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REVIEW ARTICLE

A REVIEW OF TRISOMY 21 AND ITS ORAL HEALTH IMPLICATIONS

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ABSTRACT

Down syndrome (DS) or trisomy 21 is one of the most common chromosomal abnormality associated with intellectual disabilities. It is characterized by delayed mental and physical development. With time increase in awareness about general and oral health of DS individuals has been reported among the parents and society. But still the difficulty in obtaining dental care due to disability, accessibility and dentist related factors seen as the barriers to utilization of oral health care services. Hence individuals with DS have poor oral hygiene, significant caries experience and barriers related to utilization of oral health services.

INTRODUCTION

Good mental health is important to deal with varied experiences of life. There are conditions which affects the mental health of individuals ranging from mild intellectual impairments to profound physical and intellectual disabilities (Lee, 2009). In particular children with special health care needs are at increased risk for physical and mental conditions and also require health and related services of a type or amount beyond that required by children generally (Butani, 2009). When treating individuals with Intellectual disabilities, particularly those with serious conditions, paying attention to oral health is important because it is often neglected (Emmanuelle, 2017). This review aims to highlight the problems faced by Down syndrome children and to suggest the appropriate solution for same.

Intellectual Disabilities

Intellectual disability is also referred to as developmental disability. It includes ASD (autism spectrum disorders), epilepsy, cerebral palsy, developmental delay, foetal alcohol syndrome and other disorders that occur during the developmental period. The major differences are in the age of onset, the severity of limitations, and the fact that a person with a developmental disability may or may not have a low I.Q. People with intellectual disability may have slight difficulty thinking and communicating, another may face major challenges with basic self-care and physical mobility. Children with ID may take longer to learn to speak, walk and take care of their personal needs, such as dressing or eating. As adults, some are able to lead independent lives in the community without supports. A small percentage will have serious, lifelong limitations in functioning (https://www.thearc.org/learn–about/Intellectual–disability). According to World Health Organization (WHO): Intellectual disability (ID) is a disorder defined by the presence of incomplete or arrested mental development, principally characterized by the deterioration of concrete functions at each stage of development and that contribute to the overall level of intelligence, such as cognitive, language, motor and socialization functions; in this anomaly, adaptation to the environment is always affected (WHO, 2017 and Katz, 2008).

The problem: ID comprises a substantial section of the community and it is estimated that there are about 500 million people with intellectual disabilities worldwide (WHO, 2017 and Katz, 2008). The problem presents more challenges in emerging economies around the world in terms of delivery of care to these children (Lehl, 2013). Intellectual disability (ID) it not only affects the people who suffer from it but also the family and society as a group (WHO, 2017 and Katz, 2008). Studies published during 1980-2009, has estimated ID to be 10.37 per 1000 population with higher rates in low-middle income countries and in children-adolescents as compared to

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adults. It is estimated that about 500 million people suffer with intellectual disabilities worldwide (Lehl, 2013). Each year 3000 to 5000 babies are born with Down syndrome with 30% increase in last 25 years worldwide. In India, annual birth of Down syndrome babies is around 37,000 taking incidence of Down syndrome as 1.4/1000 live birth (Sharma, 2013).

Down Syndrome: Most common ID is Down syndrome that occurs in 15 of every 10000 births. Down syndrome (DS) is the most frequent chromosomal disorder manifested in newborns worldwide also referred as Trisomy 21, Trisomy G or Mongolism because it is caused by Trisomy of 21st chromosome and Mongolian features (Asokan, 2008). All individuals with DS are mentally impaired to some degree, ranging from mild to severe (Cheng, 2017 and Desai, 1997).

Down syndrome is the most common intellectual disability with the birth ratio of 1 in 600 to 1 in 1000. It is a type of mental retardation caused by extra genetic material in chromosome 21. This can be due to extra chromosome (called trisomy 21) because of non disjunction. The cause of which is not known, although it correlates with a woman's age reference. The additional material present influences development and results in the state known as Down syndrome (WHO, 2017 and Desai, 1997).

Historical aspects (Desai, 1997): The first description of a child who presumably had Down syndrome was provided by Esquirol in 1838. Seguin in 1846 described a patient with features suggestive of an anomaly that was later known as Down syndrome. It was John Langdon Down in 1866 who accurately described some of the characteristics of this syndrome. The exact cause that Down syndrome was caused by Trisomy 21was independently determined by Le Jeune and Jacobs in 1959. Nebuhr (1974) suggested that the "Down syndrome phenotype" might be caused by the duplication of only a part of chromosome 21band q22, which itself represents about one half of the long arm.

Classification (ICD, 1996)

Down syndrome is classified by WHO under mental retardation among congenital abnormalities, deformation & chromosomal disorders[12] which is further divided based on etiology into following: Q90.0 Down's syndrome

Q90.0 Trisomy 21, meiotic nondisjunction
Q90.1 Trisomy 21, mosaicism (mitotic nondisjunction)
Q90.2 Trisomy 21, translocation

Systemic conditions associated with Down syndrome (WHO, 2017 and Desai, 1987):

Persons with DS are often short with a short neck and underdeveloped or hypoplastic mid-face giving rise to a sloping forehead and a flat face, with outer canthus of the eye higher than the inner giving rise to slant-eyes appearance. Affected individuals have greater risk in acquiring many systemic conditions such as upper respiratory tract and chest infections. Approximately 50% have some forms of heart defect, usually ventricular septal defect; some may require antibiotic cover for invasive dental treatment (Cheng, 2017; Desai, 1997; Roizen, 2003; Jain, 2009). Some of the common systemic conditions associated with DS are as follows:

Cardiovascular: Ventribular septal defects, A/V communis, Patent ductus arteriosus, Mitral valve prolapsed.

Hematopoietic: Impaired immnnity, Defective short-lived neutrophils, Risk of lymphopenia, Risk of eosinopenia, Cell-mediated immunity impaired and irregular serum immunoglobin patterns. Increased risk of leukemia and increased risk of hepatitis B carrier status (if previously institutionalized).

Musculoskeletal: Atlantoaxial instability, underdeveloped midface with relative, prognathism, narrow and partially obstructed nasal air, passages and thickening of mucosa, temporomandibular joint dysfunction, platybasia.

Nervous: Motor functions delayed; affects coordination, dementia analogous to Alzheimer's disease, Speech, delayed Expressive language, distorted phonation.

Behavior: Natural spontaneity, genuine warmth, gentleness, patience and tolerance, anxiety and stubbornness in few cases.

Down syndrome and oral health (WHO, 2017; Desai, 1997; BSDOH, 2001 and Bernal, 2005): Several dental characteristics have been described in children with Down syndrome. Some of these are macroglossia, fissured tongue, underdeveloped maxilla, tongue thrusting, congenitally missing teeth, malocclusion, high arch palate, increased salivation and microdontia (Asokan, 2008 and Oredugba, 2007). Periodontal disease is the most common oral health problem in people with Down syndrome (Oredugba, 2007; Al-Sufyani, 2014 and Zizzi, 2014). Some may have increased risk of periodontal disease due to cariogenic food choices and reduced food clearance from the mouth (Oredugba, 2007 and Al-Sufyani, 2014). Down syndrome individuals need assisted care from parents or caretakers for daily oral care because of lack of manual dexterity (Al-Sufyani, 2014) and hence high prevalence of above oral conditions (Zizzi, 2014; Hennequin, 2000). All these conditions remain static or may increase with age (Cheng, 2017; Roizen, 2003; Jain, 2009; Oredugba, 2007; Al-Sufyani, 2014).

Common oral conditions associated with Down syndrome are as follows:

Oral opening - Angle of the mouth pulled down (result of hypotonic musculature), Lower lip are chapped and everted (result of hypotonic musculature), Mouth breathing with drooling, Angular cheilitis.

Palate - "Stair palate" with "v" shaped high vault, Soft palate insufficiency.

Tongue - Scalloped, fissured, protrusion and tongue thrusting (result of hypotonic musculature), macroglossia (result of small oral cavity), desiccated tongue (result of mouth breathing).

Dental – Microdontia, hypodontia, partial anodontia, supernumerary teeth, spacing, taurodontism, crown variants, agenesis, hypoplasia and hypocalcificafion, reduced risk of dental caries, delayed eruption, bruxism, occlusion malalignment, frequent malocclusions.

Periodontal - Increased risk of periodontal disease.

Screening and prognosis (WHO, 2017): The screening methods commonly used in the case of Down syndrome are expanded Alpha Fetoprotein (AFP), Nuchal Translucency Screening (NT), amniocentesis, Chorionic Villus Sampling (CVS) and ultrasound. These diagnostic tests inform the parents of the mental and physical needs of the child and enabling them to be prepared for the challenges ahead. The prognosis of Down syndrome depends on development of systemic complications and susceptibility to infections. In the early 1900's, Down syndrome patients were expected to live less than 10 years. Now, about 80% of adult Down syndrome patients reach their 50th birthday and beyond.

Management: Many people with Down syndrome have conditions that put them to increased risks for oral health. Low priority is given to oral hygiene and there is lack of appropriate skills among carers to maintain oral hygiene, which is compounded by the behavioural and communication difficulties with DS individuals. Some of the other barriers faced include: difficult physical access to dental services, inability of individuals with Down syndrome to complain of dental or gingival pain, lack of specialist training for dentists to meet the needs of this group and fragmentation of dental services. Suggestions and recommendations for management of oral health are as follows.[15] Pediatricians are health professionals of first contact of the individuals with Down syndrome. Hence thorough oral assessment for risk factors that contribute to poor oral health in children with special health care needs should be conducted routinely and provide anticipatory guidance on appropriate oral hygiene and habits. Pediatricians should collaborate with dental partners for oral care and advocacy for children with special health care needs (Norwood, 2013).

Diet: Keep foods and drinks with sugar to mealtimes. Choose bread, toast, chapatti, cheese, fresh fruit and vegetables. Drink milk, tea or coffee without sugar. Avoid snacks and drinks that contain sugar or carbonated or fruit flavored in between meals. Always ask doctor for sugar free medicines.

Oral hygiene: Use an electric toothbrush if possible or a manual brush with a small head, even a child's one. Use fluoride toothpaste and chlorhexidine gel if necessary. The mouths should be cleaned daily with either gauze or a soft toothbrush to remove plaque wearing latex-free gloves in edentulous subjects. Dentures should be cleaned every night with a denture cleaning solution.

Dental care: People with learning disabilities should be registered with a dentist and should be assisted to visit twice a year. Oral care plan should be drawn up in partnership with carers and dental team. Carers should not decide to discontinue oral hygiene practice without consulting with the health care team.

DISCUSSION

Studies have reported low utilization of oral health services among Down syndrome individuals. From the individual perspective the barriers include lack of awareness and importance to dental visits (Habashneh, 2012 and Abdul Rahim, 2014), lack of perceived need (Shyama, 2015), low value to oral health (Shyama, 2015 and Allison, 2001), anxiety or fear[22], financial considerations (Oredugba, 2007; Habashneh, 2012; Schultz, 2001; Kaye, 2005), lack of access (Oredugba, 2007; Shyama, 2015 and Kaye, 2005), difficulty in getting an appointment (Abdul Rahim, 2014 and Shyama, 2015) and lack of cooperation (Shyama, 2015). Whereas from the dental profession point of view the barriers include inappropriate manpower resources, location, training of health personnel, insufficient sensitivity to patient's attitudes and needs (Allison, 2001).

Conclusion

Impact of intellectual disability varies considerably among people. The aim is to prevent disease and the need for operative treatment. Increasingly sophisticated tests and treatments have influenced the lives of Down syndrome patients. Medical advances, special educational programs, and increasing social acceptance of this group in the community have resulted in current trends of normalization and deinstitutionalization of these patients. It is necessary to educate the parent so that they understand the importance of dental health for their children and its relationship to their medical condition. Aspects of preventive care include dietary counseling, provision of any necessary fluoride supplements and oral hygiene instructions. It is possible that with early intervention, appropriate education and supports as an adult, one can lead satisfying life in the community.

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