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Ring Chromosome 14 Without Deletion

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A new case of a male infant with ring chromosome 14 is reported. The G banding characteristics of early metaphase chromosomes did not reveal visible deletions in the long and short arms. The present report discusses the basic symptoms characteristic of the ring 14 chromosome and also points to a certain clinical polymorphism.

About 8 cases of ring chromosome 14 have been reported in the literature, including only one male. R(14) is a well-defined clinical entity involving mental and physical retardation, seizures, craniofacial dysmorphism, hypertelorism, epicanthus, broad flat nasal bridge, large low-set ears, widely spaced nipples.

We present the clinical and cytogenetic findings of a male infant with ring 14 chromosome.

Report of a Case

The proband P.H.B. was a male, the second child in the family. The pregnancy of the mother was pathological with neuropathy and blood pressure values of 140—150/100. No treatment was applied. The birth was diffcult and protracted but at term. At birth the child weighed 4230 g, measured 54 cm and cried at once. The right collarbone was broken. The baby was first fed six hours after birth. The peculiar shape of the head and upright ears attracted attention.

The mother's age at birth was 27 years. Her first pregnancy at 21 years terminated by normal delivery of an infant female who is now six years old, with normal psychomotor development. The father was 27 years old at the patient's birth. There are no data of preceding diseases, nor miscarriages, stillborn babies or malformations.

When the child was 4.5 months of age, generalized tonic-clonic seizures had begun, with different duration and repetition rate. Some resembled epileptic statuses, which were difficult to control with anticonvulsive treatment.

At present the child is 14 months of age and does not show physical retardation: height 74 cm, weight 11 kg, head circumference 43.7 cm.

At the first glance the face is normal but the cranio-facial dysmorphism

TABLE I

Phenotypes of r(14) probands

Phenotypic features	Gilgen-krantz et al (1971)	Jalbert et al (1977)		Torri-celli et al (1978)	Abe et al (1978)	Schmidt et al (1979)	Amarose et al (1980)	Triolo et al (1981)	Present case (1984)
Sex	Female	Female	Female	Female	Female	Female	Female	Male	Male
Mother's age at birth, years	26	28	28	26	24	16	31	26	27
Gestational time at birth, weeks	39	29	29	42			39	39	
Weight at birth, g	2500	1920	1480	3100	3650	2505	2305	2300	4230
Dolichocephaly	—	+	+	+	—	+	+	+	+
Microcephaly	+	+	+	+	+		+	+	+
Broad flat nasal bridge					+	+	+	+	+
Epicanthal folds	+	+	+	+	+	+	+	+	+
Large low-set ears	+			+		+	+	+	+
Ogival palate	+			+		+	+	+	—
Micrognathia	+		+	+		+	+	+	+
Drooping mouth	+			+	—			+	—
Seizures	+	+	+	+	+	+	+	+	+
General hypotonia	+	+	+		+		+	+	—
Growth retardation	+	+	+	+	—		+	+	—
Psychomotor retardation	+	+	+	+	+	+	+	+	+

is evident: the head has a microcephalic configuration but is elongated, the fontanel is closed. The forehead is low and narrow, the frontal cranial bone is hypoplastic. The soft facial parts are well-formed, the brows are roughly outlined, there is a pronounced epicanthus and slight hypertelorism. The nasal base is large, the nostrils are broadly opened. The external ears are low-set, large, well-formed, upright. With the narrow forehead the head assumes a rabbit-like appearance. The mouth is small, the palate is high but not ogival (Figs 1 and 2).

The hair is light and thin but normal. The neck, body and extremities are well-proportioned. Clinical investigations did not reveal any internal anomalies.

Paraclinical studies

Routine blood and urine tests were normal. The wrist X-rays showed normal bone age. Cranial X-rays revealed elongations and lower limit dimensions. The eyeballs and ECG were normal, the EEG showed an abnormal record without, however, evident hemispheric asymmetry. Delta waves

were observed at the central regions of both hemispheres. Psychological study revealed moderate mental retardation, psychomotor and speech retardation in the socio-emotional sphere and sensor activity.

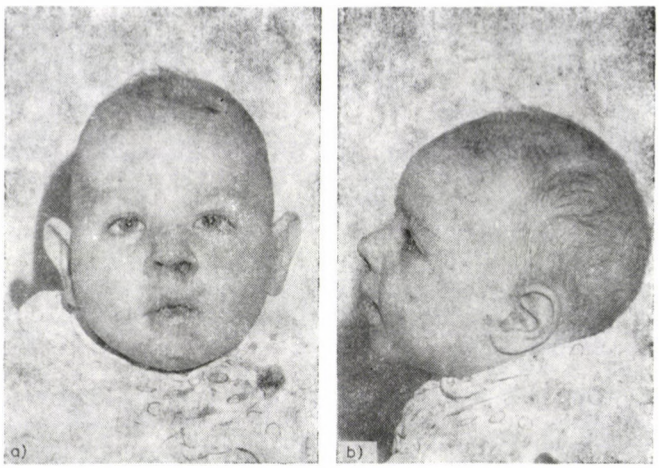
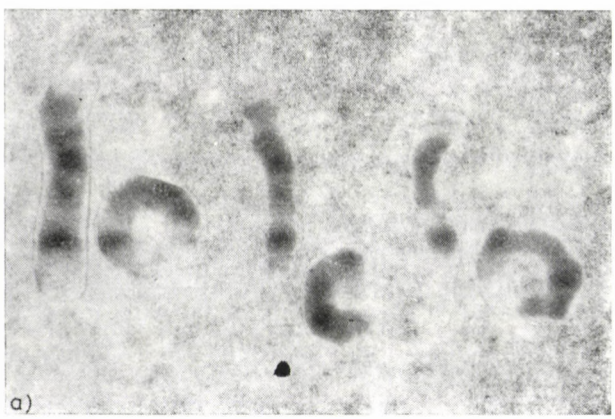


FIG. 1/a—1/b: The patient at 14 months of age



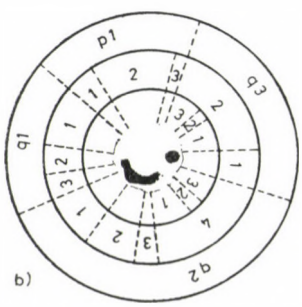


FIG. 2/a—2/b: Ring 14 chromosome identified by G banding

Cytogenetics

100 prometaphase and early metaphase chromosomes from four different lymphocyte cultures were analysed. The chromosomes were stained by G banding.

The G band characteristic of D 14 at the early stage of chromosome contraction (late prophase and early metaphase) was represented by a normally structured long arm of the ring chromosome (Figs, 2/b). The distal segment q 32 had clearly visible subbands 32.1, 32.2, 32.3 and 32.4; in the short arm p11 and p12 bands were differentiated. The aberrant chromosome was associated with acrocentric chromosomes. The infant's karyotype was 46, XY, r (14) (p 13 + + q 32); the parents had normal karyotypes.

Discussion

The proband's ring chromosome had originated as a result of mutation in the meiosis of one of the parents, or at an early stage before the zygote's first division, in view of the normal karyotype of the parents.

The diversity of clinical symptoms may be attributed to the mytotic instability of the aberrant chromosome which led to mosaic forms and secondary changes in the ring structure. The ring chromosome of our patient was stable, like in the case of Amarose (1), while Triolo et al (4) reported on a patient with ring 14 instability in 17%.

References

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