JMSCR Vol||06||Issue||02||Page 200-201||February 2018

www.jmscr.igmpublication.org
Impact Factor (SJIF): 6.379
Index Copernicus Value: 71.58
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v6i2.30

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

Detection of Pericentric Inversion of Y chromosome in an Infant with Down Syndrome

Authors
Rajesh Kumar Srivastava[1], AK Kapoor[2]
[1]Senior Technologist, Cytogenetics laboratory, RML Mehrotra Pathology Pvt Ltd, Lucknow, U.P., India
[2]Pathologist, RML Mehrotra Pathology Pvt Ltd, Lucknow, U.P., India
Corresponding Author
Ashok Kumar Kapoor
D87 Mahanagar Extension, Vigyan Puri, Lucknow, U.P., India
Email: *drashokkapoor2016@gmail.com*

Abstract
An infant aged 18 days had trisomy of chromosome 21 along with pericentric inversion of Y chromosome. He failed to develop. Later, he died. Herewith, we report a case of chromosome 21 trisomy with percentric inversion of Y chromosome.

Introduction

Autosomal chromosomal 21 trisomy is known to be associated with various clinical features, e.g. bilateral epicanthic folds, bilateral Brushfield spots, big protruding tongue, high arched palate, short broad hands with single transverse crease, short neck, wide gap between 1st and 2nd toes, Hirschsprung megacolon, atrial septal defect and Alzheimer's disease. Clinically, the disease is known as Down syndrome. Rarely, it may be associated with pericentric inversion of Y chromosome. However, pericentric inversion of Y chromosome may occur alone without any functional derangement. In another study, 4 cases with pericentric inversion of Y chromosome were detected among 239 Down syndrome patients[1]. It gave an incidence of 1.67%. Herewith, we report a case of combined pericentric in (Y) with chromosome 21 trisomy.

Case Report

A newborn infant failed to thrive in spite of best possible care. He was hospitalized in a Pediatric ICU. Infant had all the clinical features of Down syndrome. Peripheral blood lymphocytes were stimulated using a mitogen-Phytohaemagglutinin[2]. Chromosome preparation was made. Giemsa banding was done (figure 1). It was used to pair and identify each chromosome[3]. Later, sequential QFQ (i.e. Q bands by fluorescence using Quinacrine) technique was done[4]. An unusual Y chromosome was detected (figure 2). Inversion of Y chromosome was confirmed using centromere banding. Chromosomal findings were interpreted as trisomy of chromosome 21 and pericentric inversion (an inversion requires two breaks in a chromosome. In pericentric inversion, breaks are on opposite sides of centromere). Later, the patient died.

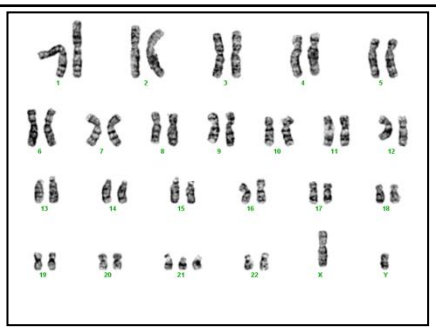

Figure 1

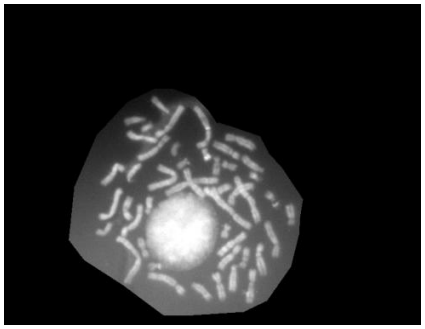

Figure 2

Discussion

Most important feature of this study was the detection of chromosomal anomalies both in an autosome as well as in a Y chromosome. Autosomal anomaly consisted of trisomy of 21 chromosome. Chromosome 21 trisomy is known to cause atrophy of superior temporal gyrus. Due to this anomaly, it results in development of various clinical features. Conversely, Y chromosomal inversion may be asymptomatic[4] or it may produce primary sterility[5]. In addition, subjects with pericentric Y chromosome inversion may have increased risk to have children with down syndrome[6]. Abnormal chromosome was considered to be a Y chromosome due to following reasons. First, it was found in a male patient. Second, size of abnormal chromosome was compatible with its derivation from a Y chromosome. Third, there was no normal Y chromosome. Fourth, it appeared to be pericentric inversion because it did not have any obvious phenotypic effect. In addition, only one chromosome was affected, which was against translocation. These features have also been described earlier in a separate study[7].

References

1. Sheth FJ, Shah ushma J, Desai Manisha J, Sheth JJ. Clinical profile of inversion Y in people of Gujarat, West India. Int J Hum Genet 2011; 11 (4): 245-248.
2. Arasaki DT, Sparkes RS. Microtechnique for culturing leucocytes from whole blood. Cytogenetics 1963; 2: 57-60.
3. Veerbhadrappa SK, Chandrappa PR, Roodmal SY, Shetty SJ, Madhu Shankari Gunjioganur S, Mohan Kumar Kumbar P. Karyotyping: Current perspectives in diagnosis of chromosomal disorders. Sifa Med J 2016; 3: 35-40.
4. Verma S, Rodriguez J, Dosik H. The clinical significance of pericentric inversion of the human Y chromosome: a rare “third” type of heteromorphism. The J of heredity 1982; 73: 236-238.
5. Jacobs PA. Structural abnormalities of the Y chromosome in man. Nature 1966; 210: 352-354.
6. Orye E, Verharren H. A peculiar t (21: 14) translocation, associated with a percentric inversion of the Y chromosome in mongol boy. Ann Genet 1974; 17: 217-220.
7. Sparkes RS, Muller HM, Veomett IC. Inherited pericenetric inversion of a human Y chromosome in trisomic Down’s syndrome. J Med Genet 1970; 7: 59-62.