Original Research Paper

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RARE CASE REPORT ON PRADER -WILLI SYNDROME

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KEYWORDS :

Prader- Willi Syndrome (PWS) is a neurodevelopmental genomic imprinting disorder with lack of expression of genes inherited from the paternal chromosome 15q11-q13 region usually from paternal 15q11-q13 deletions (about 60%) or maternal uniparental disomy 15 or both 15s from the mother (about 35%). An imprinting center controls the expression of imprinted genes in the chromosome 15q11-q13 region. In most cases, Prader Willi syndrome is caused by random genetic error and is not inherited. Determining which genetic defect caused Prader Willi syndrome can be helpful in genetic counselling. This syndrome has an incidence of affecting 1 in 10,000 or 30,000 individuals in a population and is known to affect both males and females[1]. It is termed to be a very rare disease occurring in individuals.

Diagnosis:

Diagnosis is mainly dependent on the patient history and observation of characteristic symptoms in the affected individual.

Genetic testing is performed to detect to diagnose the condition and identify the specific genetic subtypes like imprinting defect, maternal disomy 15, 15q11-q13 deletions etc.

Fluorescent in situ hybridization, DNA methylation tests and high-resolution chromosomal microarray techniques are involved in this process to detect deletions, duplications, rearrangements etc.

In cases of families with previous history of PWS, prenatal diagnosis is preferred.

CASE REPORT:

A twenty-year-old patient reported to our OPD at 7 weeks pregnancy with USG documented report of missed abortion for termination of pregnancy. On taking history we came to know history of one missed abortion at same gestational age one year back. There was no history of hypothyroidism, diabetes. After previous missed abortion she was tested for TORCH antibodies, APPLA Profile, thyroid profile, blood sugar and all these tests were normal. So couple was counselled for genetic study of fetus for which they agreed. Patient was admitted and pregnancy was terminated and the abortus was sent for genetic study (quantitative Fluorescent PCR) for trisomies and maternal blood was also sent for MLPA for sub telomeric region.

The Quantitative Fluorescent (PCR) for Trisomies showed Normal Complement of 21 ,13, 18 ad sex chromosomes.

MLPA for Sub telomeric region showed heterozygous duplication of both probes on chromosome 15 in the probe set used suggestive of three copies of chromosome 15.

Karyotyping of father revealed normal male karyotype.

Karyotyping of mother revealed normal female karyotype.

So, it was concluded that in this case this syndrome was caused by random genetic error.

REFERENCES :

1. www.mayoclinic.org/diseases-conditions/pradder willi syndrome/symptoms-causes/syc20355997