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Study reveals reasons for common epilepsy in Indian children

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In a first-of-its-kind Medgenome Labs conducted a study in partnership with doctors from Santokba Durlabhji Memorial Hospital, Jaipur, School of Medicine, University of Glasgow, UK and Pediatric Neurosciences Research Group, Royal Hospital for Children, Glasgow, UK to understand the underlying causes of Infantile Spasms (IS), a common but severe form of epilepsy in young children.

Infantile spasms also called ‘Salaam Seizures’ usually occur in the first year after birth. To date, there had been no published studies from India to comprehensively investigate for this condition. This is the first study that used Next Generation Sequencing (NGS) technology to dwell deep into the causes of infantile spasms in the Indian setting.

The other important cause of infantile spasms identified in this study were underlying genetic defects which put a child at risk to develop these types of seizures later in infancy. In this study, for the first time a detailed genetic evaluation using NGS-based Whole Exome Sequencing was performed in patients with a suspected genetic cause for Infantile Spasms. Following this exhaustive genetic evaluation, a genetic cause could be identified in a significant number of patients with infantile spasms.

The findings of the study emphasize on the importance of doing a Whole Exome Sequencing in patients of infantile spasms where no apparent cause can be found, which will help in diagnosing these genetic disorders early and prevent it from happening again in subsequent pregnancies.

The outcomes of this study will further improve the understanding of the causes of infantile spasms in India so that it can better be managed and prevented in the future.