

**CASE REPORTS**

# A Case Report of 9p Deletion Syndrome Associated with Partial Trisomy of 1q42

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## ABSTRACT

We report a case of partial deletion of 9p with partial trisomy of 1q42 syndrome, which is a rare clinical and cytogenetic report. The dysmorphic features of the patient include microcephaly, plagiocephaly, trigonocephaly with metopic ridge, arched eyebrows, hypertelorism, down-slanting palpebral fissure, ptosis, blepharophimosis, unilateral left epicanthic fold, long eyelashes, low-set and posteriorly rotated ears, long philtrum, anteverted nares, retrognathia and unilateral undescended testis. Chromosomal analysis revealed partial monosomy of 9p24 associated with partial trisomy of 1q42→qter.

**Keywords:** 9p deletion, trisomy of 1q42, mental retardation, microcephaly, trigonocephaly

## INTRODUCTION

**D**eletion 9p syndrome was firstly reported by Alfi *et al* in 1973 (1). In 1976, he described six cases of patients with terminal deletion of the 9p22 that was associated with mental retardation, trigonocephaly, upslanting palpebral fissure, wide flat nasal bridge, anteverted nostrils, long upper lip, short neck, and long digits (2). Thereafter, there were many reports describing birth defects and cerebral maldevelopment in pa-

tients with terminal deletion of the short arm of chromosome nine, either pure 9p deletion or associated with some other chromosomal rearrangement (3-7). In 1988, Huret *et al* reported 11 cases of deletion 9p of which seven had pure 9p deletion and four had 9p deletion associated with another chromosome rearrangements (8). The reported associated unbalanced chromosome segment in unrelated patients were trisomy 13q (four cases), trisomy 12q (three cases), trisomy 10q (three cases), trisomy 2q (two cases), trisomy 5p (two cases), trisomy 5q (two cases), trisomy 7q (two cases), and

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two trisomy 16q (two cases) (8). Our patient had partial trisomy of 1q42.  $\square$

### CASE REPORT

A 3-year-old boy was referred to our clinical Genetic Department because of psychomotor developmental handicap. Following an uneventful pregnancy, he was born via normal vaginal delivery at term to a non-consanguineous marriage. The patient's birth weight and length were normal (3000 grams and 51 cm, respectively). He had a normal head size at birth with head circumference (HC) of 36 cm, that is, the 75 percentile for sex and age. However, according to the health chart records, there was a regression of HC after 8 months of age. He was unable to sit till two years of age. At the time of visit (3 years of age), his physical examination was remarkable for marked microcephaly (HC: 46 cm, that is, under -2 SD for sex and age), plagiocephaly, trigonocephaly with metopic ridge, arched eyebrows, hypertelorism, down-slanting palpebral fissure, ptosis, blepharophimosis, unilateral left epicanthic fold, long eyelashes, low-set and posteriorly rotated ears, long philtrum, anteverted nares, retrognathia and left unilateral undescended testis (Figure 1). The palmar dermatoglyphics pattern was normal. He had no seizure and his vision and hearing were normal.

His neurologic examination revealed profound developmental handicap, with inability to walk. He was unable to utter even a single word. His brain MRI showed chronic bilateral fronto-temporal subdural hematoma with mild frontal atrophy. His metabolic screening was normal.

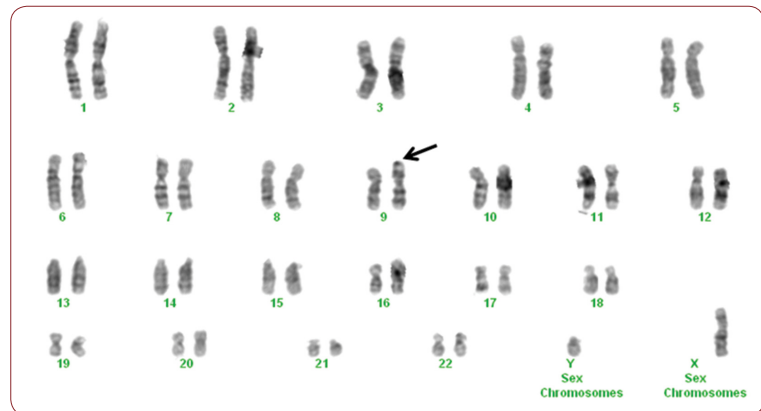
GTG banding karyotype carried out on the patient revealed derivative chromosome 9, that is, 46,XY,der(9) (Figure 2). Consequently, a chromosomal study was done on both parents to determine the origin of the rearrangement. His father's karyotype was normal (46,XY) but his mother's showed translocation between chromosomes one and nine [46,XX,t(1;9)(q24;p23)] (Figure 3). Accordingly, we concluded that the patient had partial monosomy for 9p24 and partial trisomy for 1q42→qter.  $\square$

### DISCUSSION

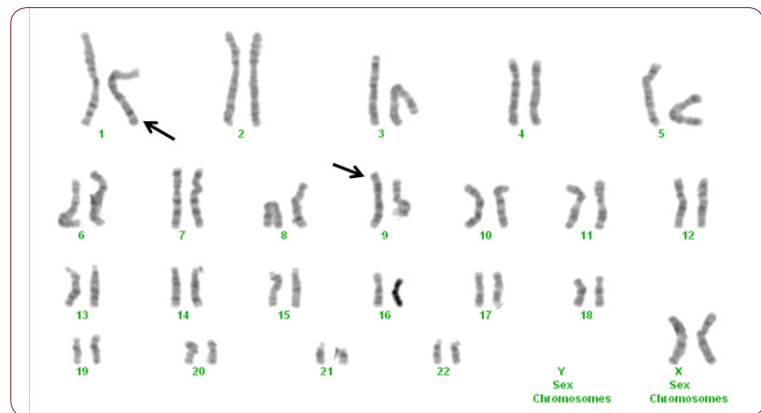
Considering the time of onset, associated dysmorphic features, and cytogenetic finding,



**FIGURE 1.** The front and lateral view of our case with partial trisomy of 1q42→qter, and partial monosomy of 9p24. There are some distinct dysmorphic features, including plagiocephaly, trigonocephaly with metopic ridge, arched eyebrows, hypertelorism, down-slanting palpebral fissure, ptosis, blepharophimosis, unilateral left epicanthic fold, long eyelashes, long philtrum, anteverted nares, retrognathia, and low-set posteriorly rotated ears.



**FIGURE 2.** The karyotype of the patient has revealed derivative chromosome 9



**FIGURE 3.** The mother karyotype showed translocation between chromosomes one and nine [46,XX,t(1;9)(q24;p23)]

our case has postnatal-onset syndromic microcephaly due to chromosomal abnormality. Besides microcephaly, distinct dysmorphic findings in our patient included arched eyebrows, hypertelorism, down-slanting palpebral fissure, ptosis,

blepharophimosis, low-set posteriorly rotated ears, long philtrum, anteverted nares, retrognathia and unilateral undescended testis.

According to a review by Huret *et al* on 80 cases with different length of 9p deletion, it seems that dysmorphic features do not differ with regard to the length of the deletion (8). All of 39 cases with pure 9p deletion described by Huret *et al* were de novo and common dysmorphic features included trigonocephaly, upslanting palpebral fissures, and long philtrum (8). However, besides trigonocephaly and long philtrum, our patient had down-slanting palpebral fissures. In our case, down-slanting palpebral fissure is in contrast with other reports that described upslanting palpebral fissures in 9p deletion syndrome.

Huret *et al* have also reviewed 41 cases of del (9p) with a partial trisomy, of which 38 were inherited from a carrier parent with balanced translocation (mother as a carrier in 25 cases, father as a carrier in 13 cases). In 23 out of 38 cases, the breakpoint occurred mainly at 9p24, which has not been reported in cases with pure 9p deletion. In pure 9p deletion, the most common breakpoint was at 9p22 (8). This breakpoint, that is 9p24, is compatible with our find-

ing where there was translocation between chromosomes one and nine.

Chew and Thong reported two cases of partial deletion 9p syndrome with trigonocephaly, arching eyebrows, anteverted nares, long philtrum, abnormal ear lobules, congenital heart lesions and digital anomalies (9). Trigonocephaly, arching eyebrows, anteverted nares and long philtrum are in common with our patient's dysmorphic features.

Monosomy of distal 9p has been shown to be associated with a wide range of gonadal dysgenesis such as hypogonadism, streak gonads, cryptorchidism, hypoplastic testes, micro-genitalia, and sex reversal (10, 11). Our patient also had unilateral cryptorchidism.

To the best of our knowledge, our case is the first report of association of 9p24 deletion syndrome with 1q42 partial trisomy in a patient with constellation of congenital birth defects.

Pilling up of clinical information of cases with similar chromosomal abnormalities, it would be helpful to understand the genetic basis of distinct dysmorphic features. □

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