestions to reduce cost (Oloyede, 2003). The introduction of scan screening would also reduce cost of screening in Nigeria.

Legal status of abortion

One of the controversial and ethical issues in Down syndrome screening is that of abortion. Most women that utilize prenatal screening have the issue of termination to contend with. In a study in Lagos, 70% of respondents would terminate a fetus that is affected (Oloyede, 2006). In taking a decision, few other factors may be important. Women with previously affected person in the family are much more likely to terminate than otherwise. Screening in the first trimester offers a better opportunity for safer termination than second trimester screening. Various associations such as the Down syndrome association of Nigeria and the Down syndrome South Africa are also cautious in the campaigning for or against abortion. This is because, it is viewed that the final decision is entirely a personal issue. Abortion law is restrictive in Nigeria and many other countries in sub Saharan Africa. Counseling following prenatal screening and diagnosis is largely non directional and leaves the option of termination in the hands of the couple.

Abortion following the screening and diagnosis of Down syndrome has been shown to reduce the prevalence of the condition in South Africa (Molteno, 1997). Considering the handicap in the care of the Down syndrome in the society in Nigeria and many other sub Saharan African countries, abortion may be a rational decision for affected cases.

Religion

Religion is sometimes a barrier to the wider utilization of prenatal screening services in Nigeria. It is responsible for poor utilization in about 28% of women in a Nigerian study (Oloyede and Oyedele, 2008). It is believed that the predominant religions in Nigeria are not well disposed to investigating the status of an unborn child, nor the decision to abort any fetus with genetic disorder (Oloyede, 2002).

The observation from Nigeria is that the influence of religion is modified by the occurrence of any previous congenital defect. Interestingly, another study in Nigeria showed that many

couples underwent prenatal diagnosis for Sickle cell disorder solely to be informed and prepare for the birth and care of an affected child (Olatubosun, 2000).

2.2 Supportive measures

Supports should be offered to all people living Down syndrome to optimize their chances of survival and quality of life. There are three levels at which these supports could be offered in an integrated approach to achieve the best effect

1. Family level
2. Community/Society level
3. Governmental level

Family level

The family system in Nigeria is more of extended rather than nuclear based. The benefit from such system is that family burdens can be shared. Unfortunately, it has the opposite effect in respect of children with Down syndrome. Consequently, many families would not share such information with other families. Indeed, it is as ridiculous in some situation as to necessitate hiding Down child from neighbours and visitors. The level of support from the family is perhaps the most important determinants of outcome in children with Down syndrome. The family should realize that love, affection and tolerance are the very key to successful outcome and perhaps this would help to discourage the usual habit of child abandonment. The children need special learning skills which have to be started very early from the home. This is because of the generally lower IQ compared with normal children. The coming together of families with Down syndrome children should be encouraged for exchange of knowledge and skill in the management of the condition.

Community/Society level

The society is to complement the roles of both the family and the government. This is better achieved through nongovernmental organizations such as the Down Syndrome Associations. In Nigeria, the Down syndrome Association of Nigeria serves as the main rallying point for all activities about Down syndrome. In particular, the body initiates and encourages the implementation of all measures that improves the survival of the Down syndrome child. Outside the body, few other organizations have come up as homes for the less privilege that try to address the challenges associated with Down syndrome.

A problem that is common to all these initiative is lack of societal support as well as governmental assistance to function optimally. In addition to this, many communities in Nigeria, patronize and discriminate against children with Down syndrome. Recently, there has been noticeable positive change in the societal attitude to Down syndrome. More public programmes are now organized to increase awareness about the condition and solicit for support in the care of those affected.

Governmental level

In Nigeria, major health policies and implementation are government driven. In particular, health problems that are considered rare in occurrence and impact are given little attention. The peculiar educational need of the Down syndrome child requires that special school curriculum is designed to optimize their potential.

The area of support would be:

Incorporation of health programmes that relates to prenatal screening and diagnosis of Down syndrome as well as screening and management of health complications into the national health system

Provision of subsidy to Down syndrome associations and other related bodies for better execution of programmes

Establishment of schools with special curriculum for those physically challenged by Down syndrome. This is considered more advantageous than educating them in regular schools in Nigeria

Organization of special events to draw attention to the plight of those with Down syndrome and to encourage community assistance. This could be in form of activities such as yearly marathon race

Design of social infrastructures that makes daily living easy for people with Down syndrome such as walk ways and bus lifts.

3. Conclusion

The up to date statistics about Down syndrome in Nigeria and many other sub Saharan countries is difficult to determine. However, empirical evidences from the few scientific publications available suggest that the condition is equally important and may even be more prevalent in the country than previously insinuated. The challenges faced in Nigeria include

1. Determine the exact magnitude of the problem
2. Early detection of cases through prenatal screening and diagnosis
3. Effective care for those affected in the population.

There are evidences to suggest that statistics can be updated in the next few years.

The last challenge is the most worrisome. This is because of the apparent lack of enthusiasm and slow response to initiatives to effectively care for the effected. In addition, there are inadequate facilities to adequately manage and optimize the survival and quality of lives of those affected.

Early detection appears the most feasible short term option in Nigeria. It gives the benefit of choice to the women. This approach involves both prenatal screening and diagnosis. Though not widely available, it has been shown to be favoured by many women. It will take a long time before a wide array of screening options become available in Nigeria. The cost, manpower and other logistics are prohibiting factors. In the meantime, the nuchal translucency scan holds the best promise for a wide scale screening. it is relatively cheaper to undertake and with less logistic required. Moreover a strong desire has been expressed by women for first trimester screening, with preference for the ultrasound scan (Oloyede and Oyedele, 2008). Efforts to improve on its availability and utilization would among other initiative involve review of training local curriculum to better emphasize prenatal medicine and collaboration between local and foreign postgraduate medical colleges for exchange and update of knowledge and skills.

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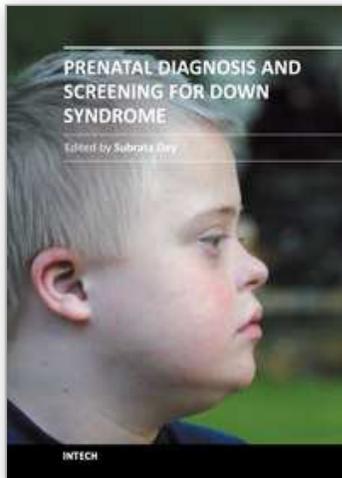
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This book provides a concise yet comprehensive source of current information on Down syndrome. Research workers, scientists, medical graduates and paediatricians will find it an excellent source for reference and review. This book focuses on exciting areas of research on prenatal diagnosis - Down syndrome screening after assisted reproduction techniques, noninvasive techniques, genetic counselling and ethical issues. Whilst aimed primarily at research worker on Down syndrome, we hope that the appeal of this book will extend beyond the narrow confines of academic interest and be of interest to a wider audience, especially parents and relatives of Down syndrome patients.

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