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# Case Report: A Stable dicentric chromosome, tdic (2: 21), in an Intellectually Disabled Female

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***Abstract***

***In human beings, dicentric chromosomes are occasionally encountered as stable chromosomal constitution except Robertsonian translocations that occur in an insignificant portion of the population. In present investigation, an 18 years female born to younger parents, who was naturally delivered but was under wt. after birth and had 42, XX, tdic (2; 21) chromosomal constitution is described. It is suggested that the stability of dicentric chromosome has been attributed to inactivation of chromosome no. 2 centromere. Stanford-Binet test was performed to ascertain the degree of intellectual disability and was found to be severe (IQ: 20-25 to 35-40). A reciprocal translocation which leads to dicentric chromosome formed from a chromosome 2 and a 21chromosome has not apparently been reported in individuals with intellectual disabilities.***

***Key Words: Dicentric chromosome, Intellectual Disability, Reciprocal Translocation, Chromosome, Disability***

## Introduction

Intellectual Disability (ID) occurs due to impairment of intellectual, social and linguistic abilities which are evident in infant stage [1, 2]. Prevalence of ID is about 2 to 3 percent of the population and numerous biological, including chromosomal abnormalities and socio-economic factors are the reasons behind the cause [3-6] but for about half of the diagnosed ID cases, the aetiological basis remains unclear. Chromosomal abnormalities accounts for 4–28% of ID cases. Among all the chromosomal anomalies, dicentric chromosomes are rarely found in patients with intellectual disabilities. Dicentric chromosomes are formed due to rearrangement of genomes that bring 2 centromeres together on one chromosome. The stability of dicentric chromosomes varies in different organisms. In humans, inactivation of one of the centromeres leads to stabilization of dicentric chromosome by creation a chromosome with single centromere that can segregate normally during division of cell.

**The objectives of Present Investigation are:**

- To ascertain the type of chromosomal abnormalities in an intellectually disabled female.
- To find out the degree of intellectual disability in the subject by Stanford- Binet test of intelligence
- To determine the origin of this abnormality in the subject.

## Material and Methods

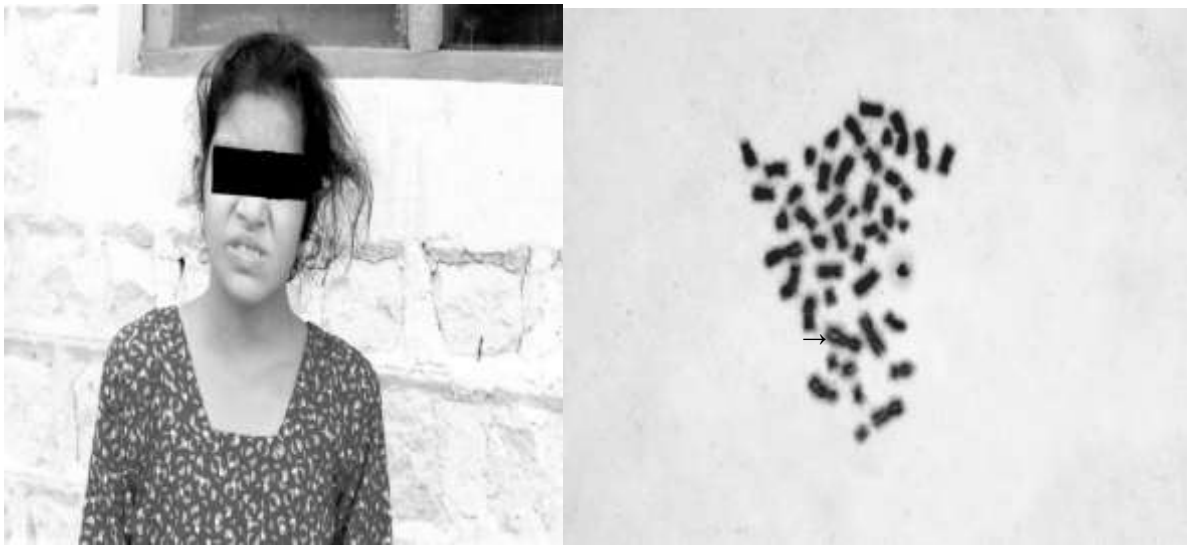
In this investigation, information about clinical features and family history was taken about the patient from her parents. IQ testing was done by Stanford- Binet test of intelligence. Age of the patient was 18 years. Blood samples of the subject and her parents were collected in sodium heparin vacutainers. Standard culture technique with some modifications [3] were followed for chromosomal preparation. Giemsa stain was used to stain the chromosomes and well spreaded chromosomal plates were selected for karyotype preparation. Photographs were taken by Leica Image analyser and karyotype preparation was were done manually and

ISCN 'International system for human cytogenetic nomenclature 2016' classification is followed to find chromosomal anomalies.

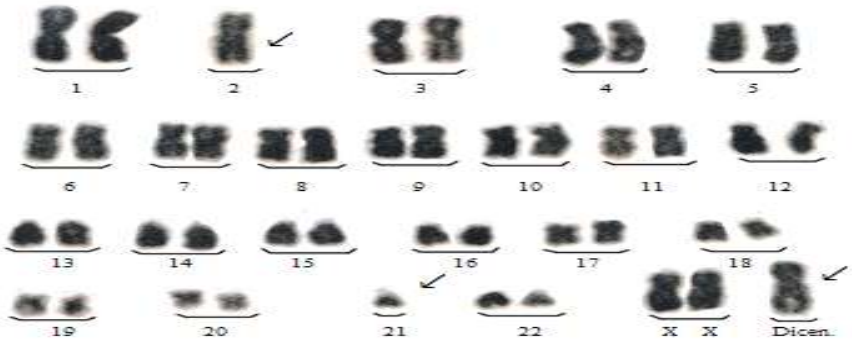
## Results and Discussion

### Case Report

Karyotype of a 18 yrs. old female was found to be 42, XX, tdic (2; 21) (Figure 1, A-C) whereas parents have normal karyotypes. She was first child, naturally delivered but under wt. after birth. Late birth cry was present. Age of mother and father during her birth was 29 and 32. No consanguinity was present. She had hypoactive behaviour and had history of epileptic seizures. Pedigree analysis hinted the sporadic cause of this abnormality (Figure 2) The degree of intellectual disability was found to be severe with IQ=33.



[A] [B]




[C]

**FIGURE 1**: [A] Subject [B] Chromosomal Plate used for preparation of karyotype

[C] Karyotypic Constituition: 45, XX, tdic (2: 21)

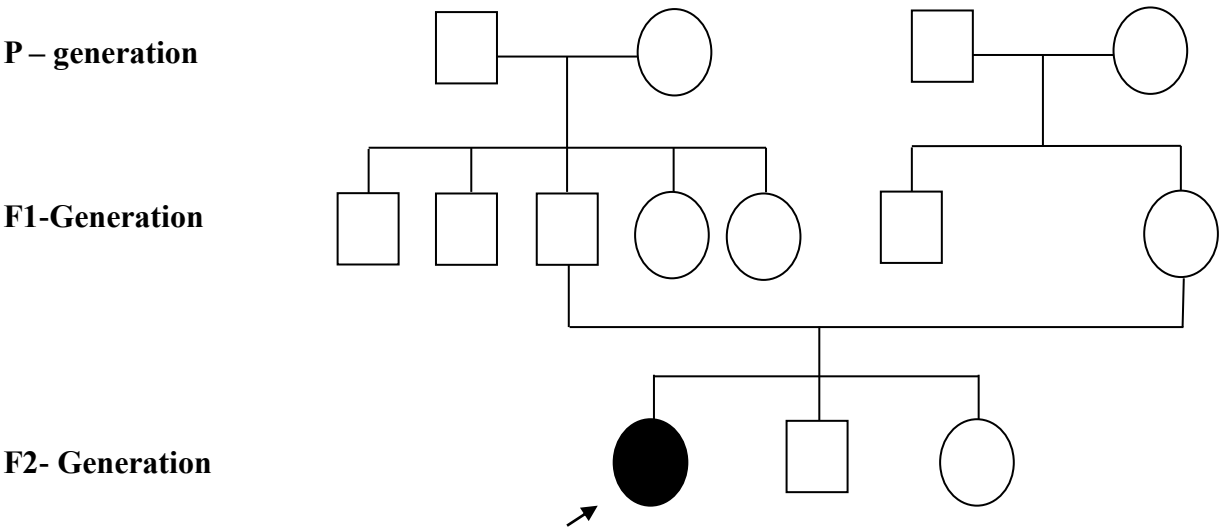



**FIGURE 2:** Pedigree Chart of the Subject

## Discussion

In dicentric chromosomes, inactivation of one of the centromeres leads to its stabilization and also prevents the formation of anaphase-bridge that result in non-disjunction. However, a small number of stable dicentric chromosomes have been reported [9,10]. The exact mechanism of inactivation of centromere is still not known. The abnormality found in present investigation is reported first time though other type of dicentric chromosomes i.e. tdic (8:22)(p23:p13), tdic(4;21)(pl6;q22) etc. are reported in several reports [11,12].From the foregoing discussion, author concluded that this abnormality is rarest among all the chromosomal abnormalities and never being reported before in individuals with ID. However, further studies are needed to unravel many unknown facts about molecular mechanism of inactivation of centromere that leads to the stability of these chromosomes.
For molecular cytogenetic diagnosis, this patient as well as her parents need revaluations as new techniques like FISH (fluorescent in situ hybridization) or MLPA (multiplex ligation dependent probe amplification) techniques and array-CGH (comparative genomic hybridization) techniques are present now.

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