Ring 20 chromosome phenotype

B PORFIRIO, M G VALORANI, A GIANNOTTI*, G SABETTA*, AND B DALLAPICCOLA

Cattedra di Genetica, Dipartimento di Sanità Pubblica, II Università di Roma; and *Ospedale Bambino Gesù, Istituto di Ricerca Scientifica, Rome, Italy.

SUMMARY A baby with a 46,XY,r(20) karyotype is described. The distinguishing features of this rare chromosome anomaly are analysed in the light of the suggested r(20) syndrome.

The existence of a ring 20 chromosome syndrome, characterised by neurological impairment, low IQ, and behavioural and psychological problems from infancy or childhood, was suggested in 1976 by Borgaonkar et al., on the basis of similarities in the phenotype of three reported patients. This has been corroborated by a small number of subsequent reports which have confirmed that these subjects have several features in common. In 1979, Stewart et al. re-examined six cases and concluded that all had seizures and three had behavioural problems of various types. Intelligence quotients ranged from 18 to 80. None had visceral malformations and physical anomalies were negligible.

We report an additional observation of r(20) found in a patient without seizures and with a complex congenital heart defect.

Case report

This boy, the only child of healthy, unrelated parents, was born in 1983, when the father was 28 and the mother 22 years old. Delivery was spontaneous, normal, and at term following an uncomplicated pregnancy. Birth weight was 3600 g and head circumference 33.5 cm. A congenital heart defect was recognised at 18 days of age. Echocardiography showed an ostium secundum type atrial defect. He smiled at seven weeks and sat unsupported at seven months.

When examined by us at 15 months, he was unable to stand unsupported and had no speech. His weight was 8.2 kg and his height 73 cm, both well below the 3rd centile. The facial appearance was not strikingly unusual (fig 1). There was no abnormality in the shape or size of the skull, but the fontanelles were still widely open. The forehead was high arched, with a flat glabella, rounded nasal tip, long philtrum, low set and prominent ears, and mild

Fig 1 Facial appearance of the patient.

Fig 2 Partial karyotype of F group chromosomes from two selected cells.
Discussion

Eight other patients with a ring 20 chromosome have been reported by banding studies. However, this case has been included in our discussion because of the similarity to our own patient. In five cases, the abnormality arose from a postzygotic event, as shown by the presence of a cell line with a normal karyotype. In three other cases, no normal cell line was present. However, in most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In four of these patients, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.

In most cases, the male to female ratio was 2:1, a ratio that is normal in the population. The mean age of fathers and mothers, respectively, was 27 and 28 years. In the 27 year old father, the mean age of the patient was 7 years. In the 28 year old mother, the mean age of the patient was 9 years. The mean age of the patient was 11 years.
and cryptorchidism in males. Mental retardation was a consistent feature, though it was severe in only half of these patients. A characteristic finding was arrest of development at a certain age, with the appearance of behavioural abnormalities of various types (immature, impulsive, aggressive) in about 50% of cases. This clinical evolution was associated with occurrence of epileptic seizures, which were poorly controlled by treatment. The age of onset of seizures ranged from three months to 10 years.4

Our patient had neither seizures nor behavioural abnormalities which are considered distinguishing features of the r(20) syndrome. It could well be that full expression of the syndrome has not yet become manifest in this young child because of the relatively late onset of encephalopathy and seizures. On the other hand, our patient presented with a complex congenital heart disease. This suggests that visceral malformations could represent the outer range of phenotypic expression in the narrow clinical spectrum of this rare chromosome imbalance.

References
1 Borgaonkar DS, Lacassic YE, Stoll C. Usefulness of chromo-

Correspondence and requests for reprints to Professor B Dallapiccola, Department of Public Health and Cell Biology, 2nd University of Rome, Via O Raimondo, I-00173 Rome, Italy.