VOLUME - 12, ISSUE - 07, JULY - 2023 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Paediatrics

A RARE CASE OF RAYNAUDS CLAES SYNDROME

Dr. Priyanka Patil	III Year Junior Resident, Pediatrics. Department of Pediatrics, MGM medical college and hospital, Kamothe, Navi Mumbai
Dr. Bageshree Seth	Department of Pediatrics, MGM medical college and hospital, Kamothe, Navi Mumbai
Dr. Vikram Patra	Department of Pediatrics, MGM medical college and hospital, Kamothe, Navi Mumbai
Dr Roshni	Department of Pediatrics, MGM medical college and hospital, Kamothe, Navi Mumbai
Dr Michelle	Department of Pediatrics, MGM medical college and hospital, Kamothe, Navi Mumbai

KEYWORDS :

Case

13y/female , 2ndby order of birth, born out of non consanguineous marriage presented with Acute exaggeration of behavioural change. Child was a k/c/o intellectual disability, behaviour disorder and seizure disorder with past history of stroke at 1 month of age with right sided hemiparesis, CT BRAIN at that time suggestive of Acute bleeding in right thalamic region and Acute Infar infarction involving entire right hemisphere, eeg s/o epileptic activity over both fro to temporal regions. In view of this early onset history of stroke and behavioural change child was worked up for homocysteinemia and urine fir homocysteine showed homocysteine uria and child started on pyridoxine supplements, anti epileptic and psychiatric medication continued. Genetic testing of the child s/o Raynaud claes syndrome . gradual improvement in behaviour was note

Raynauds claeys syndrome is a neurodevelopemental disorder , X Linked, characterised by developmental delay, intellectual disability, epilepsy, behavioural change, epilepsy and gastrointestinal dysfunction . Till date it has been Reported in 66 individual, 22 of whom are asymptomatic or only very mild selected heterozygous females. It is not known to be more prevalent in any population, and no founder variant are known.

It is inherited in an X Linked manner If the mother has a pathogenic variant the chance of transmission in each pregnancy is 50 % ,likes who inherit pathological variant will be affected females who inherit pathological variant will be heterozygous and nay be unaffected or have mild to severe manifestations.

Growth , development, neurological evaluation, psychiatric evaluation, ophthalmology, hearing assessment genetic counselling should done at initial diagnosis

CONCLUSION

organic disease should be ruled out in case of children presenting with behavioral changes. In this case early onset history of stroke, Seizures and behavioral changes was taken into consideration and the child was further investigated for homocysteinemia.

Genetic study could further help with the diagnosis