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A CASE STUDY OF RETT SYNDROME IN HYDERABAD

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Abstract:
Rett syndrome (RTT) is a rare genetic neurodevelopmental disarray in which children are mostly effected with relapse of previously acquired skills after a period of normal development. Rett syndrome is usually identified in children age between 6to 18 months as they begin to lose the abilities they gained.it is caused by the mutation on the x chromosome on a gene MECP2.There are about 900 different mutations found on MECP2 gene. It is not a degenerative disorder. The course and severity of Rett syndrome is determined by the location, type and severity of the mutation and X-inactivation. In more than 99 percent of people with Rett syndrome, there is no history of the disorder in their family.

Key words:
Rett syndrome, ME Rett syndrome, MECP2 mutation/Methyl-C-phosphate-G-binding protein 2, mental retardation, hand-wringing movement, microcephaly, autism.

Introduction:

Rett syndrome is a dominant X-linked disorder largely caused by mutations in the gene encoding methyl-CpG binding protein 2 (MECP2). Rett syndrome is caused by mutations inthe MECP2 gene, which has been mapped to the locus Xq28.

The MECP2 gene encodes the protein methyl-CpG binding protein 2 (MECP2), which functions as one of several biochemical switches; patients with RTT show improper functioning of this gene and insufficient amounts or structurally abnormal forms of the protein. Methyl-CpG binding protein 2 functions as a transcriptional repressor, an activator,and an RNA-binding protein. Systematic studies of the MECP2 gene revealed a range of mutations which were associated with different phenotypes.

In vivo studies using mice with MECP2 mutations revealed neuropathological and behavioraldeficits similar to those reported for RTT.

Mutations in the MECP2 gene were observed in 70%–80% of the girls diagnosed with RTT using current diagnostic techniques. The remaining 20%–30% of the cases were attributedto partial gene deletion and unknown mutations.

Mutations in CDKL5 (cyclin-dependent kinase-like 5) gene are associated with clinicalmanifestations in a few patients with RTT.

Rett syndrome occurs worldwide in 1 of every 10,000 female births, and is even rarer in boys.The MECP2 gene is located on the X chromosome. Between 90% and 95% of girls with Rettsyndrome have a mutation in the MECP2 gene. Most children with Rett syndrome have a mutation on the X chromosome.

Exactly what this gene does, or how its mutation leads to Rett syndrome, isn't clear. Researchers believe that the single gene may influence many other genes involved in development. Although Rett

syndrome is genetic, children almost never inherit the faulty gene from their parents. Rather, it's a chance mutation that happens in DNA.When boys develop the Rett syndrome mutation, they rarely live past birth. Males have only oneX chromosome (instead of the two girls have), so the effects of the disease are much more serious, and almost always fatal.Although There's no known way to prevent Rett syndrome. In most cases, the genetic mutation that causes the disorder occurs spontaneously. Even so, if you have a child or other family member with Rett syndrome, you may want to ask the doctor about genetictesting.

Rett syndrome primarily affects girls, with an incidence of 1:10,000–20,000 (Hagberg, Hanefeld, Percy, & Skjeldal, 2002). Most of the children with RTT are the born from a normal pregnancy and delivery (Dolce, Ben-Zeev, Naidu, & Kossoff, 2013). In an initial period the child will show the normal development from age 6–12 months after this is followed by a period of rapid decline with regression of acquired motor skills, loss of speech and purposeful hand use, abnormal walk and growth failure. This regression is sometimes sudden and often rapid, occurring in the time span of weeks to months and usually this period is associated with severe sleep disturbances, irritability, and poor eye contact (Dolce et al., 2013).

Case Report:

A 45-year-old women was the only child and she affected with mental retardation and delayed development. she was born of a consanguineous marriage and through vaginal delivery. At birth, her father and mother were 32 and 28 years of age, respectively. Normal development was observed till the age of 6 months, after that they observed developmental regression. Loss of eye contact was observed at the age of 7 months, followed by a gradual change into an introvert. The patient is examined by several doctors and she diagnosed with autism concluded by the doctor's based on her behaviour. They have even noticed that the patient's third degree relative (cousin-MALE) was suffering from same condition. Physical examination revealed loss of speech, repetitive hand-wringing movement, short stature,strabismus, microcephaly, and autistic behavior such as Improper muscle movements, muscle weakness, problems with coordination, stiff muscles, rhythmic muscle contractions, abnormal breathing patterns, shallow breathing, delayed development ,failure to thrive, irritability or repetitive movements, inability to speak or understand language or, slowness in activity, Seizures, Constipation, drooling ,teeth grinding. The patient was confirmed with Rett syndrome after a blood test confirming MECP2 mutation. Although there is no cure for Rett syndrome, doctor's providing the treatments for patient to overcome improve her symptoms. The patient is supposed to continue the treatments for her entire life. Treatment includes Physical therapy , Speech therapy, having Good nutrition , Behavioural therapy and few Medication are also recommended in controlling seizures such as Antiepilepticdrugs (AEDs) ,some Sedatives such as hypnotic agents are used to treat sleep disturbances.

X-LINKED DOMINANT

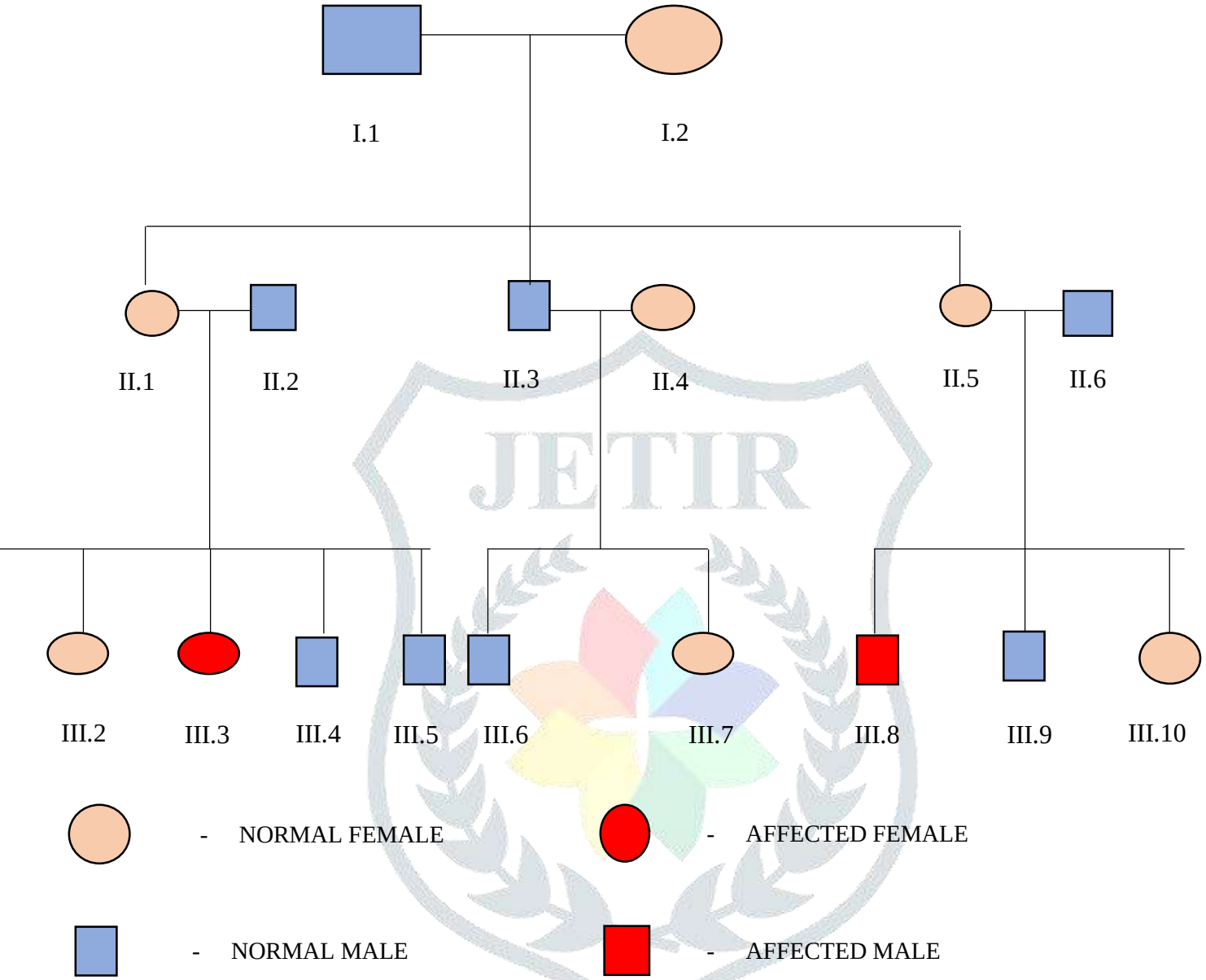

Fig: *Showing pedigree of three generations of the family.*

Discussion:

Initially it is thought to be affecting females, but it has even identified in males. The incidence in males is unknown, partly owing to the low survival of malefoetuses with the RTT associated MECP2 mutations, and partly due to differences between signs caused by MECP2 mutations and those caused by Rett's. There are several cases of 46XY karyotype males with a MECP2 mutation carried to term who were affected by neonatal encephalopathy and died before theage of 2 years. The course of RTT, including the age of onset and the severity of symptoms, varies from child to child. Currently there is no known specific treatments for RTT. Clinical care, comprises genetic counselling (with DNA tests to rule out familial transmission), support and advice for the families, anticonvulsant medication upon the development of epilepsy, and physiotherapeutic measures for alleviating scoliosisdevelopment to the extent possible .During regression,

certain features of RTT are similar to those of autism; misdiagnosis of RTT as autism is, therefore, likely. The important role played by genetic factors in these conditions should be considered by individuals concernedwith autism spectrum disorders, including psychiatrists, psychologists, or paediatricians.

Conclusion:

Rett syndrome is very complex neurological disarray, it can be diagnosed by physical elucidation. A heterogeneous spectrum of phenotypes are identified called “atypical Rett”. Nowadays, it is more precise to consider this broad spectrum of neurological disorders as the expression of a complex encephalopathy. Many efforts have been made to understand the molecular bases of RTT. With the increase of knowledge, sincere gratitude to recent genetic advances, many new genes have been identified as causative for RTT phenotype, in addition to MECP2, CDKL5, and FOXG1.

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References:

1. Bernstein BE, Schneider JH, Glaze DG. Rett Syndrome.Medscape. Updated: Jan 5,2015.

2. De Filippis B., Ricceri L., Laviola G. Early postnatal behavioral changes in the Mecp2- 308 truncation mouse model of Rett syndrome. Genes Brain Behav. 2010;9:213–223.[PubMed] [Google Scholar]

3. Das D.K., Jadhav V., Ghattargi V.C., Udani V. Novel mutation in forkhead box G1 (FOXG1) gene in an Indian patient with Rett syndrome. Gene. 2014;538:109–112. [PubMed] [Google Scholar]
4. http://www.ncbi.nlm.nih.gov/pubmed/22670134

5. http://emedicine.medscape.com/article/916377-overview
6. https://rarediseases.org/rare-diseases/rett-syndrome/

7. https://rettsyndromenews.com/2019/10/17/new-mecp2-mutation-may-cause-atypical-rett-syndrome-case-study-reports/
8. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6520293/