

Table Serum N-acetylgalactosaminyltransferase levels in proband and family

Serum donor	Genotype	Counts per minute*
I.1 Father	OO	90
I.2 Mother	A ¹ O	2296
II.1 Husband	A ² O	464
II.2 Proband	A ¹ O	1089
II.3 Brother	OO	28
III.1 Son	OO	50
III.2 Daughter	A ¹ A ² (or A ¹ O)	1801
Controls C.T.	A ¹ O	3259
R.M.	A ²	403

*Radioactivity in product of an incubation mixture containing (in a final volume of 45 μ l) 0.063 μ mol LNF-1; 0.005 μ mol UDP-N-acetyl-D-galactosamine-1-¹⁴C (2×10^5 cpm per μ mol); 1.25 μ mol MES, pH 5.6; 1.25 μ mol MnSO₄; and 10 μ l serum. The mixture was incubated at 37°C for 3 hours.

indicates that the A¹ gene is not restricted to her haemopoietic tissue.

If Mrs Co. were a twin, the presence of O red cells could be attributed to a graft *in utero* of haemopoietic tissue from a group O twin. However, Mrs Co. is not known to be a twin, and there is no history of twinning in her family. The mechanism of her blood group chimaerism remains unestablished.

We thank Mrs Co. and her family for their co-operation; Dr C. Alper (Boston) for determining the serum protein allotypes; Mr D. Della-Loggia (Boston) for technical assistance, Dr H. A. Gardner (Toronto) for provision of photographic equipment and advice; Dr P. S. Gerald (Boston) for analyses of Mrs Co.'s chromosomes; Dr E. R. Giblett (Seattle) for determining the red cell enzyme phenotypes; and Dr H. Schachter (Toronto) for provision of facilities to assay the A-specified transferases in serum.

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The r(20) syndrome

SUMMARY A 21-year-old woman with a ring-20 chromosome is described. The clinical findings, behaviour problems, epilepsy, and low grade mental retardation are the same as in the 3 cases described earlier. It seems to be justified to speak of a specific ring-20 syndrome.

Abnormalities, involving F group chromosomes, are rare. Structural changes have been found in bone marrow cells associated with haematological disease and examples of aneuploidy have been reported in spontaneous abortions. It seems that a ring-20 chromosome has been described only once without mosaicism and twice with mosaicism. We present a case with r(20) with similar clinical findings to those in the 3 previous cases.

Case report

The patient was a girl born in 1955 when the father was 35 and the mother 37 years old. She had one

brother and one sister, both of them older than she. The pregnancy and the delivery were normal. Birthweight was 3200 g. The early development was normal. She walked at 11 months.

At the age of 5 years, she began to have epileptic seizures. She went to school at 6 years of age but after 5 years her schooling was interrupted because of poor success and poor control of epilepsy. She was examined in a child psychiatric ward because of maladaptation and epilepsy and the following year she was sent to an epilepsy sanatorium where she stayed for 5 years. She then lived at home, and in 1976, at the age of 21 years, she began sheltered work in an institution for the mentally retarded.

On examination her height was 159 cm and her weight 59 kg. Her appearance was unremarkable.

In haematological investigations peripheral blood and bone marrow were normal.

Electroencephalogram showed extensive bilateral abnormality with pronounced seizure activity, predominantly occipitally and temporally on the right.

Psychological testing in 1976 showed mild mental retardation; mental development was at about the level of a 7-year-old. She reads and writes. She is co-operative and able to perform simple tasks.

Cytogenetic studies

The chromosomes of cells from peripheral lymphocyte cultures and bone marrow were examined. Lymphocytes were cultivated for 3 days according to the micromethod. Bone marrow was incubated for 1½ hours at 37°C before harvesting. The slides were made using the air-drying technique. The chromosomes from both materials were stained by Giemsa and by the G-, Q- and C-banding techniques. The chromosomes of 100 metaphases from both the blood and the bone marrow were counted.

In 90% of the metaphases of both the peripheral

blood and the bone marrow there was a ring chromosome replacing one of the homologues of pair 20. The chromosome pattern 45,XX,-20 was in 1% and 2% of the metaphases of the blood and the bone marrow, respectively. The chromosome pattern 46,XX was found in 9% of the metaphases of the blood and in 8% of the bone marrow. The size and shape of the ring varied slightly from cell to cell, more in the bone marrow than in the blood (Fig. 1 and 2). G- and Q-bandings of the ring showed two small dark and bright regions, respectively, pointing to the shorter arms of chromosome 20. C-banding showed one darkly stained centromeric region (Fig. 2).

No remnants of a possibly disappearing ring chromosome could be seen. Only a few micronuclei were found, but other abnormalities of the mitoses remained outside of this study because colchicine was added in every culture.

Discussion

Constitutional ring chromosomes have been described for all chromosome groups in man. Kisten-

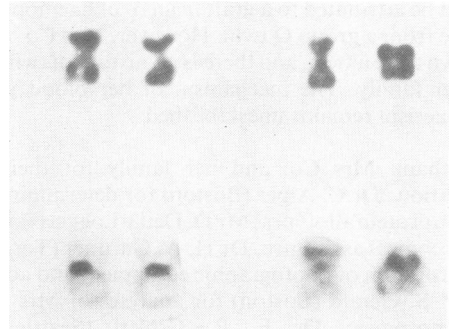


Fig. 1 *F*-group chromosomes of blood with routine stain and G-banding.

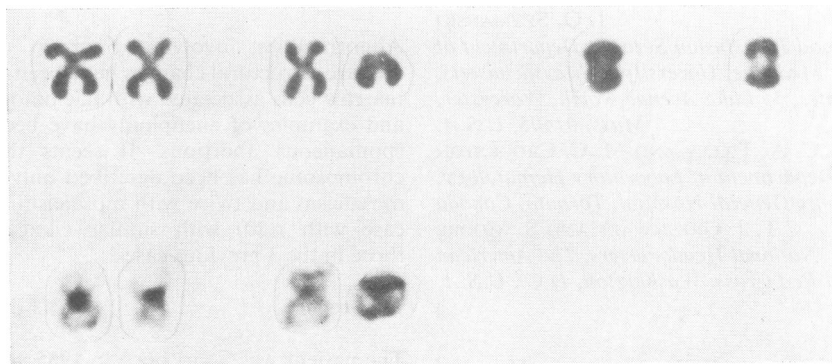


Fig. 2 *F*-group chromosomes of bone marrow with routine stain and G-banding.

macher and Punnett have presented a review of 56 cases and have studied comparative mitotic behaviour of r(9) and r(13).

Ring formation of chromosome 20 with mosaicism has been previously described twice. In a 26-year-old woman with nervous symptoms and epileptic seizures the signs and the chromosomal abnormality were presumed to be coincidental (Uchida and Lin, 1972).

Another case was a 12-year-old boy with epilepsy and behaviour disorder (Faed *et al.*, 1972). The third case with r(20) and the only one without mosaicism was a 7-year-old boy with similar symptoms (Atkins *et al.*, 1972).

Our patient is similar to all three in having low grade mental deficiency, behaviour problems, and epilepsy.

In all earlier patients and also in our patient r(20) seems to be relatively stable. This may be the cytogenetic explanation of the definite clinical picture. Leisti *et al.* (1968) suggest that the lability of the ring in r(E) patients is related to various phenotypic expressions.

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Congenital discoid lupus in the newborn

SUMMARY A female infant, born to a 21-year-old mother with systemic lupus erythematosus,

had cutaneous discoid lupus at birth. The lesions resolved spontaneously over the first few months and by the age of 1 year the infant's skin was normal. Other possible complications of this maternal disease are discussed and the need for caution in counselling mothers is recommended.

Case history

The mother was in good health until 1970 when, aged 18 years, she developed an erythematous rash on her face as a result of exposure to bright sunlight. She subsequently became unwell with general malaise, headache, nausea, and vomiting. On admission to the City Hospital, Nottingham, she was found to be anaemic with a haemoglobin level of 8.2 g/dl. She had a neutropenia of 2300 white cells/mm³, and proteinuria with total serum protein of 47 g/l, and only 12 g/l albumin. She developed the nephrotic syndrome with peripheral oedema and ascites, subsequently going into renal failure and then left ventricular failure. Renal biopsy confirmed focal glomerular changes with thickening of the basement membranes and for the first time she was found to have LE cells in the peripheral blood with a positive titre of antinuclear antibody (ANA) and a reduced serum complement level at 54 mg/100 ml (Table). Treatment was started with prednisolone, her condition improved, normal renal function returned and, despite an episode of right sided pleurisy and consolidation, she was well enough for discharge 3 months after the onset of symptoms.

As an outpatient, treatment with prednisolone was continued and she was also given azathioprine; the steroid dosage varied from 15 mg t.d.s. to 5 to 10 mg alternate days and she was maintained on azathioprine 25 mg b.d. until 1973. In February of that year, approximately 2½ years after the onset of her disease, she had amenorrhoea but a test for pregnancy was

Table Serological findings in mother and infant

Normal adult levels	ANA titre		C3 Levels
	<10	>20	
Mother			
23.9.70	400	—	54
10.11.71	—ve	16.5	185
1.12.72	20	NT	125
23.3.73	20	84.4	38
14.10.73	—ve	12.0	68
13.9.74	100	43.2	46
Infant			
14.10.73	—ve	2.2	64
23.10.73	—ve	1.9	75

NT, not tested.