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A CURIOUS CASE OF DEVELOPMENTAL DELAY : MENKE HENNEKAM SYNDROME

Nursing/Pediatrics

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KEYWORDS

A one and half year old male child presented to the ER with history of unprovoked tonic clonic seizures (2-3 episodes) each lasting 1-2 minutes more involving the right side . No history of uprolling of eyes or drooling of saliva . The child was stabilized using the Status Epilepticus protocol and was shifted to PICU . LP was done and was managed accordingly

He is a Second born baby to nonconsanguinously married parents via Normal vaginal delivery AGA / GDM / NNH on day 3 of life and was discharged on Day 5

Hypotonia picked up during the vaccination visit of Day 45 . started on Developmental Therapy . Delay in Gross motor and Language domains .continued DT and Speech therapy Head banging noted at 9 months of age and was started on Sizodone and was symptomatically better

O/E : mild dysmorphic features prominent forehead broad nasal tip depressed nose , open mouth short philtrum

EEG : prefrontal poly spikes MRI brain : small thin walled simple intra ventricular cyst in frontal side of lateral ventricle

Elder sister had febrile seizures with ho developmental delay

NGS : pathogenic variant in exon 31 suggestive of MENKE HENNEKAM SYNDROME 1

Parents refused genetic testing on financial grounds . At present he is on Leviteracetam and Sizodone DT and speech therapy . His anthropometric parameters are in the lower centile . he remains seizure free since then but has IP for LRTI

Menke hennekam syndrome is a recently diagnosed genetic disorder caused by specific heterozygous pathogenic variants in exons 30 or 31 of CREBBP gene .it presents with a wide array of subtle phenotypical features often associated with intellectual disability and microcephaly mutations in the same gene causes rubinstein taybi syndrome but they do not have any overlapping features

Life expectancy of this condition depends on the severity . only very few cases have been reported so far in the world