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

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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 pericentric 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 patients¹. 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-Phytohaemagglutinin². Chromosome preparation was made. Giemsa banding was done (figure 1). It was used to pair and identify each chromosome³. Later, sequential QFQ (i.e. Q bands by fluorescence using Quinacrine) technique was done⁴. 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.
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 or it may produce primary sterility. In addition, subjects with pericentric Y chromosome inversion may have increased risk to have children with Down syndrome. 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.

References