31. A Case of Partial Trisomy for the Long Arm of Chromosome 9

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Currently five cases associated with genuine partial trisomy for the long arm of chromosome 9, or partial trisomy 9q, are referred to in the literature (Turleau *et al.* 1975; Faed *et al.* 1976; Šubrt *et al.* 1976; Chamla *et al.* 1977). We wish to describe in this article an additional case with an abnormal chromosome no. 1 carrying an inserted portion of the long arm (9q12→9q13) as partial trisomy 9q.

Case reports. The propositus was a thirteen-year-old boy, 32 Kg in weight, with circumferences of 51 cm in head, 66 cm in breast, and 133 cm in length. He was born to a 22-year-old father and a 18-year-old mother as the first child. There was three spontaneous abortions in the first trimester, but no consanguinity in this couple. Both of her mother and father were phenotypically normal on the clinical examination.

Following clinical features were remarkable in the affected child: Imbecile, repeated convulsion, hard-dry and goose-fresh skin, dolichocephalia, flat occiput, upward slanted palpebral fissure, blepharophthalmosis, weak-sight, enlarged nasal bridge, irregular placement of teeth, low set ears, incomplete development of external genitalia, cryptorchism, and small penis (Fig. 1). Electrocardiogram was normal.

Cytological findings. Chromosome preparations were made from leucocyte cultures established from the propositus and his parents. For differential staining, the G-banding procedure was applied to air-dried slides prepared from the above subjects. Chromosome counts were done on 60 well-delineated metaphases in each sample. Karyotypes were analysed on 24 cells in each based on conventional Giemsa and G-banding preparations.

The karyotype of the proband showed 46 chromosomes in which one no. 1 chromosome was outstanding by its enlarged long arm. The unusual element could not be assessed until the maternal karyotype came under study. The examination of parents indicated that the

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mother was the balanced carrier of direct insertion of a long arm segment of no. 9 into a long arm of no. 1 chromosome. The G-banding

Fig. 1. Phenotypes of the propositus.

analysis revealed that the segment q12q13 of chromosome 9 was missing and that this segment was inserted into the long arm of chromosome 1. Then, the formula of the mother was given as 46,XX,ins(1; 9)(q11; q12q13) (Fig. 3).

Based on the result from the maternal translocation, the aberration found in the propositus was interpreted as partial trisomy for

Fig. 2. G-banding karyotype of the propositus having an aberrant no. 1 chromosome which is a partial trisomy for long arm of no. 9 chromosome: 46,XY,der(1),ins(1; 9)(q11; q12q13)mat.

Fig. 3. G-banding karyotype of the mother having a direct insertion into a long arm of no. 1 chromosome from a long arm of no. 9: 46,XX,ins(1; 9)(q11; q12q13).

the long arm of 9. The formula was shown as 46,XY,der(1),ins(1; 9)(q11; 12q13) mat (Fig. 2).

The father of the propositus was apparently normal in his karyotypes without showing any detectable chromosome change.

Remarks and conclusion. Recent banding techniques for the chromosome studies have served as very essential tools for the identification of the chromosome, as well as for the determination of translocation and of derivative chromosome.

The cases with partial trisomy for a long arm of no. 9 chromosome without association with trisomy 9q are rarely referable to the recent literature.

Turleau *et al.* (1975) reported two unrelated patients with a partial trisomy for a long arm of chromosome 9. One patient had the duplication of the region q11q33, while the other showed trisomy for the segment q31qter. Faed *et al.* (1976) dealt with a case with partial trisomy for a long arm of chromosome 9 carrying a substantial portion of the long arm (q12→q32) present in duplicate. Šubrt *et al.* (1976) reported a case with partial trisomy 9q which showed trisomy for the segment q32qter. Chamla *et al.* (1977) described a case with partial trisomy 9q involving the 9q11→9q32 region.

The major physical features of our case are shown in Table I in comparison with those reported in cases with partial trisomy 9q by the above-mentioned authors. It is evident that the clinical features are rather divergent depending on the difference of 9q segments.

The partial trisomy 9q observed in our case was shown to involve

Table I. Major clinical features of partial trisomy 9q patients described by different authors

	Turleau <i>et al.</i> (1975)				Faed <i>et al.</i> (1976)	Šubrt <i>et al.</i> (1976)	Chamla <i>et al.</i> (1977)	Present case
	Case 1	Case 2						
Trisomic segment	q11→q33	q31→qter	q12→q32	q32→qter	q11→q32	q12→q13		
Sex	Male	Female	Male	Female	Male	Male	Male	Male
General condition	Severe mental retardation	Alive at 4 months	Severe mental retardation	Poor development	Psychomotor development retardation	Psychomotor development retardation	Repeated convulsion	
Cranium	Micro- and dolichocephaly	Dolichocephaly	—	—	—	Dolichocephaly	Dolichocephaly	
Eyes	Deeply set	Deeply set	Wide set	Enophthalmos	Strabismus	Weak-sight	Weak-sight	
Palpebral fissures	Horizontal, Bilateral epicanthal folds	Horizontal	—	Narrow	Hypertelorism	Upward slanted	Blepharophimosis	
Ears	Normal	Low-set, large, round	Simple	Low-set, large, soft	epicanthus	Low-set, large lobes	Low-set	
Nose	Beaked	Beaked	Beaked	Beaked, Prominent, Enlarged bridge	—	—	Enlarged bridge	
Chin	Receding, round	Receding, round	Retromicrognathia	Receding	Receding	Normal	Normal	
Mouth	Small	Small	—	Long upper lip	Down-slanting corners	Normal	Normal	
Hand	Long fingers clutched over the thumbs, the index over the other fingers	Long fingers clutched over the thumbs, the index over the other fingers	Tapered finger	Arachnodactyly	Low-set thumbs, Single palmar creases	Normal	Normal	
Feet	Long toes with abnormal implantation	Long toes with abnormal implantation	Flexion contractures	Both thumbs were held transversely in the palms, over the 3rd and 4th fingers	—	Normal	Normal	
Genitalia	Normal	Normal	Hypoplastic	Normal	Engulfed penis	Cryptorchism	Small penis	

a very small segment of a long arm of a no. 9 which was represented by the band q12→q13 (Fig. 4). The mother of the proband is most probably a carrier of the direct insertion of a long arm of no. 9 (q12q13) into a long arm of no. 1 (1q11). She had in the past three spontaneous abortions. Very likely it might be from the unbalanced state of her gametes caused by deleted 9q12→q13.

Fig. 4. Diagram showing direct insertion of a long arm segment of no. 9 (q12q13) into a long arm of no. 1.

The conclusion is then made that the propositus possesses the karyotype, 46,XY,der(1), ins(1;9) (q11;q12q13) mat, and his mother is a carrier of direct insertion, 46,XX,ins(1;9) (q11;q12q13).

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