Two Familial Cases with Trisomy 15q dist Due to a rep(5;15)(p14;q21)

M. Tzancheva¹, M. Krachounova¹, and Zv. Damjanova²

¹Department of Medical Genetics, Medical Academy, 8 Belo More Str 1040, Sofia, Bulgaria
²Department of Pediatrics, 1st Pediatric Clinic, Medical Academy, 1, G. Sofijski Street, Sofia, Bulgaria

Summary. A trisomy of the distal long arm of chromosome 15 (q21–qter) resulting in similar phenotypic and developmental abnormalities in two related children (a boy and a girl) is described. The chromosome defect was due to malsegregation of a balanced translocation (5;15)(p14;q21) in one of the parents. It was inherited in four generations and accompanied by recurrent miscarriages. Comparison of these patients with four previously published cases of trisomy 15q dist reveals a pattern of common features including: microdolichocephaly with characteristic strikingly protuberant occiput and predominance of the visceral over the cerebral cranium; peculiar facial dysmorphism—narrow antimongoloid palpebral fissures; hypertelorism and convergent strabismus; voluminous nose with upward nostrils and nasal septum pending downward; large mouth with long philtrum without expressed design and tight lips; high hard palate; large, malformed and asymmetrically low-set ears; micrognathy. Furthermore the following were also observed: short neck, cardiopathy, unilateral hydrops gravidarum. The examination at the age of six months revealed good physical development (weight 7,900 g, length 69 cm, head circumference 45.5 cm). The following abnormalities were observed: facial dysmorphism, dolichocephaly, facial dysproportion (visceral larger than cerebral cranium), projection in the cheek-bone region; rough facial features; high, projected forehead; strikingly prominent occiput; narrow antimongoloid palpebral fissures; hypertelorism and convergent strabismus; voluminous nose with upward nostrils and nasal septum pending downward; large mouth with long philtrum without expressed design and tight lips; high hard palate; large, malformed and asymmetrically low-set ears; micrognathy. Furthermore the following were also observed: short neck, cardiopathy, unilateral

Introduction

Cases of partial trisomy of the distal part of the long arm of chromosome 15 are rarely observed. The present paper reports on two related patients with partial trisomy 15q dist, resulting from malsegregation of a balanced translocation (5;15)(p14;q21) in one of the parents. The translocation is traced in members of four generations and is characterized by recurrent miscarriages.

Case Reports

Patient 1. V.I.I. was a boy born at term from a first pregnancy, which was threatened by miscarriage in the third month due to...
Simian creases, muscle hypertonia, profound encephalopathy with lack of suck and swallow reflexes. The skull X-ray examination revealed arched backward projection of the skull roof, and strong thinning of lamina occipitalis interna and externa. The vertebral column X-ray showed irregular body shape of the first and fifth and no sixth and seventh cervical vertebra. The patient died three days after birth. Autopsy revealed microgyri and internal hydrocephalus, malformation of the back cranial pit, and open ductus Botalli (4 mm).

Pedigree and Cytogenetic Examinations

The patients were children of a brother and sister (III-8 and III-9). The pedigree was notable for the many recurrent miscarriages (Fig. 3). There was also one child (III-2) who died at seven months of age from a heart defect. (It is possible that the child had the same chromosome disease.)

Chromosome analysis after G-band staining using the Seabright banding technique (1971) revealed unbalanced karyotypes in both patients representing one derivative chromosome 5 with a prolonged short arm (Fig. 4a). The karyotype of Patient 1’s mother (III-9) included a balanced translocation, probably reciprocal, between the short arm of chromosome 5 and the long arm of chromosome 15 (Fig. 4b)—rcp(5;15)(p14;q21).

The same balanced translocation was found in her brother (III-8), Patient 2’s father. Based on this Patient 1’s karyotype could be interpreted as 46,XY,der5,rcp(5;15)(p14;q21)mat and Patient 2’s karyotype as 46,XX,der5,rcp(5;15)(p14;q21)pat. Therefore Patients 1 and 2 are partially trisomic for the distal part of the long arm of chromosome 15 (q21–qter).