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Singapore sheds new light on unsolved genetic diseases in children

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The study highlights the role of genes inherited from mothers in genetic diseases in children, and improves the understanding of such diseases

The development of an embryo is a well-orchestrated string of processes, ensuring correct formation and positioning of vital organs of the growing organism. At the molecular level, these processes are controlled in a precise manner by switching on or off specific factors such as genes or proteins. Any errors in these processes could result in physical defects or disease in the newborn organism.

A team of scientists from the National University of Singapore (NUS) led by Assistant Professor Xue Shifeng from the Department of Biological Sciences has discovered a new way to interpret unsolved Mendelian diseases – diseases inherited from either parent due to gene mutations in the developing egg or sperm – through studying the inheritance of a protein known as SMCHD1 which is coded by the *SMCHD1* gene.

Mutations in the *SMCHD1* gene can cause diseases such as facioscapulohumeral muscular dystrophy (FSHD) which is a muscle degenerative disorder, and Bosma arhinia microphthalmia syndrome (BAMS) which causes abnormalities of the nose and eyes.

According to the researchers, this will change how we think about unsolved inherited diseases.