

A case report of infant with robertsonian translocation

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Abstract

Robertsonian translocations carry reproductive risks that are dependent on the chromosomes involved and the sex of the carrier Robertsonian translocation (14;15)(q10;q10)4p is rare and account for only 0.6%.

We describe a case with rob (14;15) in which the phenotype includes:

Generalized hypotonia, respiratory distress, high arch palate, left ear-grooves abnormal, prominent nasal bridge, left impatent nasolacrimal duct, dysmorphic face.

Chromosome analysis with peripheral blood was performed, while the karyotype was interpreted as 45,XX, der (14;15)(q10;q10)4p.out of two chromosome number 4, one Chromosome 4 has found deleted from 'p' arm small arm In wolf-hirschhorn Syndrome.

Molecular studies associated with spinal muscular atrophy and progressive muscular dystrophy also had negative findings. We suggest that rob(14;15)(q10;q10)4p could be Related to clinical presentation like this case.

Keywords: PGD, reproductive risks, Robertsonian translocation, Chromosomes, Down syndrome, Aneuploidy, Trisomy, Translocation (genetics), Mosaicism, Preimplantation diagnosis.

Introduction

American zoologist and cytogeneticist William Robertson (1881–1941) who first described a Robertsonian translocation in grasshoppers in 1916.² and called as full arm translocations or centric-fusion translocations.

In humans, Robertsonian translocations occur in the five Acrocentric chromosome pairs (chromosome pairs where the short arms are fairly short) namely 13, 14, 15, 21 and 22.

Robertsonian Translocations Are the commonest structural Chromosomal Abnormalities found in 1.23/1000 live births.

Translocations of two main types: reciprocal and Robertsonian.

Reciprocal translocations represent the exchange of chromatin blocks between two non-homologous chromosomes.

Robertsonian translocation involves two acrocentric chromosomes which fuse at the centromeric region and lose their short arms.

The peri-centric region of soft two acrocentric chromosomes fuse to form a Single centromere or double there sulting karyotype having 45 chromosomes including the Translocate done.

Case Presentation

A primi gravida (27year old female and 29 year old male) came for routine check up to the clinic.

The reports showed:

Normal TIFFA Scan: Placenta-Anterior, Liquor-Normal

Umbilical cord-Two arteries and one vein

BPD-45mm, HC-176mm, AC-150 mm, FL-33mm.

Normal Quadruplet Marker: Alpha fetoprotein-28.7ng/ml, Unconjugated Estriol (uS3) 1.68ng/ml, BETA-HCG 25968.0mLU/MI, Inhibin A Serum -282.6pg/ml.

Normal Triple Marker: Trisomy21-risk1:408, Normal N-T scan-2.9mm.

Fetal Anatomy:

Head

Mid line falx seen

Both lateral ventricles appears normal

Posterior fossa, cerebellum and cisternmagna appears normal.

No definite intracranial lesion seen.

Atria of lateral ventricles-6.0mm

Cisternamagna-4.9mm

Transcerebellar diameter-22mm.

Spine

Entire spine seen in longitudinal and transverse axis.

Vertebrae ands Pinal canal appears normal.

Face

Both orbits, nose and mouth appears normal

Heart

Heart appears with normal cardiac situs.

Four chamber view, three vessel view and outflow tracts appear grossly normal.

The baby was delivered with birth asphyxia and was admitted in NICU.

The Sample Was sent for karyotyping and that treatment given to baby was: IV Fluids, Inj. febrinil, Tab sildenafil.



Fig. 1

Discussion

Robertsonian translocation (ROB) is a chromosomal abnormality in which a certain type of a chromosome becomes attached to another chromosomes. It is the most commonest form of chromosomal translocation in humans, affecting 1/1,000 live births. It does not usually cause health problems, but can result in genetic disorders in some cases as Down syndrome and Patau syndrome.¹

In us, Robertsonian translocations occur in the five acrocentric chromosome pairs (chromosome pairs where the short arms are fairly short) named 13, 14, 15, 21 and 22.

The participating chromosomes break at their centromeres and the long arms fuse to form a single, large chromosome with a single centromere.

A Robertsonian translocation is involving two homologous (paired) or non-homologous chromosomes (i.e. two different chromosomes, not belonging to a homologous pair). A feature of chromosomes that are commonly found to undergo such translocations is that they possess an acrocentric centromere, partitioning the chromosome into

a large arm containing the vast majority of its genes, and a short arm with a much smaller proportion of genetic content. The short arms also join to form a smaller reciprocal product, which typically contains only nonessential genes also present elsewhere in the genome, and is usually lost within a few cell divisions. This type of translocation is cytologically visible, and can reduce chromosome number (from 23 to 22 pairs, in humans) if the smaller chromosome that results from a translocation is lost in the process of future cellular divisions. However, the smaller chromosome lost may carry so few genes (which are, in any case, also present elsewhere in the genome) that it can be lost without an ill effect to the individual.^{2,3}

In humans, when a Robertsonian translocation joins the long arm of chromosome 21 with the long arm of chromosomes 14 or 15, the heterozygous carrier is phenotypically normal because there are two copies of all major chromosome arms and hence two copies of all essential genes.⁴ However, the progeny of this carrier may inherit an unbalanced trisomy 21, causing Down syndrome.

About one in a thousand newborns have a Robertsonian translocation.⁵ The most frequent forms of Robertsonian

translocations are between chromosomes 13 and 14, 14 and 21, and 14 and 15.¹

A Robertsonian translocation in balanced form results in no excess or deficit of genetic material and causes no health difficulties. In unbalanced forms, Robertsonian translocations cause chromosomal deletions or addition and result in syndromes of multiple malformations, including trisomy 13 (Patau syndrome) and trisomy 21 (Down syndrome).

Most human with Robertsonian translocations having only 45 chromosomes in each of their cells, yet all essential genetic material is present, and they appear normal. Their children, however, may either be normal, carry the fusion chromosome (depending which chromosome is represented in the gamete), or they may inherit a missing or extra long arm of an acrocentric chromosome (phenotype affected). Genetic counseling and genetic testing is offered to families that may be carriers of chromosomal translocations.

Rarely, the same translocation may be present homozygously if heterozygous parents with the same Robertsonian translocation have children. The result may be viable offspring with 44 chromosomes.⁶

Conclusion

Genetic counseling is the process by which the patient so relatives at risk of an inherited disorder (or may be carrying a child at risk) are advised of the consequences and nature of the disorder, the probability of developing or transmitting it, and the option so pentohem in management and family planning.

Conflict of Interest: None.

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