Microcephaly

Source: TH

Microcephaly, a **neurological condition** characterised by an **abnormally small head and impaired brain development**, has been the focus of extensive research, with the **SASS6 gene** emerging as a key player in this complex genetic disorder.

- Children with microcephaly often have a **small brain, poor motor function, speech impairment**, abnormal facial features, and **intellectual disability.**
- The roots of microcephaly lie in the peak phase of brain development in the embryo when the **cells destined to become neurons fail to divide normally.**
- Since 2014, a gene called SASS6 and its variants have been implicated in this developmental process.
- Researchers have observed that mutations in the SASS6 gene can lead to **abnormal centriole formation,** crucial for **cell division and neural development.**
- The **Ile62Thr Mutation in the SASS6 gene** has been linked to microcephaly, with the protein made using the mutated gene still being functional enough to allow survival, **but causing brain and head deficits.**
- **According to researchers,** consanguineous marriages **(cousin marriage)** increase the risk of inheriting a mutated copy of a gene, including the SASS6 gene, leading to a higher incidence of microcephaly.

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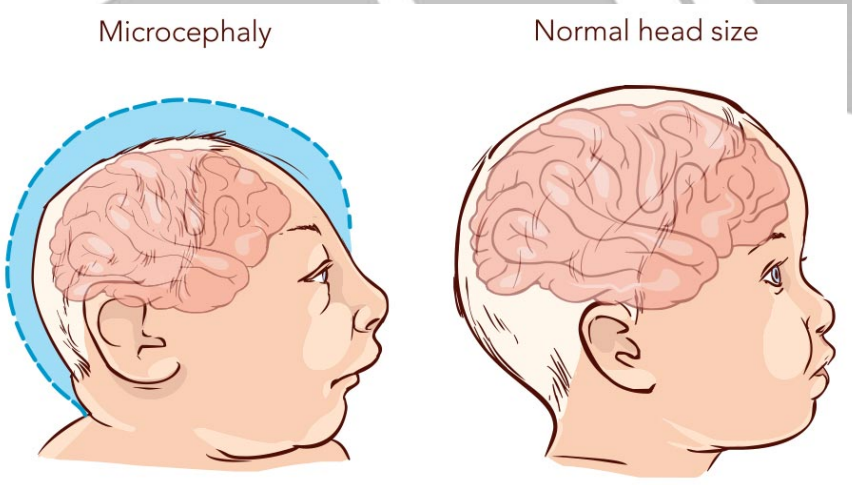

Read more: NBRC Researchers Decipher the Cause of Microcephaly