MICROCEPHALY: A Developmental Disorder

Humera Kausar
¹Department of Biotechnology, Kinnaird College for Women, Lahore
*humera.kausar@kinnaird.edu.pk

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Microcephaly is a result of abnormal in utero development resulting in an unusually small head size. It is caused by a number of variables, including chromosomal abnormalities as well as other genetic conditions, infections during pregnancy, for instance; rubella, toxoplasmosis, and prenatal exposure to dangerous toxicants. Although drastically deficient cognitive growth is prevalent, problems with motor control processes not showing up until much later in life. Most negatively impacted infants have severe neurological abnormalities and sometimes seizures, as well. Motor function and verbal advancement may also be deferred while hyperactivity and intellectual disability are both prevalent, however to varying degrees. Convulsions are also possible with variations in motor ability; from clumsiness to spastic quadriplegia in some people. The majority of cases of microcephaly are caused by genetic variations. On the one hand, linkage has been discovered between autism, gene duplications, and macrocephaly. On the contrary, a link has been discovered between schizophrenia, gene removals, and microcephaly.

Being caused by a reduction in cerebral cortex, and it can occur throughout embryonic and foetal growth phases because of inadequate neural stem cell advancement, impeded neurogenesis, or decrease of neural stem cells. Many genes needed for standard neural development have been discovered through studies in animal models such as rodents. The genes associated with the Notch pathway, for instance, govern stem cell advancement and neurogenesis. Genetic variations induced in mouse models in an experimental setting can induce microcephaly that in comparison is similar to that of the human beings. Abnormal Spindle-like Microcephaly-Associated (ASPM) gene abnormalities have been linked to human microcephaly, and a knockout-model of a ferret with extreme microcephaly has now been designed. Furthermore, viruses like Zika virus and Cytomegalovirus (CMV) have been found to affect and destroy the brain’s primary stem cells and radial glial cells, resulting in the destruction of daughter neurons.

A comprehensive physical and history assessments are carried out on patients with microcephaly. Neuroimaging, metabolic evaluation, and genetic examination should be taken into consideration in cases of deteriorating microcephaly. Neuroimaging with Magnetic Resonance Imaging (MRI) is mostly utilized as the very first diagnostic analysis in children suffering from microcephaly. Genetic screening is frequently the very next process after imaging techniques. Microcephaly is a long-term condition with no specific treatment available.