FINAL DIAGNOSIS:

Chromosomal analysis of a 5-months-old baby with balanced chromosomal rearrangements along with phenotypic abnormalities [46,XY,t(3;10)(p21; p13)].

DISCUSSION

Chromosomal abnormalities are one of the major causes of birth defect in new born babies. Also, balanced translocations are frequent structural chromosomal abnormalities observed in patients with secondary infertility or recurrent spontaneous abortion[4]. We present a case with unique balanced autosomal translocation of t(3;10)(p21;p13). The index case presented with dysmorphic facial features and had a balanced translocation 46,XY,t(3;10)(p21;p13). This translocation with break points at (p21;p13) has not been reported in the literature in a child with facial dysmorphism. The risk of phenotypic abnormality from a de novo balanced translocation has been estimated at approximately 7% [5].To the best of our knowledge, this is the first report of novel balanced translocation t(3;10) with break points in a child with dysmorphic features.

The most common causes of dysmorphic feature in a new born are Down's syndrome, Patau syndrome, and other unbalanced aberrations[6,7]. To the best of our knowledge, the association of reciprocal balanced translocation of p arm of chromosome 3 and p arm of chromosome 10 with dysmorphic features has never been reported in the literature.

Previously, two balanced chromosomal rearrangements were reported in a 17-week fetus by prenatal karyotyping of amniocytes. The fetal karyotype was 46,XX,t(2;16) (q33;q24) along with inv(7)(p15q11.23). Parental karyotypes were normal, indicating a strong evidence of de novo origin of chromosomes rearrangements in the fetus. The pregnancy was terminated because of the detection of intrauterine growth retardation. In the absence of additional experience, the minimum presumed risk of phenotypic abnormality for de novo, multiple, or complex chromosome rearrangements identified prenatally may be estimated as the additive risk of the number of chromosome breakpoints involved[8].