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DOWN SYNDROME CHALLENGES AND MANAGEMENT STRATEGIES; THE PREVENTIVE APPROACH

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Abstract: Down syndrome constitutes one of the congenital genetic challenges affecting human being globally. Although not recognized as an illness but substantially affect a person's physical and intellectual capabilities and also could increase the likelihood of some health problems and over all development hence the need to explore ways and means of assisting individuals living with it. The identified cause of the condition is believed to have strong link with chromosomal defect resulting in conditions such as intellectual disability, heart problems, auditory and visual problems, gastrointestinal problems and other conditions such as autism, Alzheimer diseases and thyroid abnormalities. The physical characteristics of Down syndrome include: flabby muscles, slant eyes, skin folds and white spots on the iris, small stature with short neck, protruding tongue, flat nasal bridge and large space between the large and second toe. Besides, Down syndrome patients present other physical and psychological problems with its attendant consequences. The reported preventive management strategies include; dietary intervention through supplementations, regular exercise participation targeting cardiovascular efficiency and other alternative approaches. Conclusively, Down syndrome is a genetic defect and patients could be assisted with the identified management strategies to assist the individual function effectively within the limit of his or her capabilities. It is recommended among others that a survey be carried out immediately to ascertain the incidence ratio in Nigeria while government through the ministry of health should come up with a comprehensive health policy for special people targeting Down syndrome patients.

Keywords: Down syndrome, Management, Prevention.

Introduction

One of the several genetic challenges affecting man congenitally is Down syndrome (DS). Down syndrome is a genetic condition that occurs when there 1s an extra copy of a specific chromosome: that is chromosome 21(Crosta, 2017). Generally it is not recognized as an illness but a term describing the features resulting from this chromosome can affect a person's physical features, intellect and overall development and it could also increase the likelihood of some health problems (Freely & Jones, 2006; Crosta, 2017). Historically, the name Down syndrome was originally called Mongot by one Dr. John Langdon Down sometime in 1866. The name however, did not go down well with people who have DS and their advocates who understandably were sensitive about the words used to describe this chromosomal condition. The chromosomal basis of DS was identified in 1959 which resulted in a gradual trisomy 21 as being a variation of normal has done a lot to remove some of the disability and uniformed debates Over the humanity of people with Down syndrome (Trumble,2012).

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The global incidence of DS is not robust in literature. The few reported cases show considerable variations across the globe. DS has a prevalence estimated at 1 in 700-1000 live births and these individuals are living longer into adulthood than ever before. In England and Wales, the prevalence ratio is 1:89 per 1000 live births In Australia, DS is identified in 1:1150 live births. In the absences of parental diagnosis and therapeutic abortion, the prevalence of DS in developed countries is 1-2 per 1000 live births making it the most frequent identifiable cause of sever learning difficulty. Advances in maternal is by far de strongest of epidemiological variables with birth prevalence increasing from 0.6 to 4.1 % per 1000 between age 15 and 45 (Cuckie, Walds &Thompson, 2007). Besides, there is familial aggregation: having had a previous DS pregnancy confers a risk 42 per 1000 higher risk than the age specific prevalence (Arbuzova, Cuckle, Maller & Schmi, 2011).

Down syndrome is the commonest identifiable cause of intellectual disability accounting for almost 1/3 of cases. It occurs equally in all races with an overall incidence of approximately 1 in 800 births. This is much lower than the actual conception rate due to a high incidence of spontaneous and surgical abortion (Australia, Bureau of Statistics, 2009). The increase in incidence with advancing material age is well-known but what is not commonly realized is that most children with DS are born to mothers who are less than 30. This is due to greater number of pregnancies in this age group (Australian Bureau of Statistics, 2009). One study however, observes that before the age of 30 it occurs in fewer than in 1000 pregnancies and that after the age of 40, this figure rises to about 12 in 1000 (Down Syndrome Association of Victoria, 2011). These statistics regarding the incidence ration point to the fact that DS is of public health concern and has known causes.

Causes of Down Syndrome

Down syndrome causes could be linked to its genetic origin. All cells in the body contain genes that are grouped along chromosomes in the cell nucleus. There are normally, 46 chromosomes in each cell; 23 inherited from the mother and 23 from the father. When some or all of a person's cells have an extra full or partial copy of chromosome 21, Down syndrome occurs (Andensome, 2011). This singular identified cause of DS manifest with several characteristic among the individuals suffering it.

Characteristics of Down Syndrome Individuals

Individuals with DS presents certain identifiable characteristics which could result in certain disease conditions. People with Down syndrome often have associated medical conditions that are either present at birth or develop over time. Doctors can manage these conditions with medications or other care. Common conditions of Down syndrome include: Intellectual disability (Low 1Q), Heart problems, frequently present at birth, Thyroid abnormalities, Hearing and vision problems, Gastrointestinal problems, like constipation, gastro esophageal reflux (when fluid from the stomach re-enters the esophagus and ceilial disease (intolerance for wheat protein), Autism, with challenges with social skills, communication and repetitive behavior, Alzheimer's disease, which cause memory and thinking problems in older age (Cleveland Clinic, 2020).

Muscular and orthopedic anomalies are well known in DS. For example flat foot, patella instability main causes of working problems and sever static troubles such as scoliosis and kyphosis postural problems (Scola & Pezzullo, 2010). An increased risk of dislocation exists after cervical traumas or abrupt head movements with neurological complications occurring by cervical cord compression. Dislocation can also produce a condition of quadriplegia with incontinence which can be preceded by head tilt, abnormal staggering gait and emergence of neurological signs (Cremers, 2013). Sexual

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maturation is similar to that of the general population for both sexes. However, menstrual pause occurs -early in females. Fertility ratio is reduced in females; only a small number pregnancy has been observed. Males are almost invariably sterile (as only a cases of fatherhood have been reported (Aicardi, 2012).

Down syndrome individuals present anatomical abnormalities, mental and orofacial problems that present a large impact in quality of life (Shyama Al-Mutawa & Honkala, 2010). Furthermore, DS patients are more susceptible to infections including an increased prevalence of periodontal diseases, almost 100% under the age of 30 years, present mild to moderately reduced T and B cell counts, suboptimal antibody responses to immunization, decreased immune globulin A in saliva (Ram & Chinen, 2011). A study found other conditions that patients with DS present to indicate advanced tongue position periodontal disease (dental diseases), an expression of immune related gene in children with DS (Zampieri,. Selli-Perico, de Souza, Burgen, Silva & Goloni-Bertollo, 2014).

People with DS frequently present alterations in the development, structure and functions of several organs or systems that can lead to complications sometimes sever for their health and determination in their quality of life. Many of these complications can be prevented if they are kept in mind and managed at early stage, with the correct application of a specific programme of truly prevention medicine. They are easily identified by certain physical characteristics. Some distinct physical health issues and variability in cognitive development are obvious. The physical features include: eyes that slant upward, have oblique fissures, skin folds on the inner corner and have white spots on the iris, Low muscles tone, small stature and a short neck, A flat nasal bridge, Protruding tongue., A large space between the large and second toe, A single flexion furrow of the 5th finger (Anderson, 2011).

Developmental delays show that people who have DS usually have cognitive development profiles that suggest mild to moderate and intellectual disability. However, cognitive development and intellectual ability are highly variable. They often reach developmental milestones later than their peers for example they may be delayed in learning to talk. Fine motor skills may also be delayed. They can take time to develop after the child acquires gross motor skills (Crosta, 2017). On the average, a child with Down syndrome will Sit at 11 months, Crawl at 17 months walk at 26 months (Crosta, 2017). There may also be problems with attention, a tendency to make poor judgments and impulsive behavior. Most people with DS can attend school and become active valued members of the community

Sometimes there are general health problems that can affect any system or organ, heart defect. There may also be higher risk of respiratory, hearing difficulties, childhood leukemia, epilepsy, thyro1d conditions, however, there seem to be a lower risk of hardening of the arteries, diabetes and most kinds of cancers (Crosta, 2017). On the general outlook, a person with DS can do many of the things that ordinary people do. Children may take longer to acquire skills such as walking and talking but with such stimulation, they can acquire key life skills and attend school up to college. Depending on how the condition affects a person ,it often possible for someone to work and live semi-independently with DS. These individuals constantly need friendships. Some will live with a partner or get married and possibly have an independent life. This however, could be dependent on management strategies adopted.

Challenging behavior is a common occurrence. Children with DS are often described as stubborn and obstinate. In fact, references to challenging behavior have historically been seen in the clinical literature and continue to exist today for example children with DS show high rates of attention problems, social withdrawal, noncompliance and compulsions (Such as arranging

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objects and repeating certain actions) as well as high rates of self-talk (Feeley & Joncs, 2006; Dykens & Kasari, 2007). With increasing age, they may be having associated anxiety, depression and withdrawal will also increase (Dykens & Kasari, 2009).

Several other factors, specific to children with DS including sleep disorders and increased incidence of illness may also increase the likelihood of challenging behavior in children with DS (Feeley & Jones, 2006). As the child grows through the pre-school years, it is common to have development delayed, physical milestone will be delayed due to joint weakness, speech is likely to be difficult and socialization may be delayed (Anderson, 2011). Further evaluation shows that most children with DS have intellectual functioning in the moderately disabled range but the range is enormous (Down syndrome Association of Vitoria 2011). There is significant hearing impairment in the majority of children with DS, visual impairment is present and sometimes could develop to cataracts. The teeth of children with DS tend to be small, irregularly spaced and misshapen.

Psychologically, DS patients show temperament and are often frustrated in their expression, their skin tend to be dry and susceptible to eczema (Down syndrome Association of Victoria, 2011). There are peculiar problems which sometimes hinder effective management of individuals with DS. For instance, these individuals still experience significant barriers to the receipt of high quality healthcare and health outcomes compared with those of general population. Additional problems include that they are at increased risk of range of physical and mental health conditions, often live multiple unrecognized health issues and have limited access to disease prevention and health promotion interventions (Korista & Lacono, 2011). Mental health disorders such as depression, Obsessive compulsive disorder, abuse and conduct disorder occur frequently than other mental health disorders in individuals with DS to differentiate between dementia and depression. The Dementia Screen for DS (DSDS) is a validated scale of adults (Fenech, 2011). Currently, this scale is only available for use by licensed psychologists; other scales numbers of adults with DS, but the Down syndrome Digital Scale (DSDS) appears to have the greatest reliability.

Adults with DS can develop health problems normally caused by aging. These include: dementia, memory loss, high cholesterol, diabetes, cataracts, and early menopause (American Academy of Family Physician, 2019). The focus of this paper is to present possible preventive strategies that could probably reduce the impact of the health condition so that individuals living with DS could function with the limit of capability. However, preventive methods sometimes present disappointing outcomes in this group of patients, requiring special approaches. These factors justify the necessity of further research efforts.

Management Strategies of Down syndrome

There is no single standard management for Down syndrome. Management is based on each individual physical and intellectual needs as well as his or her personal strength and limitations. It has been suggested that management strategies of diet and exercise should be realistic, comprehensive and adaption to the person living and employment situation where necessary. There may be factors that people with DS have to overcome that are not necessarily present in the wider population for example lower metabolic rate, less Physical activity as a result of lower muscle tone and delayed development and hormonal conditions. Recreational social activities can provide the benefits of exercise without feeling too much like hard work. Building regular family walks with friends into the week can help them. Social activities such as dancing or drama groups can be really good motivators for people with DS. Exercise Digital Video Disks (DVDs) particularly those featuring a favorite actor or television (TV) personality are enjoyed by many adults (Chicoine & MeGuire, 2010).

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Awareness of increased DS risk means an individual couple may decide to avoid pregnancy at advanced age. This raises the possibility of a public health strategy based on routinely informing couples of their age specific in a family planning context. It is not possible to judge how effectiveness this might be in practice but a theoretical maximum can be estimated for the prevalence if all families are coupled before different ages. These strategies could be further enhanced with good dietary supplementation.

Supplementation

Dietary intervention studies show that genomic instability is minimized when the plasma tolerance level exceed about 34 nmol and Hey level is less than 7.3 umol (Fenech, 2011). These levels can only be achieved when folic acid intake is above 5mg per day. Currently, the recommended daily dose for the prevention of neonatal DS in women with pervious affected pregnancies is 0.4mg but it has now been estimated that higher intake would be required to have a substantial benefits und the authors recommend, 5mg (Wald, Law, Morris & Wald, 2011), In author publication, 5 women with foliate deficiencies who were given 10mg per day for 2 months, showed an average of 53% reduction in plasma Hey (Zittoun, Tonetti, Bories, Pigion & Tulliez, 2008).

The importance of a well balanced diet should be emphasized, as is wearing in due time. This may result in the control of growth retardation since linear growth retardation in characteristics of DS. Statue is generally stabilized at minus 2-3 standard deviations on normal growth charts. Special growth charts for children with DS are also available (Myrolyed et al, 2012). The mechanisms responsible for short stature have not yet been completely explained, but multiple causes such as malabsorption, congenital heart disease may be responsible. There is also the use of hormone therapy for growth enhancement. The notable hormone therapy is insulin like growth factors (IGF). These hormones have been found not only useful for body growth, but also for the development and maintenance of nervous system as low levels of these hormones have been found in children with DS (Rasore- Quartino et al, 2007). Also intestinal malabsorption in DS is responsible for intestinal disturbances and for growth retardation in some children. Ghuten which is a component of wheat, rye, barley, and other cereals have been identified as having high intolerance among 1ndividuals with DS with prevalence in normal population 0.43%,, in DS.it is 6% (Bonamico, Rasore -Quartino et al, 2011).

Intestinal malabsorption is usually very sever::it usually develops in early childhood after the introduction of gluten into the diet (approximately at 6mouths) manifests itself with diarrhoea, bulky stools prominent abdomen and poor, thriving. Presently, more frequently moderate or atypical forms are found, appearing late childhood or in adolescence and showing scare or absent intestinal symptoms (Marsh, 2012). These diet strategies are part of rehabilitation programmes.

The tendency to obesity among DS individuals is obvious. It manifests itself mainly in young adults. Therefore, it is important to establish a correct prevention from childhood, with particular attention to adolescence. This implies that intervention in nutrition and physical activity (PA) is critical. The assessment of caloric intake should be regularly carried out. Excess of glucose and lipids-rich food should be avoided. Protein rich food should be preferred, but in a well balanced diet (Lowe, 2009). Poor management of diet could result in dental anomalies.

Dental anomalies is a common problem, the situation to which is not an easy task. A peculiar oral and dental anatomy, developmental anomalies are frequent. On the contrary, carries seems to be less frequent than in normal persons. If oral hygiene is poor, gingivitis and periodontal disease are likely to occur leading to early and total tooth loss. Dental checks should be constant from childhood and for the whole life. Accurate dental checks should be available in order to avoid the

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depressing consequences of dental decay (Lowe, 2009). Muscular and orthopedic anomalies are well known in DS. For instance flat foot, patella instability are main causes of walking problems. There are other postural problems such as scoliosis and kyphosis. Prevention is necessary and is achievable through early and correct mobilization and an active life associated with sport activities (Smith 2012). An increased risk of dislocation exists after cervical trauma as or abrupt head movements, with neurological complications occurring by cervical cord compression. This is preceded by head tilt, abnormal staggering gait and emergence of neurological sign (Cremers et al 2013). Resistance trainings targeting the lower body muscles such as leg press, leg extension, leg curl and squat could be helpful for stability. The threshold however, should be low to avoid injuries to the muscles and joints already adjudged weak due to existing conditions.

Summary and Conclusions

Based on the review of Down syndrome challenges and management strategies by means of preventive medicine, the following conclusions are made:

1. Down syndrome is not an illness but a chromosomal defect.
2. The prevalence ratio is low in developed societies while the statistics in developing countries to the best knowledge of the researchers is nonexistent.
3. Down syndrome incidence increases with maternal age suggesting early procreation by couples possibly before 30 years of age.
4. The condition comes with multiple health conditions such as low LQ auditory and visual impairments, gastric abnormalities, cardiac problems, muscular and orthopedic anomalies, postural defects as well as skeletal and neurological complications, mental health disorders such as depression and anxiety.
5. The physical characteristics of Down syndrome individuals include: eyes that slant upwards, small stature and short neck, flabby muscles, white spots on the iris, protruding tongue flat nasal bridge and others.
6. On management strategies; the preventive approach, light exercises such aerobic exercises at moderate intensity, light resistance training, dance activities, cycling and swimming are excellent for the efficiency of the cardiovascular system. However, the exercise load should at all times be kept at low threshold.
7. Dietary management strategies include good nutrition with emphasis balanced diet should also be inclusive of calcium (ca) and iron (fe) for building strong bones and teeth. There is also the need for alternative therapies such as supplementation with folic acid, gluten which is a component of wheat and cereal. On the other side, excess glucose and lipid-rich food should be avoided as poor dietary management could result in dental anomalies.

Recommendations

Arising from the conclusions of the study, the following recommendations are made:

1. There is the need for a national survey to determine the prevalence ratio in Nigeria because there are numerous DS individuals in our society.\
2. The study has successfully established that with proper management, individuals with DS can function optimally within the limit of capabilities. Recreation arena should be created and equipped with exercise equipment at the rehabilitation centres to enhance their physical fitness status.
3. The high incidences of DS with maternal age mean that intending couples should begin and end procreation on or before the age of 30 years.
4. The dietary arrangement for Down syndrome individuals should be nutritious with plenty of balanced diet. Diet should also be inclusive of Ca

and Fe for the formation of strong bones and teeth.

5. Supplementation with folic acid and gluten and other cereals should form part of the diet plan for DS individuals.
6. Government through the ministry of health should come up with comprehensive health policy for special people targeting DS individuals in our society.

References

ACSM, (2014). Plhysical Activity Readiness Self-Assessment Guidance. Available from http:/www.nasin.org /does/default source/PDF/nasm- par- g (pdf-21k) pdf, accessed on 27" January, 2020.

American Academy of Family Physicians(2019). Down Syndrome. Avaliable from https://familydoctor.org/condition/down-syndrome retrieved on 26 December, 2019.

Bonamico, M., Marcani, P., Danesi, H.M., Crisogianni, M., Failla, P., Gemme, G, Resore Quartino, A., Gianohi, A et al (2011). Prevalence and clinical picture of celiac disease in Italian Down Syndrome Patients: a multicenter Study. *J Pediatric Gasstroenetrol Nutr. 2011;* 33:139-143

Cremers, M. J. G., Bol, E, de Roos, F., Van Giin, J., (2013). Risks of sport activities in children with down syndrome and atlantoxial Instability. *Lancent 2013*, 342:511-514.

Daskapan, A., Tuzun, E.H.,& Eker, I.,(2000). Received barriers to physical activity in University Students. *Journal of Sports Science and Medicine* 5:615-620.

De Beurde audhuji, I., philippaerts, R, Crombez.G., Matten, I.. Wijiodacle.K. Balduck A,& Lefevre, J., & Lefevre,J.(2005). Stages of change for PA in community sample of adolescents. *Health Education Research*, 20(30):116-124.

Fenechi, M. (2011). Recommended dietary allowances (RDAS) for genomic instability. *Mutat Res.* 2011:480:51 -52.

Lindsay-Reid, E& Osborn, R.W., (2008). Readiness for exercise adoption. *Soc Sci Med Psychol Med Social 2008*,14: 139-146.

Marsh, M.N., (2012): Gluten, major histocompatibility complex and the small intestine.A molecular and immunobiologic approach to the spectrum of gluten sensitivity (Celiac sprue). *Gastroenterology 2012*, 102:350-354.

Myrolid, A., Gustafsson, J., Ollars, B., Anneren G.(2012). Growth charts for Syndrome from birth to 18years of age. *Arch Dis Child*, 2012, 87:97-103.

Prezi User, (2013), Assessing the body's readiness for exercise. Available from https/prezi.com/18hdq-q2/assessing-the-body-readines, accessed on 27 January,2020.

Rasore- Quartino, A., Cominetti, M. (2007). Clinical Follow-up of adolescents and adults with Down syndrome. In Nadel L., Rosenthal. D. (eds.): Down, Syndrome: Living and Learning in the Community. Wiley-Liss, New York. 2007 pp238-245.

Smith, D.S., (2011). Healthcare management of adults with Down syndrome: Preventive approach. *Amer Family Phys. 2011*, 64:1031-1038

Stroliacker, K. Boyer, W.R., Smitherman, E.C., & Fazzino,D..(2017). Assessing energy level as a marker of aerobic exercise readiness: A pilot investigation. *International Journal of Exercise Science*, 10(1) 62-75

Wald, N.J., Law, M.R. Morris, J.k. & Tulliez, M., (2008). Plasma hormoccysteine level related to

interactions between folate status and methylenetetrahydrofolate reductase: a study in 52 healthy subjects. *Metabolism 47*(11):413-418

Wald, N.J., Law, M.R. Morris, J.k. & Wald, D.S. (2011). Quantifying the effect of folic acid. *Lancet 2011*; 358:2069-2073