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Case Report

Evidence of Rare Robertsonian Translocation (14; 21) Inherited From Mother: A Case Report from North-Eastern State Assam

Debajit Saikia[1], Giriraj Kusre[2]

[1]Senior Research Fellow, CFDMGD Lab, Department of Anatomy, Assam Medical College & Hospital, Dibrugarh, Assam, India.
[2]Associate Professor, Department of Anatomy, Assam Medical College & Hospital, Dibrugarh, Assam, India.

Corresponding Author: Debajit Saikia

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ABSTRACT

Robertsonian translocation is a rare event out of which maternally inherited ROBS is about 1%. Maternally inherited Robertsonian Translocations (14; 21) have not been reported from North East India. A female baby was born with sluggish reflexes, flat occiput and facial dysmorphism. In suspection of chromosomal abnormality the baby was subjected to cytogenetic evaluation at our lab. Age of the mother and father of the baby was 28 years and 33 years respectively. The baby was the first child of the parents. Parents were phenotypically normal. After karyotype of the baby was found to be 46, XX, t (14; 21) (q10:q10) and the karyotype of the mother was found to have 45, XX, t(14;21) (q10:q10) whereas her father was cytogenetically normal i.e. 46, XY. The heterozygous carrier for ROBs is phenotypically normal though the total number of chromosomes may be 45 but they can transmit the translocation to their baby along with an extra copy of 21 chromosome resulting in unbalanced translocation. This is supported by the present case as the baby had a karyotype of 46, XX, t (14; 21) (q10:q10) and her mother had a karyotype of 45, XX, t (14; 21) (q10:q10). ROBs may be sporadic or familial. In familial cases one of the parents is a carrier of balanced translocation. If such translocations are seen in the mother the recurrence rate of an unbalanced trisomy child is much higher compared to translocations of paternal origin.

Keywords: Robertsonian translocation, maternally inherited, cytogenetic evaluation, trisomy 21, North East India.

INTRODUCTION

Down Syndrome (DS) is the most frequent live born aneuploidy and recognizable form of mental retardation. The frequency of Down syndrome in Indian population is roughly estimated at 1 in every 920 live births with an annual incidence of 18,000 cases. [1] Almost 90% of DS cases are due to pure trisomy of the 21st chromosome and remaining cases are due to either mosaic cell line or unbalanced Robertsonian translocation (ROBS) involving the acrocentric groups of chromosomes especially chromosomes 14 and 21. [2] ROBS are either de novo in origin or are inherited from one of the carrier parents for that translocation. [3] Frequency of inherited translocation is about 1% of the total cases. [4] Robertsonian translocation is a rare event and maternally inherited ROBS have not been reported from North east India. Here we report a case of maternally inherited ROBS from this region.

CASE REPORT

A female baby born at 34th week of pregnancy with sluggish reflexes was referred to Neonatal care unit. On

examination the baby had flat occiput and facial dysmorphism. Suspecting chromosomal abnormality the baby was subjected to cytogenetic evaluation at Diagnostic Genetic lab. Age of the mother and father of the baby was 28 years and 33 years respectively. The baby was the first child of the parents. Parents were phenotypically normal. There was no history of any facial dysmorphism or mental retardation in the family. A pedigree chart was prepared (Figure 1).

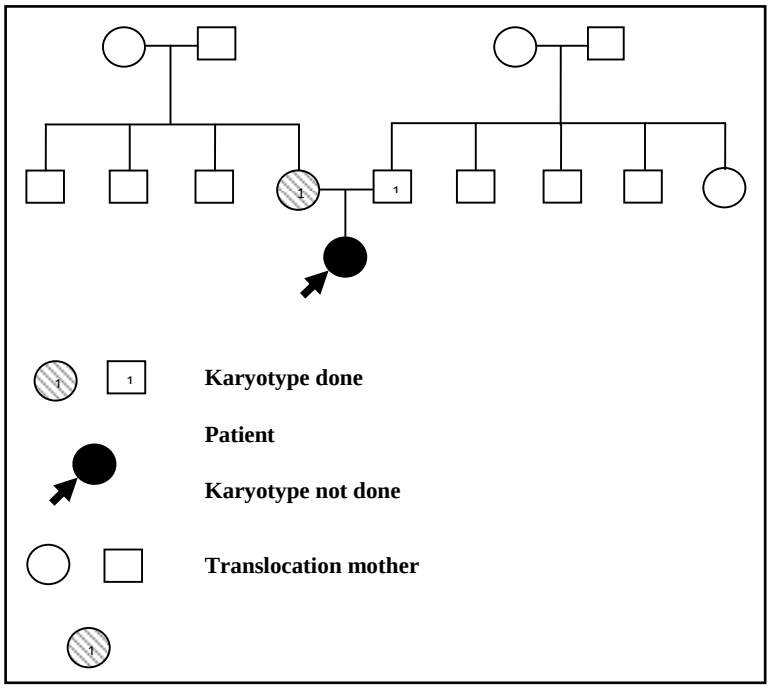

Figure 1: Pedigree chart of the patient

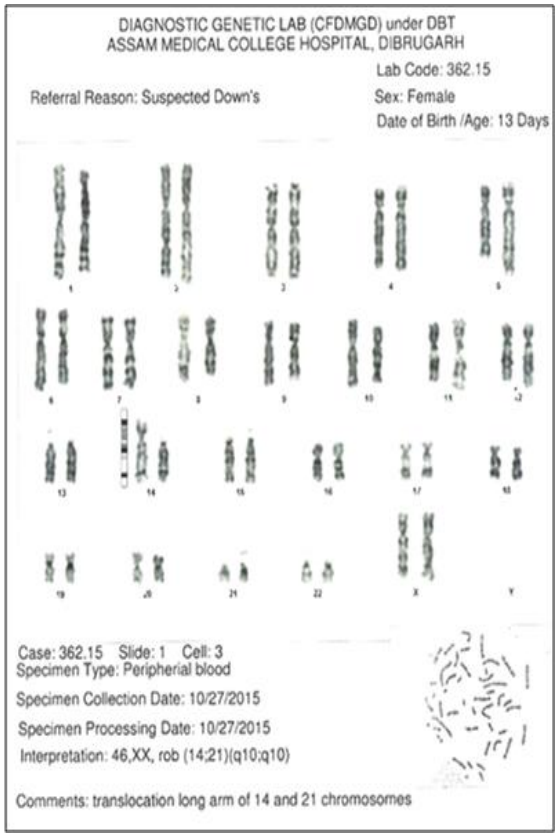

Figure 2: Karyotype of the patient

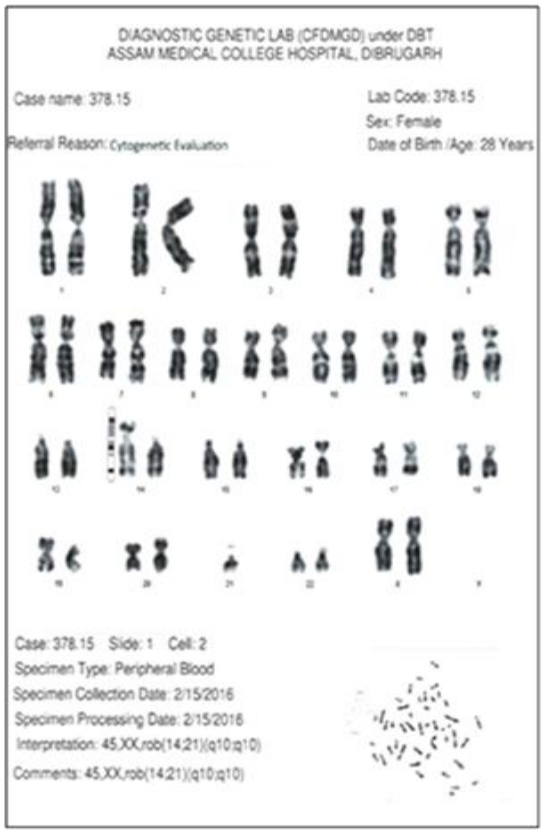

Figure 3: Karyotype of the mother

After obtaining signed informed consent, 3 ml of whole blood was collected in heparinised tube from the baby and her parents. The blood was cultured in peripheral blood culture medium (HikaryoXL™ RPMI Medium, HIMEDIA®), harvesting was done at 67 and half hours. Slides were prepared for GTG banding. The slides were stained by Giemsa's stain (HIMEDIA®) and 20 spreads were examined in the CytoVision® workstation (Leica DM6000B, Leica Microsystem, Germany). The karyotype of the baby was found to be 46, XX, t (14; 21) (q10:q10) (Figure 2). The parents were also cytogenetically analysed. The karyotype of the mother was found to have 45, XX, t(14;21) (q10:q10) (Figure 3) whereas her father was cytogenetically normal i.e. 46, XY. Cytogenetic analysis of the siblings of the mother was not done as consent could not be obtained.

Parents were informed about the condition and counselled. The risk of recurrence in subsequent pregnancies was also explained.

DISCUSSION

Most ROBs arise through adjacent chromatid exchanges corresponding to meotic chiasmata, in the pericentric regions of the acrocentric chromosomes. [5] The breakpoints of ROBs occur in the short arms of the participating chromosomes, leading to dicentric rearrangements. [6] In most organisms, dicentrics typically break during cell division; dicentric human chromosomes can be stable in mitosis and meiosis. This stability is reflected particularly in those chromosomes which are inherited. [7,8] The risk of transmission of D/G type of ROBs from carrier father is approx 1% and from that of carrier mother is approximately 10-15%. [9]

The heterozygous carrier for ROBs are phenotypically normal though the total number of chromosomes may be 45 as he/she has all the genes but they can transmit the translocation to their baby along with an extra copy of 21 chromosome resulting in unbalanced translocation. [10] In the present case the baby had a karyotype of 46, XX, t (14; 21) (q10:q10) and her mother had a karyotype of 45, XX, t (14; 21) (q10:q10).

In pure, the karyotype of the mother is normal and the average age of the mother is towards higher side, whereas the mother of the baby with inherited Robertsonian translocation has an abnormal karyotype and is of younger age. [11] As the mother of the present baby was young, phenotypically normal, had an abnormal karyotype and had the same type of translocation, the ROB (14; 21) in the baby must have been inherited from the carrier mother.

As inherited ROBS are inherited from phenotypically normal parents and carries a risk of recurrence in subsequent pregnancy, counselling of the parents and prenatal analysis should be attempted in all subsequent pregnancies.

CONCLUSION

Down syndrome is caused by trisomy of chromosome 21. Though more than 90% of the cases show free trisomy about 5-6% exhibit Robertsonian translocation. While free trisomy is attributed to rising maternal age the Robertsonian translocation is seen in young mothers. It may be sporadic or familial. In familial cases one of the parents is a carrier of balanced translocation. If such translocations are seen in the mother the recurrence rate of an unbalanced trisomy child is much higher compared to translocations of paternal origin. In familial cases of Robertsonian translocation close relatives should also be investigated for balanced carrier translocation status. The study emphasises the importance of chromosome examination in all Down syndrome infants, and in relatives when an unbalanced chromosome constitution is discovered.

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REFERENCES

1. Bandopadhyay Debasis, Bhatnagar Rajan. Robertsonian Translocation in a Down syndrome: A Case Report. Clinical Medicine and Diagnostics. 2015; 5(1):4-7.
2. Hanna EJ, Johnston WP, Nevin NC. Down syndrome associated with a familial 14/21 translocation. Ulster Med J. 1981; 50:95-98.
3. Ohno S, Trujillo JM, Kaplan WD, Kinosita R. Nucleolus-organisers in the causation of chromosomal anomalies in man. Lancet. 1961; 2:123-125.
4. Kolgeci S, Kolgeci J, Azemi M, *et al.* Cytogenetic study in children with down syndrome among kosova Albanian population between 2000 and 2010. Mat Soc Med. 2013; 25(2):131-135.
5. Therman E, Susman B, Denniston C. The non-random participation of human acrocentric chromosomes in Robertsonian translocations. Ann Hum Genet. 1989; 53:49-65.
6. Page SL, Shin JC, Han JY, *et al.* Breakpoint Diversity Illustrates Distinct Mechanisms for Robertsonian Translocation Formation. Hum Mol Genet. 1996; 5(9):1279-1288.
7. Shaffer LG and Lupski JR. Molecular mechanisms for constitutional chromosomal rearrangements in humans. Annu Rev Genet. 2000; 34:297-329.
8. Stimpson KM, Song IY, Jauch A, *et.* Telomere disruption results in non-random formation of de novo dicentric chromosomes involving acrocentric human chromosomes. PLoS Genetics. 2010; 6(8):e1001061.
9. Emery AE and Mueller RF. Elements of Medical Genetics; in Students note. 8th ed. ELBS: Churchill Livingstone; 1992.295.
10. Hassold T, Abruzzo M, Adkins K, *et.al.* Human aneuploidy: incidence, origin, and etiology. Environ Mol Mutagen. 1996; 28:167-175.
11. Mutton D, Alberman E, Hook EB. Cytogenetic and epidemiological findings in Down syndrome, England and Wales 1989 to 1993. National Down syndrome Cytogenetic Register and the Association of Clinical Cytogeneticists. J Med Genet. 1996; 33(5):387-94.

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