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SHORT COMMUNICATION

A PARTIAL TRISOMY 2p(p21→pter) DERIVED FROM A PATERNAL t(2;4)(p21;q33) KARYOTYPE

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ABSTRACT

We describe a 7-year-old boy with additional material on 4q whose karyotype was 46,XY,der(4) t(2;4)(p21:q33). The father's karyotype was 46,XY,t(2;4)(p21;q33) and the mother's was normal. These results indicate that the extra material on 4q in the patient originated from the father's chromosome 2p. The patient had dysmorphic facial features, prominent ears, long fingers, developmental delay, speech delay and suffered from seizures.

Key words: Trisomy 2p, Dysmorphic facial features

INTRODUCTION

Trisomy duplication of 2p is associated with a characteristic syndrome as described in (1-8). Signs and symptoms mentioned in these sources for the chromosome 2p duplication syndrome include: small head; prominent high forehead; dysmorphic ears; low-set ears; widely spaced eyes; strabismus; short nose, broad nasal bridge; anecephaly, long, tapering, hyperflexible fingers; limited hip movement; scoliosis; reduced muscle tone; congenital heart defect; bronchial aplasia; pulmonary aplasia; undescended testes and small penis in males, mental and growth retardation, and neuroblastoma (1-8). We present here a patient with distal partial trisomy 2p that resulted from a familial balanced reciprocal translocation.

CASE REPORT

A 7-year-old male was referred to our clinic for developmental and psychomotor delay. He was born after a normal gestation to a normal and healthy couple who were not consanguineous. He was born at term by natural delivery.

At the time of diagnosis his weight was 14.5 kg, his height was 100 cm, and his head circumstance was 49.5 cm. He had a prominent high forehead, maxillary hypoplasia, small mandible, widely spaced eyes, pectus excavatum, long fingers, undescended testes, small penis, reduced muscle tone, slender body, and was mentally and developmentally retarded (Figure 1). Both mother and father had an apparently normal phenotype with no history of major clinical abnormalities in their families.

Cytogenetic Study. Chromosome study was

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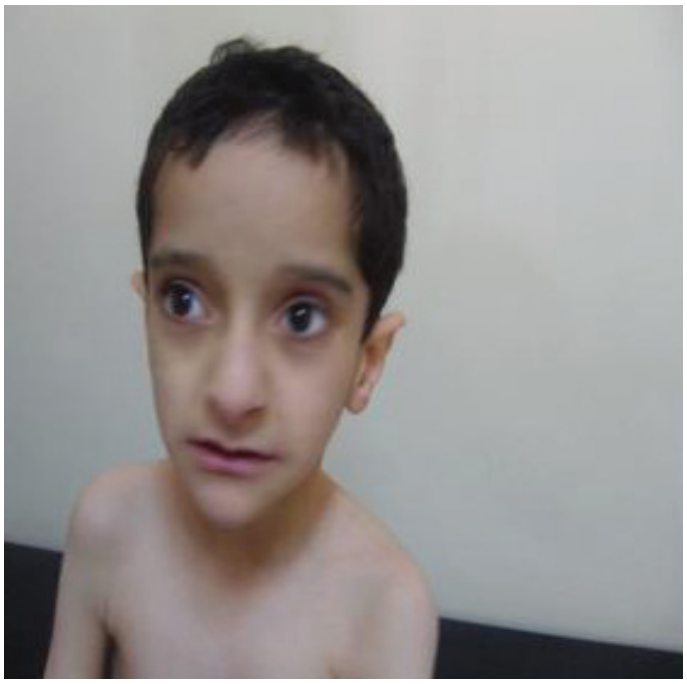


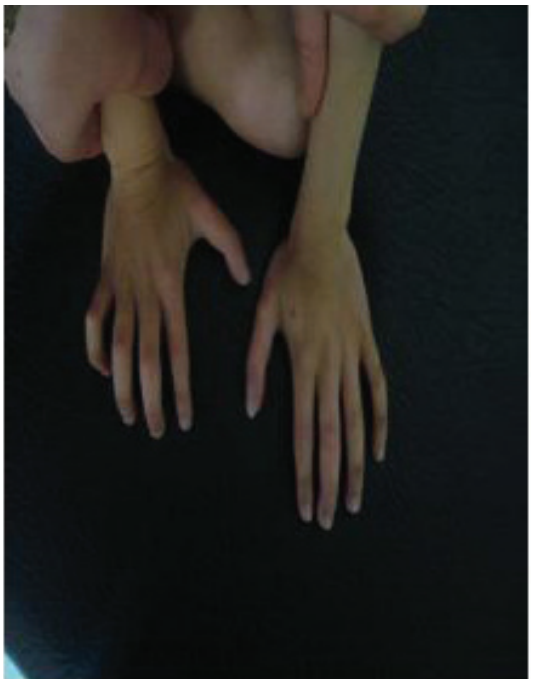


Figure 1. The proband at age 7.

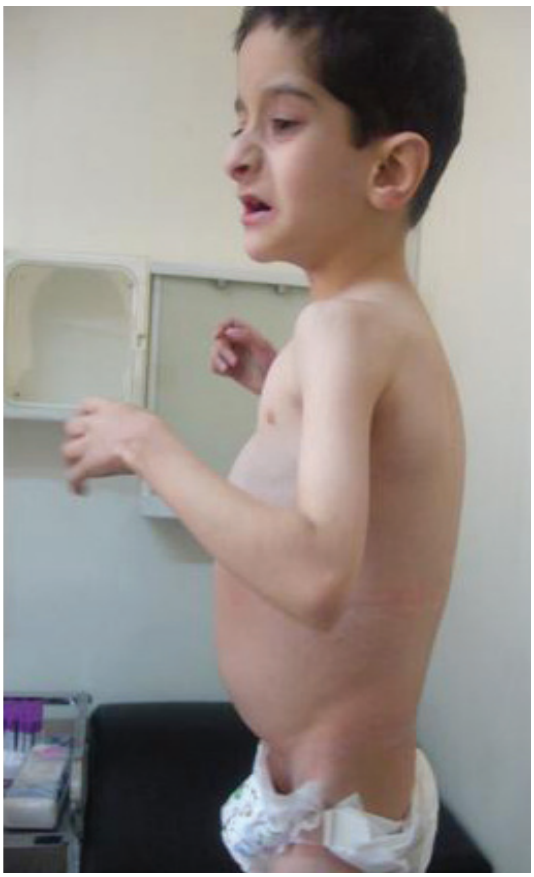
performed on a peripheral blood leukocyte culture according to standard protocols (9). The proband's karyotype was 46,XY,der(4),t(2;4)(p21;q33) (Figure 2). The father's karyotype was 46,XY,t(2;4)(p21:q33) (Figure 3), while the mother's karyotype was normal. We concluded that the extra chromosomal material in the patient was derived from the father.

DISCUSSION

Our patient had a derivative chromosome 4, that arose from a paternal balanced reciprocal translocation, *i.e.,* 46,XY,t(2;4)(p21;q33). As a consequence, the clinical features associated with the presence of an extra 2p21 to 2pter region are seen in the patient.

Carriers of balanced translocations do not generally reveal any abnormal phenotype. However, they are at risk of infertility/miscarriages. Furthermore, they may have child or children who inherit either one of the aberrant chromosomes with the partial monosomy or one of the abnormal chromosomes with partial trisomy as in our case. These chromosomal segments might contain or lack a variety of important genes that are able to cause multiple congenital malformations in the outgoing generations. A summary of all clinical findings in similar published reports is listed in Table 1.

Partial trisomy of different segments of 2p with some uncommon features has been presented. For example,



trisomy 2p21→p25 is associated with broncho-pulmonary hypoplasia (17); trisomy 2p23 with flat, wide glabella and depressed nasal bridge (3); trisomy 2p24 with neural

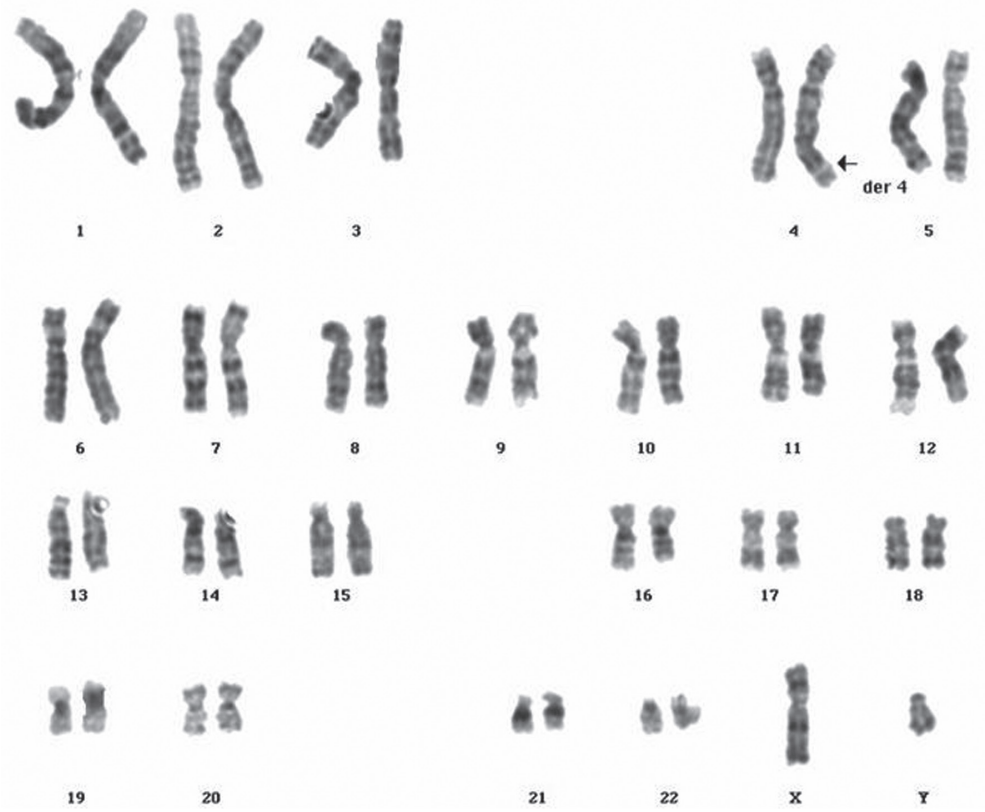


Figure 2. The karyotype of the proband.

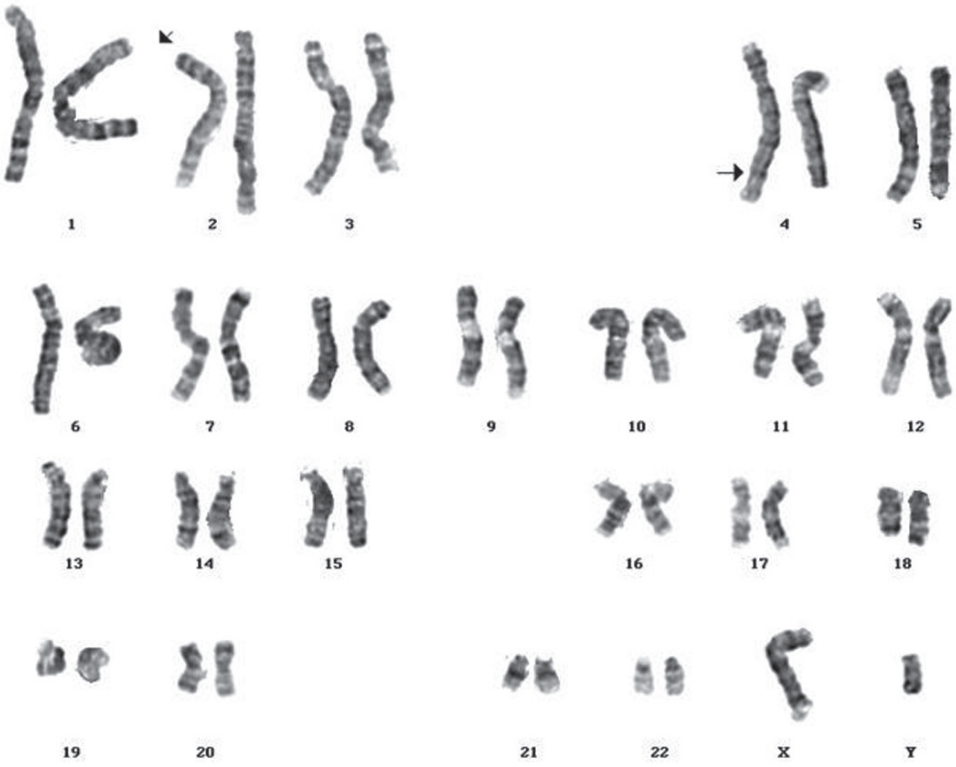


Figure 3. The karyotype of the father.

Table 1. A summary of clinical findings of patients with partial duplication of 2p.

Region	Sex-Age	Signs/Symptoms	References
2p21→2pter	F-4	Mental retardation; short stature; congenital heart disease; dysmorphic facial features	10
2p23→2pter	F-7 months	Prominent forehead with a flat hemangioma; depressed nasal bridge; protruding tongue; posteriorly angulated ears; retinal hypopigmentation; esotropia with poor abduction of the right eye; foveal hypoplasia; striking left optic nerve hypoplasia	11
2p21→p24.2	F-18 months	Microcephaly; open fontanelles; prominent forehead; flat occiput; hypertelorism; sparse eyebrows; small nose with depressed nasal bridge; bulging philtrum; thin upper lip, high arched palate; low-set and posteriorly rotated ears; small mandible, short neck with a a low hairline; eye malformations	12
2p25→p23	F-53 months	Microcephaly; dilated leteral horns of the cerebral ventricles; transient cortical blindness; myopia; muscle hypotonia; dilatin of the left renal collecting system	13
2p25.3→2pter	M-at birth	Mental and growth retardation; psychomotor delay; microcephaly; ptosis, micrognathia, a nalow palate and cryptorchidism	15
2p21→2pter	M-7	Mental retardation; prominent high forehead; maxillary hypoplasia; small mandible; widely spaced eyes; pectus excavatum; long fingers; undescended testes, small penis; slender body; seizures	Our study

tube defects and hand and foot deformities (16); trisomy 2p23→2p25 with myopia (12); trisomy 2p25 with cryptorchidism and small penis in males (15), trisomy 2p23→ p24 with abnormal rotation of the heart or L-transposition of large vessels (with or without visceral heterotaxia (17).

The human prolactin regulatory element binding (PREB) protein gene located on 2p23 may have a role in human development and abnormal dosage of this transcription factor. It may also be involved in developmental abnormalities of the face, skeletal defects, growth and mental retardation, congenital heart and neural tube defects, and abnormalities of the genitalia (18).

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REFERENCES

1. Cassidy SB, Heller RM, Chazen EM, Engel E. The chromosome 2 distal short arm trisomy syndrome. J Pediatr 1977; 91(6): 934-938.

2. Francke U, Jones KL. The 2p partial trisomy syndrome. Duplication of region 2p23 leads to 2pter in two members of a t(2;7) translocation kindred. Am J Dis Child. 1976; 130(11): 1244-1129.

3. Francke U. Clinical syndromes associated with partial duplications of chromosomes 2 and 3: dup(2p), dup(2q),dup(3p),dup(3q). Birth Defects Orig Artic Ser. 1978; 14(6C): 191-217.

4. Armendares S, Salamanca-Gómez F. Partial 2p trisomy (p21 leads to pter) in two siblings of a family with a 2p-:15q+ translocation: Clin Genet. 1978; 13(1): 17-24.

5. Zuñiga R, Torres de Caballero OM, Mondragon A. Direct duplication 2p14 to 2p23. Hum Genet. 1979; 48(2): 241-244.

6. Kishi K, Matsutani Y, Iwatsubo T, Nishimura K, Tonomura A. A case of partial trisomy 2p (region 2p21 to 2pter) derived from a maternal t(2;15)(p21;q26). Jinrui Idengaku Zasshi. 1980; 25(1): 47-53.

7. Nagano H, Kano Y, Kobuchi S, Kajitani T. A case of partial 2p trisomy with neuroblastoma. Jinrui Idengaku Zasshi. 1980; 25(1): 39-45.

8. Parruti G, Di Ilio C, Calabrese G, Stuppia L, Guanciali Franchi P, Aceto A, Palka G. A new case of partial 2p trisomy due to de novo interstitial duplication 2p21-22. Ann Genet. 1989; 32(1): 55-58.

9. Barch MJ, Knutsen T, Spurbeck JL: The AGT Cytogenetics Laboratory Manual. Philadelphia: Lippincott-Raven Publishers, 1997.

10. Pueschel SM, Scola PS, Mendoza T. Partial trisomy 2p. J Ment Defic Res. 1987; 31(Pt3): 293-298.

11. Al-Saffar M, Lemyre E, Koenekoop R, Duncan AM, Der Kaloustian VM. Phenotype of a patient with pure partial trisomy 2p(p23→pter). Am J Med Genet. 2000; 94(5): 428-432.

12. Mégarbané A, Souraty N, Prieur M, Theophile D, Chédid P, Augé J, Vekemans M. Interstitial duplication of the short arm of chromosome 2: report of a new case and review. J Med Genet. 1997; 34(9): 783-786.

13. Aviram-Goldring A, Fritz B, Bartsch C, Steuber E, Daniely M, Lev D, Chaki R, Barkai G, Frydman M, Rehder H. Molecular cytogenetic studies in three patients with partial trisomy 2p, including CGH from paraffin-embedded tissue. Am J Med Genet. 2000; 91(1): 74-82.

14. Le Caignec C, Winer N, Boceno M, Delnatte C, Podevin G, Liet JM, Quere MP, Joubert M, Rival JM. Prenatal diagnosis of sacrococcygeal teratoma with constitutional partial monosomy 7q/trisomy 2p. Prenat Diagn. 2003; 23(12): 981-984.

15. Chen CP, Liu FF, Jan SW, Lin SP, Lan CC. Prenatal diagnosis of partial monosomy 3p and partial trisomy 2p in a fetus associated with shortening of the long bones and a single umbilical artery. Prenat Diagn. 1996; 16(3): 270-275.

16. Hahm GK, Barth RF, Schauer GM, Reiss R, Opitz JM. Trisomy 2p syndrome: a fetus with anencephaly and postaxial polydactyly. Am J Med Genet. 1999; 87(1): 45-48.

17. Lurie IW, Ilyina HG, Gurevich DB, Rumyantseva NV, Naumchik IV, Castellan C, Hoeller A, Schinzel A. Trisomy 2p: analysis of unusual phenotypic findings: Am J Med Genet. 1995; 55(2):229-236.

18. Taylor Clelland CL, Levy B, McKie JM, Duncan AM, Hirschhorn K, Bancroft C. Cloning and characterization of human PREB; a gene that maps to a genomic region associated with trisomy 2p syndrome. Mamm Genome. 2000; 11(8): 675-681.