Down’s syndrome

Name

Institution

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Introduction

This paper is aimed at documenting the FDA regulations, scientific advances, role of family, description of disease, laboratory analysis, chromosomal analysis, causes, policy, nutritional influences, nutritional assessment and prevalence of Down’s syndrome. This paper will enable the reader to understand the significance of genetics in the determination of physical, mental and emotional wellbeing of the individual and how a subtle genetic mutation affects the developmental appropriateness gravely (Heller et. al., 2010). The on-the-field experience of registered nurse working in the psychiatric unit will be documented thoroughly.

FDA regulation

When it comes to the standard care of Down’s syndrome, drug therapy is not an integral component currently. This is because genetic disorders cannot be treated but can only be managed; medications are only prescribed for the symptomatic treatment of pain and other psychological abnormalities such as delusions, hallucinations and other psychotic symptoms associated with Down’s syndrome (Costa, 2011). Needless to say, using analgesics without any sufficient diagnostic evaluation and profound comprehension about the underlying etiology of disorder is never encouraged. FDA approved diuretics and digoxin for maintaining congestive heart failure and congenital heart defect (Costa, 2011).

Scientific advances

Some year ago, individuals with Down’s syndrome were prescribed with amino acid supplements but later on researches indicated that this treatment gives adverse reactions in controlled trials. With the technological advancements, more efficient psychoactive drugs are now being developed to manage the associated neurological symptoms (Guralnick, 2010). Moreover, children with Down’s syndrome are provided with the assistive devices such as amplification tools, specialized pens, bands, touch screen computers and system with larger display that help them accommodating their academic and other tasks efficiently. Other evidence based non- pharmacological treatment options for Down’s syndrome include occupational therapy, emotional and behavioral therapies, speech and language therapy, physical therapy and family education (Heller et. al., 2010).

Role of the family

The role of family is irrefutably significant in the management of Down’s syndrome because as mentioned earlier, patients require assistance in their daily task management and full time supervision. They suffer language-related, hearing, walking and cognitive impairments hence become unable to execute even subtle moves on their own (NDSC, n.d). This is why; education of the family members is quite essential component of psychotherapy; the way parents and siblings interact with the patient determines the level of improvement that he attains from medications and psychotherapy. The role of family encapsulates following: learning more about Down’s syndrome, loving and playing with the child, encouraging child to become independent, assigning chores to the child, working with the professionals collaboratively, finding out what the child is learning in school, looking for more social opportunities and developing a social circle having other parents with a Down’s syndrome child (NDSC, n.d).

Description of disease

Individual with this rare condition are prone to get certain physical and psychological features in common such as small ears, flat nose, mild to moderate issues in learning, understanding, reasoning and thinking and difficulty in attaining the expected milestones of development appropriately such as talking, walking, running and developing communication skills (Guralnick, 2010). Many individuals with Down’s syndrome do not report any other health issue however exceptions are always there; cardiovascular problems, hearing and vision related problems may occur. Above all, individuals having Down’s syndrome cannot live an independent life; they always need someone to manage their daily tasks.

Laboratory analysis

Two types of tests are administered on the suspected patient in the laboratory; the screening and diagnostic tests (Ravel et. al., 2019). Screening test is usually done during pregnancy including blood test, Nuchal translucency test, Chorionic villus sampling (CVS) and Amniocentesis. The distinctive facial features of affected patient are prominent at the time of birth that call for further diagnostic testing; blood test is done for identifying the extra chromosome in the genetic material; the hallmark of Down’s syndrome (Ravel et. al., 2019). However in the in vitro fertilization, pre-implantation genetic diagnosis is an excellent option for detecting this unwanted condition.

Chromosomal analysis

Typically, during the cell division, two copies of all the 23 chromosomes are prepared for the developing embryo which number 46 in total. In some cases, the chromosome number 21 gets replicated three times instead of two that result in the formation of triplet of 21st chromosome, typically referred to as the *non-disjunction* (Ravel, et. al., 2019)*.* As a result of this non- disjunction, the resulting individual develops deteriorated mental capabilities, developmental delays and abnormal physical features. The portion of copied chromosome determines the severity level of Down’s syndrome i.e., mild, moderate and sever.

Causes of disorder

Three types of genetic variations can cause Down’s syndrome; Trisomy 21, Mosaic Down syndrome and Translocation Down syndrome. Trisomy 21 is caused due to the triplet copy of all the chromosomes number 21; it is the most common condition. In the Mosaic Down syndrome, some cells have an extra copy of 21st chromosome whereas the Translocation Down syndrome is when some portion of chromosome number 21 becomes attached to other chromosome erroneously however it is a rare condition (Glasson et. al., 2012). Other risk factors of the Down’s syndrome include mother aged greater than 35, mother being carrier of the genetic translocation, having another child with the similar condition (Sentilhes et. al., 2015).

Policy

It is important to consider all the potential etiologies of Down’s syndrome before planning associated policies. The most exclusive part would be to determine the actual cause of disorder specific to the person through eliminating all the other plausible causes. For that matter, appropriate medical and psychological testing coupled with the medical history of the parents must be kept in account before developing and implementing any care plan. The prenatal testing would render immensely effective in this regard. Moreover, management of the condition would be prioritized than mere medical treatment; other interventions include non-pharmacological techniques such as cognitive and behavioral therapies. Education of the patient’s family is another integral component of the policy making (Glasson et. al., 2012).

Nutritional influences

The conductive feeding relationship between parents and children is essential for accomplishing the valued goals of holistic wellbeing. Their daily calorie need is less than the children of identical ages without this condition due to lower muscle activity and subsequent metabolic rates (Bennett et. al., 1983). Infants are prescribed to take more iron and vitamin D rich food whereas vitamin A, Calcium. Fluoride and iron must be given to the toddlers.

Nutritional assessment

Nutritional assessment revolves around various parameters such as clinical examination (anthropometry and physical examination), biochemical (functional tests and nutrient stores), and dietary (historical data and present nutrient intake). Individuals with Down’s syndrome require some extra assessment of their feeding skills because their developmental milestones are delayed; they might feel chewing difficulty, coordination of tongue with teeth and lacking energy for grinding food (Beal, 1961).

Prevalence

Down’s syndrome is quite rare among other physical, neurological and psychological impairments however it is the most common genetic disorder. A report from Center of Disease Control and Prevention suggests that about 6000 babies each year are born with this unfortunate condition; 1 out of 7 babies is affected with this genetic abnormality (Parker et. al., 2010).

Conclusion

In a nutshell, Down’s syndrome is not curable but only manageable because it is a genetic birth defect. Its management strategies mainly encapsulate cognitive and behavioral techniques and family education. The need of the hour is to make active attempts in diagnosing this condition correctly so that interventions could be applied accordingly ensuring effective outcomes.

References

Beal, V. A. (1961). Dietary intake of individuals followed through infancy and childhood. *American Journal of Public Health,* 51, 1107.

Bennett, F.C., McClelland, S., Kriesgsmann, E.A., Andrus, L.B., & Sells, C.J. (1983). Vitamin and mineral supplementation in Down’s syndrome. *Pediatrics*, 72 (5), 707–713.

Centers for Disease Control and Prevention (2016). *Facts about Down syndrome*. Atlanta, GA: Author. Retrieved from  
https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html

Costa, A. C. (2011). On the promise of pharmacotherapies targeted at cognitive and neurodegenerative components of Down syndrome. *Developmental Neuroscience*, 33, 414–427.

Glasson, E. J, Sullivan, S. G., Hussain, R., Petterson, B. A., Montgomery, P. D, et al. (2012). The changing survival profile of people with Down’s syndrome: implications for genetic counselling. *Clin Genet.* 62, 390–393.

Guralnick, M. J. (2010). Early intervention approaches to enhance the peer-related social competence of young children with developmental delays: A historical perspective. Infants and Young Children, 23, 73–83.

Heller, J. H., Kishnani, P. S., Spiridigliozzi, G. A., Lott, I., Escobar, L., Richardson, S., Zhang, R., et al. (2010). Treatment of cognitive dysfunction in children with Down syndrome aged 10–17. *American Journal of Medical Genetics Part A*, 152A, 3028–3035.

National Down syndrome Congress (NDSC) (n.d.). New and expectant parents. Available online at: http://www.ndsccenter.org/new-and-expectant-parents/

National Human Genome Research Institute. (2010). Learning about Down syndrome. Retrieved June 11, 2012, from http://www.genome.gov/19517824

Parker, S. E. Mai, C. T. Canfield, M. A. et. al. (2010). Updated National Birth Prevalence estimates for selected birth defects in the United States, 2004-2006. *Birth Defects Res A Clin Mol Teratol.* 88(12), 1008-16. doi: 10.1002/bdra.20735.

Ravel, A. Mégarbané, A., Mircher, C., et al. (2019). The 50th anniversary of the discovery of trisomy 21: the past, present, and future of research and treatment of Down syndrome. *Genet Med.* 11, 611–616.

Sentilhes, L., Salomon, L. J., Vayssiere, C. (2015). Cell-free DNA Analysis for Noninvasive Examination of Trisomy*. N Engl J Med.* 373, 2581–2582.