Commentary

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Oxidative Phosphorylation Mechanism Contribute To Genome-Wide Deregulation

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Description

Due to a triplication of chromosome 21, Down Syndrome (DS) has unique health care requirements. Abnormalities in the cardiac, digestive, and vertebral systems are among the congenital conditions associated with DS. Normal obtained conditions for people with DS incorporate thyroid illness, diabetes, dental issues, and hearing misfortune. A person with DS has a unique psychological profile, including the possibility of having major depressive disorder, bipolar disorder, and autism spectrum disorder as co-occurring disorders and an earlier onset of Alzheimer disease and cognitive decline. The demand for long-term specialty care is driven by these acquired and congenital conditions. Despite this, individuals with Down syndrome receive inadequate care. The study also found that less than half of the adolescents with Down syndrome who attended primary care clinics were up to date on preventative measures such as vaccinations, well-visits, and cancer screenings.

Amyloid Precursor Protein Gene

Due to trisomy of chromosome 21 and the resulting extra copy of the amyloid precursor protein gene, nearly all adults with Down syndrome develop Alzheimer's disease pathology by the age of 40. Given their longer life expectancy than adults with Down syndrome in the past, they are also at high risk for dementia. In order to improve our comprehension of the disease mechanisms in these two genetic groups at high risk for Alzheimer's disease, we set out to compare the patterns of CSF biomarkers in Down syndrome with those in carriers of autosomal dominant Alzheimer's disease mutations. Despite their dedication to their work, young adults with Down syndrome struggle to make friends at work. However, they were encouraged by the conversations they had with coworkers who they recognized as friends. They looked forward to interacting with their friends, despite the fact that there were few opportunities for them to do so.

In people with Down Syndrome (DS), mitochondrial dysfunction and impairment in the oxidative phosphorylation mechanism contribute to genome-wide deregulation, which causes oxidative stress, an increase in the production of Reactive Oxygen Species (ROS), and cell death. The muscles, brain, and heart, as well as other cells that require more energy, are significantly impacted. Patho-mechanics at the cellular and systemic levels are directly correlated with mitochondrial network impairment in DS patients, who also experience generalized metabolic perturbations. In the DS population, where mitochondrial dysfunctions play the central role, numerous clinico-phenotypic features, including intellectual disability, early aging, neurodegeneration, and AD-related dementia, are inevitable. Together, the altered energy metabolism and mitochondrial abnormalities disrupt several signaling pathways, particularly those related to neurogenesis, which are directly linked to cognitive development and the early onset of AD in DS patients.

Production of Reactive Oxygen Species

As a result, the DS population's quality of life was thought to improve as a result of therapeutic challenges to correct mitochondrial defects. Various pharmacologically dynamic normal mixtures, for example, polyphenols, cancer prevention agents and flavonoids have shown persuading result for inversion of the useless mitochondrial network and oxidative digestion, and improvement in scholarly expertise in mouse models of DS and people with DS. As a result, these DS patients have 46 chromosomes despite carrying one structural and one numerical abnormality in their genome. This DS variant is more common than other variants with mosaicism or translocation. All of the offspring of the twill parents have variant DS. The Robertsonian translocation carriers will have 45 chromosomes in their genome, and their DS offspring will have 46 chromosomes, two of which will be translocated inherited from the carrier parent and one of which will be free of HSA21.Thus, in this case, the disorder is inherited; Otherwise, it is a condition that develops over time and is not inherited.

Meiotic non-disjunction and the fertilization of the aneuploid gamete are the main causes of trisomy 21, but mosaicsm can also occur during embryonic development when mitotic non-disjunction occurs. Down syndrome is no longer confined to children. Better care standards for adults with Down syndrome are required by an increase in life expectancy. A common childhood diagnosis for chromosomal disorders is Down

syndrome. The most frequently cited care guidelines need to be updated to include children. Life expectancies for people with Down syndrome have reached middle to late adulthood as a result of advancements in health care. Obstructive Sleep Apnea (OSA), Central Sleep Apnea (CSA) syndromes, sleep-related hypoventilation disorders, and sleep-related hypoxemia disorder are all included in the heterogeneous group of disorders known as Sleep Disordered Breathing (SDB) .Intermittent hypoxia, sleep fragmentation, and obstructed respiratory efforts caused by disruptions in respiration during sleep have been linked to cardiovascular, metabolic, and neurologic consequences. Down disorder trisomy 21 is a typical recognizable reason for scholarly inability.

Individuals with Down syndrome exhibit a wide range of manifestations, despite the fact that there are many shared characteristics. Understanding the condition and its effect on individual lives can help keep up with and advance wellbeing, and empower people to accomplish their true capacity. It is essential not to fall into the trap of labeling all symptoms as part of Down syndrome because some conditions are more prevalent in Down syndrome patients. People will be able to get the support and care they need if they are aware of some of the conditions that are seen more frequently. Understanding these conditions will also help with timely investigation and management. This article focuses on our role as health professionals in assisting Down syndrome families, children, and adolescents. We talk about some of the conditions that happen more frequently to people with Down syndrome and our role in screening, finding, and treating them.