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RESEARCH ARTICLE

Medical Care of Infants with Down Syndrome in Sub-Sahara Africa; Challenges and the Way Forward

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ABSTRACT

Down syndrome is the most common chromosomal malformation, it affects multiple organs, necessitating a need for medical care for different systems. The care needed by infants with Down syndrome is complex and needs planning and coordination at different levels.

Medical services in Sub-Saharan Africa are not robust and face challenges in providing accessible and affordable health care.

We will review the situation of the medical care provided to infants with Down Syndrome, looking at diagnosis, counselling, and management within the Sub-Saharan context. Exploring the challenges and suggesting recommendations.

Introduction

Down syndrome (DS) is the most common chromosomal malformation affecting about 1:700 - 800 life born[1], DS the most common chromosomal condition associated with intellectual disability, and is characterized by a variety of additional clinical findings.

There is multi-organ involvement associated with DS, which makes an early diagnosis of DS important for the care of affected children[2]. Early interventions for abnormalities associated with DS, such as congenital heart disease (CHD) , hypothyroidism, and leukemia will prevent additionally irretrievable physical and mental disabilities[3].

Early diagnosis of DS will help in delivering the timely needed and effective medical care [4]. Early implementation of intervention programmes and support packages for the the affected children has been shown to improve their quality of life [2].

DS affects many organs spanning cardiovascular, central nervous system , gastrointestinal tract, endocrine and growth, respiratory, ear nose and throat, haematology oncology, Immune system, musculoskeletal system and the eyes [5]. The life expectancy of children with DS have seen a considerable improvements In the last few years in high-income countries (HICs), this has been mainly attributed to the recent advances in neonatal congenital heart surgery (CHD) [5]. Surgery for CHD in Sub-Saharan Africa is not well established and neonatal cardiac surgery services is not readily available in most countries in the region [6]. The inetgration of medical guidelines with preventative health care programmes in the care of children with DS will help improve their life expectancy and quality of life [7].

Epidemiology and Burden of Down Syndrome

Originally, there was a widespread belief that Down Syndrome is rare or non-existant among native Africans[8]. The report in 1955 by Luder and Musoke, from Uganda[9] was the first to draw attention to the occurance of DS among native African children; it is now known that the condition is not as rare as previously believed.

Most, if not all, Sub-Saharan African countries do not have an accurate record of the incidence of Down syndrome. This may be associated with the fact that the majority of residents reside in rural areas, with little or no adequate access to maternity care. Home delivery is still widely practised, and there is no accurate birth record; in fact, many children do not even have birth certificates[8]. Epidemiological studies from Sub-Saharan Africa about DS are few . The reported incidence of DS in Nigeria is 1:865 live birth [10], and a large study from South Africa investigating at the incidence of DS over twenty years period found an incidence of 1:671 live birth [11].

A systematic analysis of the global disease burden of DS from 2010 to 2019 revealed that although the rate of global DS disability-adjusted life years (DALY) and years of life lost (YLL) is stable, the years lived with the disability (YLD) rate is increasing. In addition, it reveals that the DS burden varies significantly based on geography; the burden was higher in Low Income Countries (LICs), including Sub-Saharan Africa. Down syndrome disability-adjusted life years (DALY) rate was mostly attributable to the years of life lost (YLLs) of children below the age of one year[12]. Building on these findings, we can see that the data suggests furure strategies should concentrate on low-income countries and DS-related deaths in children under the age of one year [12].

Diagnosis

There has been a recognition of the difficulties in recognising DS in Black African neonates[13]. This delayed diagnosis has a detrimental effect on the care of children with DS , and a study from South Africa showed only 16.4% of cases were diagnosed during the postnatal stay in hospital [13]. Another study from South Africa found that the average age of cytological diagnosis was around 13 months[4] and most of the of cases were clinically diagnosed. The same study showed the clinical diagnosis to be correct only in 55% of cases, while a study from the Netherlands found that the clinical diagnosis was correct in 83% of cases[14].

The absence of robust routine prenatal screening for DS in the majority of sub-Sarahran African countries and poor uptake of the few exesting programmes contributes to the delay in diagnosing the condition[15]. The practice of home deliveries is still widely practised in rural Africa [16], hence some infants with DS will have no contacts with or access to medical professional during the delivery and postnatal period; this contributes to the delay in the diagnosis of DS. Antenatal ultrasound screening can be an effective short term solution to antenatal diagnosis of DS in the absence of full antenatal screening program [17,18].

Counselling

It can often be daunting as a doctor to break bad news. And while it is one of the most difficult areas for the doctors to deal with, there is an unfortunate lack of adequate formal training in breaking bad news[19,20].

The coping of parents of infants with DS, with the unanticipated diagnosis of Down Syndrome, is affected by a variety of factors. This includes the time invertal between birth annd the disclosure of clinical suspicion of DS, the confidence level of the attending physician, and the time interval between the disclosure of clinical suspicion and the confirmation of karyotpe. Delays in diagnosis and initial uncertainty are harmful to parental coping. Premature communication of the news before the parents had time to spend with the infants is also detrimental to the parents coping[21]. It is beneficial to inform parents early, to be told about the prognosis, and to be connected with other families with affected children[22].

This initial experience may influence the way the parents react or accept the child's illness in the long run[19]. A study from Sudan, exploring the parents experience regarding the initial counselling , found that 36% of parents of children with DS received no counselling on the condition. 34% of those who received counselling had a negative experience about the counselling, and felt the quality of the counselling was not good as it failed to address their concerns and worries[19].

The majority of DS cases in Sub-Saharan Africa are diagnosed postnatally, and antanatal diagnosis of cases is only seen in 1-8% of DS cases in Africa[19,23] (19) (23). As a result, most of the counselling will be done by the paediatrician rather than obstetricians. There are no genetic services in most, if not all, of the countries in Sub-Saharan Africa. Home deliveries are still widely available in Africa [16] and as a result diagnosis may be missed or delayed and this will lead to poor or no counselling about DS.

Practically all the Sub-Saharan countries do not have their own guidelines for antenatal and postnatal counselling for DS, and although there are good international practice guidelines, such as recommendations of the American National Society of Genetic Counselors [24], these are not usually followed[15,20]. The problem with counselling for DS is not particular to Africa, studies from different parts of the world e.g. UK, Sweden, Australia, USA, have also reported strong parents dissatisfaction with the way in which the diagnosis was conveyed to them during the immediate postnatal period [25,26].

Health professionals' verbal and nonverbal communication has a potentially lifelong impact on parents[21]. Counselling for parents with children with DS need to be improved, this issue should be addressed by issuing local guidelines for the care of children with DS, and by formal training to paediatrician in counselling skills and in delivering bad news[20].

Training programs for obstetricians [27] and paediatricians [28] in counselling for DS have been shown to improve the counselling skills and their perceived comfort level for delivering a prenatal diagnosis or postnatal diagnosis respectively. Similar courses could improve the quality of counselling and hence the care received by children. The effect of uncertainty of the diagnosis in the first few days after birth is considerable. This have to be balanced with raising the clinical suspicion of Down Syndrome as soon as possible. Confirmation of the clinical suspicion of Down Syndrome should be done as soon as possible. Promoting parent–infant bonding and normalising postnatal care should be encouraged in the postnatal care[21].

The majority of countries in subsaharan Africa, do not have proper genetic services and the counselling is done by the paediatricians. Counselling of parents who have DS children is very important for the long term care of the children, so it must be done in a sensitive and a compassionate manner (20). Parents should be provided with up-todate written information about Down syndrome and information about peer support while they are still in hospital[21]

Training programs in DS counselling for obstetricians[27] and paediatricians[28] have been shown to improve counselling skills and perceived comfort level for delivering a prenatal or postnatal diagnosis, respectively. Similar courses could improve the quality of counseling and, as a result, the care provided to children.

The impact of diagnosis uncertainty in the first few days after birth is significant. This must be balanced against the need to raise the clinical suspicion of Down syndrome as soon as possible. The clinical suspicion of Down syndrome should be confirmed as soon as possible. Postnatal care should encourage parent-infant bonding and normalisation of postnatal care[21].

The majority of Sub-Saharan African countries lack proper genetic services, and genetic counseling is provided by paediatricians. Counseling parents with DS children is critical for the long-term care of the children, so it must be done sensitively and compassionately[20]. While their child is in the hospital, parents should be given current written information about Down syndrome as well as information about peer support[21].

Reduction of DS cases can be achieved by social uplifting, increasing awareness of the public and especially women about the increased maternal age as a risk factor for having a baby with DS.

Women should be encouraged to start their families in a younger age and women with advaned age should be advised to use appropriate family planning measures to reduce such risk [1,29].

Such measures have been shown to reduce the incidence of DS in high income countries, through reducing the mean maternal age[30].

Termination of pregnancy has not shown to be an effective measure to reduce the incidence of DS in Africa, as most pregnant African women tend not to accept termination of pregnancy for antenatally diagnosed cases; due to religious and cultural beliefs[30].

Care & Support

There are well established international guidelines for the medical care and support needed for children with DS [7,31].

The American Academy of Paediatrics (AAP) revised their DS clinical guidelines in 2011[31] focusing on improving the level of functioning and quality of life of children with DS through screening for the associated medical conditions [32].

All the countries in Sub-Saharan Africa do not have a nationally recocnised guidelines for health provision of children with DS. Children with DS are missing essential health care because International or local guidelines are not followed [19,32]. South Africa adopted the AAP guidelines[31] , study from South Africa looking at the care of DS using AAP guideline as a standard of care, found that the diagnosis and screening test were not achieved within the time scale advised by the AAP , and suggested that a local and resource-appropriate guideline is needed[32].

Early social and educational Support for families with children with DS is essential for the development and integration of people with DS into the society[1]. Support group like DS organisation are not well established in african countries , such groups can have a major role in increasing the awareness about DS and in cordinating the medical, educational and social care of children with DS at the policy makers level [8].

Health services in sub saharan Africa is not well organised and there are problems with awareness , affordability and access especially for the poor and rural population[33]. There is an urgent need to raise awareness of DS among medical and nursing staff as well as in communities, particularly in hospitals and clinics that lack specialized care and supervision[4]. A designated health care worker, who can assist the parents with coordinating and getting the needed care and could help provide continuity of care for families with an infant with Down syndrome[21].

Early detection and intervention, as well as ongoing medical monitoring, are critical for minimizing disability and maximizing potential in all children with Down syndrome. These interventions not only save money but also improve the quality of life for the child and his family. To ensure the health of children with Down syndrome, paediatricians should follow health supervision guidelines[34]. All stakeholders involved in the care of children with DS, including medical, social, and educational professionals, should be included. To be effective and sustainable, such care must be well coordinated.

In the absence of genetic services, Down syndrome clinic should be promoted as it can provide and cordinate all the medical services needed by the affected children such as cardiac, respiratory,endocrine , opthamology. A Down syndrome specialty clinic can identify and address many healthcare needs of children and adolescents with Down syndrome that go beyond what primary care or secondary care physicians can provide [35].

Conclusion and recommendation

Reliable epidemiological data about Down Syndrome in Sub-Saharan Africa is missing. The limited available data suggest that the burden of the of DS in Sub-Saharan Africa is higher compared to the high income countries. Further research about the prevalence of DS in the region is needed , this should increase the awareness about the condition among the health professionals and policy makers. Antenatal screening is importanat especially for high risk groups such as advanced maternal age. All countries should ensure having lacal guidelines for counselling and for the care of DS.

Counselling of parents for DS need to be improved, this issue should be addressed by issuing local guidelines for the care of children with DS and by formal training to obstetrician and paediatrician in counselling skills and in delivering bad news. Counselling of parents who have DS children is very important for the long term care of the children, so it must be done in a sensitive and a compassionate manner.

Antenatal ultrasound screening for DS can be an effective antenatal diagnosis measure especially in the rural settings, genetic services should be develop to improve the care of DS and other genetic disorders.

The lack of availability and accessability of specialised health services in sub-Saharan Africa such as cardiac surgery is detrimental to the quality of life and care received by children with DS. Estbalishment of DS associoations will improve the awareness of DS and the care received.

Down syndrome multidisciplinary clinics should be promoted to improve the accessability to the medical care. Involvement of all the stakeholders

such as medical, social and education is vital for the care and the good quality of life for DS children.
Countries should establish their own guideline for the care of children with DS, taking into consideration the local context, the guidelines should provide advise on diagnosis, health screening and medical care and it should also provide for referral to social,educational and support group.

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