16. Elongated Long Arm of No. 1 Chromosome in Heavy Mental Retardates

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Due to the application of differential staining methods, various specific chromosome changes have been demonstrated in healthy normal subjects as well as in those with phenotypical abnormalities. The elongated long arm of no. 1 chromosome (1qh+) has been found in both normal persons and subjects with congenital malformations.

In a study on 70 cases with heavy mental retardation, it was found that 5 cases had 1qh+.

Materials and methods. All the 1qh+ cases were among 70 cases with heavy mental retardation, who were admitted in Kamo National Sanatorium.

The chromosome slides were prepared following the standard blood culture procedures. The karyotype analyses were made with the application of the conventional Giemsa staining and G-banding differential staining.

Results and remarks. Out of 70 cases, 5 cases had the elongated long arm of no. 1 chromosome, being 7.1% in incidence. Table I showed the results of the clinical examinations and karyotypes of the 5 cases having 1qh+. Fig. 1 showed the partial karyotypes of those cases.

Fig. 1. The partial G-banding karyotypes of the patients and their fathers, having 1qh+ chromosome. 1: Case 1. A; the patient, 46,XY,1qh+,rcp(3;4)(q23;p14). B; his father, 46,XY,1qh+. 2: Case 2. 46,XX,1qh+. 3: Case 3. A; the patient, 46,XY,1qh+. B; his father, 46,XY,1qh+. 4: Case 4. 46,XX,1qh+. 5: Case 5. 46,XY,1qh+.

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Table I. Clinical and family data on the 1qh+ cases

Case no.	Age	Sex	Karyotype	Clinical features	1qh+ relatives discovered		
					No. of relatives	Karyotype	Phenotype
1	13y.	M	46,XY,1qh+ rep(3;4) (q23;p14)	growth retardation, hypertelorism, epicanthal fold, downward slanted palpebral fissure, ptosis of eyelid, blepharophimosis, strabismus, low-set and malformed auricle, dysplasia and mal-implantation in teeth, kyphosis, bronchial asthma, nephrosis	1 (father)	46,XY,1qh+	normal
2	15y.	F	46,XX,1qh+	low-nasal bridge, cleft lip, seizure, behavior disorders, slight cerebral palsy	0		
3	31y.	M	46,XY,1qh+	congenital cataracts, blindness, frontal bossing, hypertelorism, prominent nasal bridge, dysplasia and malimplantation in teeth, (father) aged facies, seizure, behavior disorders	1	46,XY,1qh+	normal
4	18y.	F	46,XX,1qh+	oxycephalus, seizure, slight cerebral palsy	0		
5	31y.	M	46,XY,1qh+	short stature, flat occiput, head deformity, dysplasia and mal-implantation in teeth, behavior disorders	0		

All the cases had heavy mental retardation and speech disorders.

Table II. Clinical and family data on the 1q+ cases in literatures

Authors	Proband ascertainment	Pheno- typically normal 1q+ relatives discovered		Pheno- typically abnormal 1q+ relatives discovered	
Patau <i>et al.</i> (1961)	Oral-facial-digital syndrome	0		Mother has OFD*	
Gorlin & Psaume (1962)	Oral-facial-digital syndrome	—		—	
Yunis & Gorlin (1963)	Jaw cyst, basal cell cancer, bifid rib syndrome	4		0	
Cooper & Hernis (1963)	Male pseudohermaphroditism	2		0	
Koulischer & Zanen (1964)	Miner palpebral anomalies	0		Mother same anomalies	
Philip <i>et al.</i> (1965)	Primary amenorrhoea, adrenocortical hyperfunction	2		0	
Genest <i>et al.</i> (1966)	Apert's syndrome	—		—	
Büchner <i>et al.</i> (1967)	B/D translocation**	3		0	
Sofuni & Sandberg (1967)	Primary amenorrhoeas, short stature	3		0	
Donahue <i>et al.</i> (1968)	Phenotypically normal	9		0	
Fergament <i>et al.</i> (1968)	Mother of repeated abortions and/or stillbirths	—		—	
Ibid	Mother of repeated abortions and/or stillbirths	—		—	
Ying & Ives (1968)	Epicanthic fold, unilateral simian crease	5		—	
Wilson (1969)	Mother of three spontaneous abortions	—		—	
Jacobs <i>et al.</i> (1970)	Borstal survey	9		0	
Saner (1970)	Psychomotor retardation, multiple minor abnormalities	0		—	
Ibid	Trisomy 18	1		0	
Ibid	Epilepsy	—		Sib with minimal cerebral palsy and hypertelorism, father hypertelorism	
Kamaryt <i>et al.</i> (1971)	Trisomy 21**	7		0	
Prigozhina <i>et al.</i> (1971) & Prigozhina (1973)	Chronic lymphocytic leukemia	8		0	
Lobitz <i>et al.</i> (1972)	Mild mental retardation	3		0	

Table II (continued)

Authors	Proband ascertainment	Pheno- typically normal 1q+ relatives discovered	Pheno- typically abnormal 1q+ relatives discovered
Holzer <i>et al.</i> (1972)	Short stature, Webbed neck, ptosis, undescended testes	9	2 short stature
Howard <i>et al.</i> (1972)	Mildly dysmorphic and retarded	4	generation transmission
Østergaard (1973)	Immunological defect	0	—
Osztovcics <i>et al.</i> (1973)	Trisomy 18	—	—
Kulazhenko <i>et al.</i> (1973)	Fetus with phenotypical Patau's syndrome***	1	0
Gardner <i>et al.</i> (1974)	Translocation trisomy D	2	0
Ibid	Scoliosis, webbed neck	1	0
Ibid	Short stature	—	—
Ibid	Clinically Patau's syndrome	3	0
Ibid	Clinically Patau/Meckel syndrome	1	0
Ibid	Severe mental retardation, abnormal facial appearance, R. microphthalmia, cleft lip and palate, scoliosis	—	—
Nielsen <i>et al.</i> (1974)	Organic psychosyndrome	2	0
Ibid	Personality deviation	—	—
Ibid	normally developed newborn	0	0
Ibid	normally developed newborn	3	0
Ibid	normally developed newborn	2	0
Halbrecht & Shabtey (1976)	Polycystic kidney, fibrosis of liver and pancreas, polydactyly, malformed ears	3 (parents and sister)	0
Ibid	Parents of malformed boy and microcephalic girl	—	—
Ibid	Hydrocephalus, antimongoloid slant, micrognathia, retrognathia, bone malformations, poly cystic kidney, 1qh+, 15p+, Yq+	1 Mother 1qh+, 15p+	0
Ibid	Prune Belly syndrome	2 (parents)	—
Ibid	Stillborn anencephalic boy	2 (parents)	—
Ibid	Patau's syndrome	2	—
Ibid	Stillborn malformed girl (hydrocephalus, cleft lip and palate, congenital cataracts)	1	—
Ibid	Mongol child	1	—
Halbrecht & Shabtey (1976)	18 trisomic mosaic child with multiple stigmata	1	—
Ibid	Polycytemia	—	—
Ibid	Polycytemia	—	—
Ibid	Chronic myelocytic leukemia	—	—
Ibid	Systemic Lupus Erythematosus	—	—
Ibid	Mother of a macerated fetus	—	—
Tsenghi <i>et al.</i> (1976)	Husband of a couple with repeated abortions	—	—
Salamanga-Gómez <i>et al.</i> (1978)	Chediak-Higashi syndrome	2	—
Kadotani <i>et al.</i> (1980)	Husband of a couple with repeated abortions	—	—

* OFD: Oral-facial-digital syndrome.

** Did not carry 1q+.

*** Chromosome not analyzed.

In reference to 1qh+, the phenotypes of probands were various (Table II). And it is very interesting that, although the random variety of abnormalities in the 1qh+ index cases, the almost relatives, who carried the 1qh+, were phenotypically normal.

Further, more 1qh+ cases were found in the abnormal population than in the normal population (Gardner *et al.* 1974). Tüür *et al.* (1974) made the chromosomal study on 208 normal adults and reported that the incidence of 1qh+ was 1.9%. In the present study, 1qh+ indicated higher incidence (7.1%).

The elongation of long arm of no. 1 chromosome was found in the area of the constitutive heterochromatin. Recently several studies concerning the quantity of constitutive heterochromatin have been reported (Baliček *et al.* 1977; Lopetequi *et al.* 1980; Maes *et al.* 1983). But the significance of the variation in the quantity of the constitutive heterochromatin remains to be elucidated. We hope further investigation.

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