Commentary

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Moyamoya Disease Is a Rare Occlusive Cerebrovascular Condition

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Description

When compared to children who do not have Down syndrome, those with Down syndrome belong to a distinct genetic population that is more likely to develop Acute Myeloid Leukemia (AML) and Acute Lymphoblastic Leukemia (ALL).The risk of developing solid tumors is also different from that of people without Down syndrome. In the case of myeloid leukemias, Transient Abnormal Myelopoiesis (TAM), a pre-leukemic condition caused by genetic factors such as GATA1 mutations, begins the process of leukemogenesis in Trisomy 21 in the early stages of fetal development. Myeloid Leukemia with Down Syndrome (ML-DS) can develop as a result of a number of other mutations in genes that encode cohesin, epigenetic regulators, and the RAS pathway. The striking paradox of the Down syndrome population is that, despite having a higher risk of developing AML, they are also extremely sensitive to chemotherapy agents, particularly cytarabine.

Rare Occlusive Cerebrovascular Condition

This explains why the cure rates for ML-DS are so much higher than those for AML in children who do not have Down syndrome. By de-intensifying chemotherapy doses, if at all possible, current clinical trials for ML-DS attempt to strike a balance between effective curative therapies and the reduction of treatment-associated toxicities, such as infections. The little extent of patients with backslid ML-DS have an incredibly unfortunate forecast and require the advancement of new treatments. In the Down syndrome population, pulmonary hypertension and myocardial impairment are well-known risk factors for cardiorespiratory morbidity. Their intricate relationship, including the potential negative impact of LV diastolic impairment on pulmonary hypertension indices, has not been fully characterized. When compared to the population without DS, the global incidence of solid tumors is lower. A disorder caused by an excess copy of trisomy 21 is known as Down syndrome, also known as trisomy 21.The condition has even been shown to be the most common chromosomal-related disorder and has a very high prevalence in various global communities. The various DS patients have very different levels of disease severity. The appearance of DS patients is distinctive, and typical manifestations include intellectual disabilities and developmental delays that last a lifetime. In addition, the disease may be associated with a number of complications, such as obstructive sleep apnea, heart defects, ear infections, eye diseases, and hearing loss. DS can have a negative impact on the quality of life of those who care for the affected children as well as the significant morbidity it causes.

Moyamoya disease is a rare occlusive cerebrovascular condition that typically affects children. It is characterized by stenosis of the internal carotid artery and the circle of Willis, which results in ischemia of the brain. A Moyamoya-like arteriopathy known as Moyamoya syndrome has risk factors such as Down syndrome, thyroid disease, sickle cell disease, or autoimmune disorders. Trisomy 21 is a genetic condition associated with intellectual impairment and particular physical and behavioral characteristics. An adult with Down syndrome presented with ischemic stroke in a rare case of Moyamoya syndrome is the subject of our report. A partial or full trisomy of chromosome 21 has been linked to mitochondrial dysfunctions in Down Syndrome (DS).In eukaryotic cells, mitochondria are essential for a number of essential functions, particularly energy production, calcium homeostasis, and programmed cell death.

Mitochondrial Dysfunctions

Genes encoding in the nucleus and mitochondria are primarily responsible for regulating the function of mitochondria. The effects and molecular mechanism of gene therapy, as well as drugs that exert protective effects in DS cells through modulation of mitochondrial function and attenuation of oxidative stress, were all discussed as potential therapeutic strategies for modifying DS's energy deficits. It is prudent to target something as straightforward as cellular mitochondria biogenesis and function to improve DS pathophysiological conditions or quality of life. The underlying molecular mechanisms that contribute to the syndrome and the phenotype of accelerated aging remain largely unknown, despite the fact that the cause of Down Syndrome (DS) is well-established. Although DS significantly alters DNA methylation profiles, it is still unclear how various methylation regions and probes are organized into a network of interactions. The strategy is expected to be broadly applicable to other diseases.

People with Williams Syndrome (WS) or Downs Syndrome (DS) are frequently referred to as being overly friendly, sociable, and trusting of other people. Due to the increased risk of exploitation and victimization, this hypersociability is a major concern for parents and caregivers. Two mind locales - the amygdala and the Orbito Frontal Cortex (OFC) - have been ensnared in driving this hypersociability in WS, and in everybody and have relationship with close to home assessment, danger discovery and social inspiration. However, there has been relatively little neuroimaging research on this subject to date, particularly in DS. The purpose of this study was to look into the possibility of neuroanatomical and neuropsychological links between hypersociability in WS and DS. As was to be expected, people with WS had the best overall social approach, especially when it came to the need and drive to interact with strangers and inappropriate or overly friendly behaviors. In terms of social trust and unconditional positive regard, both groups scored similarly. The DS group had some trouble recognizing negative emotions especially anger, but the other groups had similar emotional recognition abilities. Both WS and DS had impairments in flexibility and inhibition that were comparable. The DS group had larger bilateral amygdala volumes compared to neurotypical controls, while the WS group had a larger right medial OFC.