attach itself to the spindle. Thus it would allow the dicentric to behave as a monocentric element dominated by the centromere of the No. 8. There was no evidence of mosaicism or breakage, but these events could not be ruled out as they might be lethal or sublethal and consequently not seen in the cultured cells. Such instability of the dicentric at critical stages of embryonic development might have contributed to the abnormalities of the proposita. However, it is likely that the formation of the dicentric was preceded by the deletion of the tips of both chromosomes, and it is probable that any loss of material from the No. 8 would have contributed to the abnormal phenotype.

It is of interest that, in the stable dicentric autosomes so far reported in man (present case; Niebuhr, 1972a, b; Warburton *et al*, 1973; Šubrt *et al*, 1971), acrocentric short arm material was translocated into an intercentric position, where, in at least two cases (present case; Warburton *et al*, 1973), it retained its function of nucleolar organizer, as indicated by its association with acrocentric chromosomes. It is possible that satellites are particularly susceptible to participation in chromosomal rearrangements. This, and the compatibility of loss of acrocentric short arm material with normal development (Nielsen *et al*, 1974) might, in part, explain the involvement of acrocentrics in the dicentric chromosomes referred to above. However, in the present case and those reported by Niebuhr (1972a, b) and Warburton *et al* (1973), the centromere of the acrocentric chromosome (or one acrocentric in dicentric Robertsonian translocation chromosomes) usually appeared to be partially or completely non-functional. It is, therefore, suggested that there may be a tendency for the centromeres of acrocentrics in dicentric chromosomes to become inactivated, making it possible for stability to be achieved. This may be an important factor in the involvement of acrocentrics in the stable dicentric autosomes of man.

S. H. ROBERTS, R. T. HOWELL, K. M. LAURENCE,
and M. E. HEATHCOTE

*Child Health Laboratories, Department of Child
Health, Welsh National School of Medicine,
Heath Park, Cardiff CF4 4XN*

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Mental retardation with 45 chromosomes 45,XX,-5,-14,+der(5)t(5,14)(p15;q13) mat due to familial balanced reciprocal translocation

SUMMARY A girl with severe mental retardation and odd facies and some features of the cri-du-chat syndrome was found to have only 45 chromosomes. Her karyotype was 45,XX,-5,-14,+der(5)t(5,14)(p15;q13) mat. Her mother and her two sisters were found to be balanced reciprocal translocation carriers having 46 chromosomes, one of which was a very small (14pter→14q13::5p15→5pter) that was missing in the proposita.

The very few cases of 3:1 meiotic disjunction resulting in 45 chromosome offspring have been recently reviewed in this journal by Lindenbaum and Bobrow (1975). To the best of our knowledge the present report is the first of a family with a balanced reciprocal translocation involving chromosomes 5 and 14 detected because the proposita was a mentally retarded unbalanced 45 chromosome offspring. A

somewhat similar family with a balanced reciprocal translocation involving chromosomes 4 and 15 has been recently reported by Cohen *et al* (1975).

Case report

The *proposita* (III.2, Fig. 1) was born on 3 November 1968. The father and mother were 30 and 22 years of age, respectively, at the time of her birth. She was born after a full-term pregnancy. Delivery was by vacuum extraction because of deep transverse arrest of the head. Birthweight was 3800 g. At the age of 1 week she was found to be slightly apathic with cephalohematoma, slightly increased anterior fontanelle, weak reflex responses, and was thought to have brain damage. She had a weak cry. Her development was slow and she suffered from constipation. When she was admitted to hospital at the age of 9 months because of fever of unknown origin she was still unable to sit and was hypotonic. She was thought to have Hirschsprung's disease, and hypertelorism and epicanthic folds were noted.

At the age of 6½ years the child was admitted to hospital with acute gastrointestinal disease. She was thin (weight 14 300 g, height 107 cm), and severely retarded (Fig. 2) she behaving like 1½-year-old infant (IQ estimate of about 23). The child had an odd facies (Fig. 2B, C) with telecanthus (inner canthal distance 3.7 cm), that is above the 97th centile for adults, mongoloid slant of the eyes, epicanthic folds, and a small chin. The above combination of features was the reason for chromosomal investigation.

X-rays of the abdomen revealed megacolon with the sigmoid on the right side. The long bones were delicate and osteoporotic. The skull and the dorsal and lumbar vertebrae were normal. Bilateral cervical rib was present and the anterior part of the first and second ribs were widened with a bridge formation on the right side. Intravenous pyelography and cystography re-

vealed right hydronephrosis and left vesicoureteral reflex. Electroencephalogram showed mild diffuse disturbance. Dermatoglyphs showed whorls on all the fingers of the left hand and on the fourth finger of the right hand and ulnar loops on the remaining fingers. The axial triradii were distally placed (*atd* angle, right 70°, left 75°) and hallucal patterns revealed bilateral distal loops.

Cytogenetic studies and family report

Chromosome investigations were performed on phytohaemagglutinin stimulated peripheral lymphocytes obtained from the family members. Chromosomal banding was obtained by the ASG method (Sumner *et al*, 1971) and/or the trypsin methods (Seabright, 1971).

The karyotype of the *proposita* had only 45 chromosomes and was found to be 45,XX,-5,-14,+der(5)t(5;14)(p15,q13) mat (Fig. 3) in all the 150 cells counted. The buccal smear was as expected X-chromatin positive.

The mother of the *proposita* had 46 chromosomes in all the 150 cells counted, with 2 abnormal elements; the identical large marker, morphologically resembling a No. 1, as seen in the *proposita*, as well as a small acrocentric marker similar to the G group but about two-thirds in size (Fig. 4 and 5). Banding techniques showed that the mother was a balanced reciprocal translocation carrier with the following karyotype 46,XX,t(5;14)(5qter→5p15::14q13→14qter;14pter→14q13::5p15→5pter). The karyotypes of the two normal sisters of the *proposita* (III.4 and 5, Fig. 1) were identical with that of the mother. The first pregnancy of the mother (III.1) ended in 1967 in a missed abortion, after 6 months of pregnancy, of a macerated female fetus having a weight of 190 g and length of 22 cm.

The maternal grandparents (I.1 and 2) were first cousins of the Moroccan Jewish community. The grandfather (I.1) and grandmother (I.2) were 22 and 17

FIG. 1 Family pedigree. *Proposita* is 45,XX,-5,-14,+der(5)t(5;14)(p15;q13) mat.

FIG. 2 A. The proposita 6½ years old. B and C. The face of the proposita, 6½ years old.

years of age, respectively, at the time of the birth of the mother (II.2). The maternal grandmother and 4 maternal uncles and aunts were found to have normal karyotypes but the grandfather refused testing.

Blood group markers were not informative in this family, but did not indicate any nonpaternity.

Discussion

The problem of reciprocal translocation and 3:1 meiotic disjunction resulting in 47- or 45-chromosome offspring has been recently reviewed by Lindenbaum and Bobrow (1975). There were

only 3 cases with 45 chromosomes derived from a parent heterozygote for a reciprocal translocation. The present case seems to be the first tertiary monosomy segregant 3:1 of the type 45,XX,-5,-14,+der(5)t(5,14)(p;q) though one case of a *de novo* tertiary monosomy of the same type if not identical has been described by Reinwein and Wolf (1965) and Wolf *et al* (1966). In the *de novo* case reported by Capotorti and Ferrante (1966) only the groups of the chromosomes involved in the translocation were identified 45,XY,-B,-D,der(B)t(B;D)(p;q). Those 2 cases as well as our own showed

FIG. 3 Partial karyotype (trypsin) of the proposita 45,XX,-5,-14,+der(5)t(5,14)(p15;q13) mat.

FIG. 5 Schematic partial maternal karyotype. Arrows indicate break points in the two translocation chromosomes (chromosomes 14 drawn upside down for easier orientation).

FIG. 4 Karyotype (trypsin) of balanced carrier (mother of proposita) 46,XX,t(5;14)(5qter→5p15::14q13→14qter;14pter→14q13::5p15→5pter). Arrows indicate break points in the two translocation chromosomes.

some phenotypic manifestation of the cri-du-chat syndrome. The fact that the proposita showed only some of the typical features of the cri-du-chat syndrome can be explained by the variable expressivity of the syndrome. Moreover, because the proposita lost roughly the terminal fifth of the short arm of

chromosome 5 and the proximal fifth of the long arm of chromosome 14 as well as its centromere and short arm (Fig. 3 and 5), she is not expected to be a typical case of cri-du-chat.

The mother (II.2) was advised about the possibility of prenatal diagnosis in her future pregnancies

and the pregnancies of her daughters (III.4 and 5) in the more distant future.

K. FRIED, M. TIEDER, S. BEER, M. ROSENBLATT,
and H. I. KRESPIN

*Departments of Genetics and Paediatrics 'B',
Asaf Harofe Hospital, Tel-Aviv University
Medical School, Zerifin, Israel*

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Sex linked hydrocephalus

SUMMARY A family showing the syndrome of X-linked hydrocephalus is presented and the relevance of this condition in genetic counselling discussed. A method of decompressing a grossly enlarged after-coming head is described.

Case history

A 29-year-old patient presented for hospital booking at 16 weeks' gestation by dates. Her only previous confinement, conducted elsewhere, had resulted in an intrapartum anoxic stillbirth after clinical evidence of fetal distress in early labour. A necropsy of this female infant (birthweight 3360 g) had shown no evidence of congenital abnormality.

Examination confirmed the presence of a 16-week pregnancy. When seen at 32 weeks' gestation clinical examination revealed a breech presentation. This presentation was maintained and delivery by elective caesarean section was planned. An abdominal x-ray film, requested to exclude the possibility of gross fetal

abnormality, showed an overlarge fetal head with thinning of the skull bones and widening of the cranial sutures. Sonar cephalometry at 36 weeks' gestation calculated a biparietal diameter of 11.4 cm confirming the diagnosis of hydrocephalus.

The patient was admitted to hospital at 38 weeks' gestation when a further sonar measurement showed an increase of 1.1 cm in the biparietal diameter. The findings were discussed with the patient and her husband and the reason for preferring vaginal delivery were explained. Labour was induced by low amniotomy and an escalating syntocinon infusion. Epidural analgesia was provided and, after a first stage lasting 6 hours, the patient reached full dilatation and was, therefore, encouraged to bear down with contractions. The breech was delivered easily but difficulty was experienced with the shoulders and general anaesthesia was induced. There was no associated spina bifida. The aftercoming head remained high in the abdomen and a wide bore needle was inserted per abdomen releasing approximately 600 ml cerebrospinal fluid. The head then entered the pelvic brim and an incision was made in the skin over the highest accessible part of the fetal neck; with blunt ended scissors, a 'tunnel' was then created upwards between the scalp and the underlying skull. A Simpson's perforator was inserted and, while steadying the fetal head with the left hand on the lower abdomen, the skull was perforated. The blades of the instrument were opened and rotated within the cranium releasing a further gush of cerebrospinal fluid and brain tissue. The collapsed head was then readily delivered without the use of forceps. The infant, a male, weighed 2860 g. In view of the destruction of the skull contents necropsy was not requested.

The patient made an uncomplicated recovery and was discharged home two days later.

Family history

The patient's mother had had a stillborn hydrocephalic male infant in her first pregnancy in 1938, but no records remain of the details.

The patient stated that her elder sister had also had a stillborn hydrocephalic male infant in her first pregnancy. This was confirmed by reference to the hospital notes, which showed that hydrocephalus had been diagnosed by x-ray film one week before the onset of labour. During labour, with the vertex presenting, the head had to be perforated to allow delivery of the child. No necropsy had been requested because of damage to the brain and skull during the birth. Subsequently a normal daughter and son were born to this sister.

The family pedigree has been traced for three