associated with a male karyotype including an abnormal X chromosome was reported by Bernstein et al. The abnormal X was interpreted as a duplication of p21–pter. In addition to sex reversal, H-Y antigen was found to be virtually absent. This finding was interpreted as interference with a putative regulatory element on the X chromosome.

The recent tentative localisation of the H-Y antigen suppressor gene distal to Xp22 would perhaps explain why the sexual development in Bernstein’s patient with a distal duplication was severely affected, while our patient, with a more proximal duplication, had a male phenotype; however, unfortunately, H-Y antigen studies were not carried out in our patient.

Anomalous segregation of the Xg<sup>a</sup> allele in Bernstein’s patient might suggest that the Xg locus was involved in the inactivation process, but recombination at meiosis was also a possibility. Since both our proband and his brothers were Xg(a<sup>−</sup>) and the mother was Xg(a<sup>+</sup>), the findings in our patient do not seem to help in elucidating this problem. Furthermore, there was no evidence from BrdU incorporation for inactivation of short arm material on the X chromosome in the proband.

We thank Dr Ruth Sanger and the staff of the MRC Blood Group Unit, London, for performing the Xg blood group studies, and Mrs Birthe Jespersen for expert technical assistance.

KAREN BRØNDUM NIELSEN* and FINN LANGKJÆR†
*The John F Kennedy Institute, DK 2600 Glostrup; and †Andersvange, Institution for the Mentally Retarded, DK 4200 Slagelse, Denmark

References

Requests for reprints to Dr K Brøndum Nielsen, Department of Medical Genetics, The John F Kennedy Institute, 7 Gl Landevej, DK 2600 Glostrup, Denmark.

A complex chromosome rearrangement resulting in trisomy 15q22→qter

SUMMARY A black infant with malformations was found to have trisomy 15q22→qter. The mother had a complex chromosomal rearrangement involving three chromosomes (5, 13, and 15). A comparison with previously published cases of trisomy for distal 15q suggests a pattern of clinical findings including retardation in growth and development, microcephaly, asymmetrical facies, prominent occiput, antimongoloid slant of the palpebral fissures, micrognathia, prominent nose, and congenital heart disease.

Partial trisomy for the distal third to half of the long arm of chromosome 15 has been observed rarely. In the four previously reported families, the affected children were the result of maternal malsegregation of balanced translocations involving two chromosomes. We report an affected child resulting from the maternal malsegregation of a complex chromosome rearrangement involving three chromosomes, 5, 13, and 15.

Case report

The proband, a newborn black male, was the 1820 g product of an uncomplicated term pregnancy. He was born to a 28-year-old gravida 3, para 0, abortis 2 mother. There was no history of smoking nor alcohol or drug abuse. Labour was spontaneous and delivery was vaginal with mid-forceps assistance. The infant was meconium-stained and subsequently received

Received for publication 4 July 1981.
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oxygen therapy but never required mechanical ventilation. Apgar scores of 1 and 3 were assigned at 1 and 5 minutes, respectively. He remained in the nursery for 3 weeks and was discharged on nasogastric feedings. The baby was readmitted at 7 weeks of age for irritability, symptoms of upper respiratory infection, possible congestive heart failure, and failure to gain weight.

Physical examination showed a small, cyanotic, unusual appearing infant with multiple dysmorphic features (fig 1), which included length and weight markedly below the third centile; asymmetrical facies; sparse hair; large anterior fontanelle; severe microcephaly; prominent occiput; receding forehead; hypertelorism; microphthalmia; corneal clouding; low set, posteriorly rotated ears; right-sided preauricular pit; micrognathia; high arched palate, prominent upper lip; widely spaced nipples; short sternum; grade 4/6 holosystolic murmur; hepatosplenomegaly; flexion deformities of both elbows; micromelia; arachnodactyly; 3rd and 4th fingers of the same length; partial syndactyly of the 3rd and 4th fingers; bilateral simian creases; hypoplasia of all interphalangeal creases; flexion contractures of the proximal interphalangeal joints; fisting with adduction deformity of the thumb and radial sway of the fingers; syndactyly of the 2nd to 4th toes; the 2nd and 5th toes overlap the 3rd and 4th toes, respectively, on the right; the 2nd toe overlaps the 3rd toe on the left; metatarsus adductus and cryptorchidism with both testes palpable in the inguinal canal. Laboratory data were unremarkable. Chest x-ray showed cardiomegaly with increased pulmonary vasculature. Sector scan of the heart

FIG 1 The patient at the age of 6 months.

FIG 2 (a) Partial karyotype of the mother with the derived chromosome on the left; (b) diagramatic illustration of the complex rearrangement. Breakpoints are indicated by arrows.
was compatible with an A-V canal and possible subaortic stenosis. Radiographic studies showed mild gastro-oesophageal reflux and a small hiatal hernia. Intravenous pyelogram and voiding cystourethrogram were normal.

The infant was discharged on digoxin and was continued on nasogastric feedings. Further cardiac evaluation is planned when the baby has attained a weight of 42 kg.

He was seen at follow-up at 6 months of age. Weight, height, and head circumference were 4050 g, 58 cm, and 32.5 cm, respectively, all still markedly below the third centile. He had attained none of the expected developmental milestones and was still maintained on tube feedings since his suck was too weak to deliver adequate calories orally. The baby has been seen in the emergency room on several occasions for recurrent bouts of otitis media and upper respiratory infections but has not required readmission to hospital.

**CYTOGENETIC STUDIES**

Chromosome preparations were obtained from short term phytohaemagglutinin-stimulated lymphocytes. Cells were analysed using GTG banding. The initial analysis of the proband’s karyotype revealed two abnormalities, 5p+ and 13q+. The karyotype of the father was normal. The mother’s analysis revealed a complex chromosome rearrangement involving chromosomes 5, 13, and 15, as illustrated in fig 2, and was interpreted as 46,XX,t(5;15;13)(p15;q22.00; q32). The proband would be interpreted as 46,XY, -5,-13,+der(5),+der(13),t(5;15;13)(p15;q22.00; q32)mat. Thus, he is trisomic for 15q22→qter and it is assumed he is also monosomic for the telomeric region of 5p.

**Discussion**

Complex chromosomal rearrangements occur rarely, as recently reviewed by Pai et al. In those cases where derivative chromosomes come from one parent, only maternally derived abnormalities have been described. This report is consistent with this observation.

The clinical features of patients with trisomy for distal 15q are compared in the table. Only those findings reported in three or more of the five published cases are included. The similarities noted suggest a pattern of common features that may result in an established chromosomal syndrome as more cases are accumulated.

We wish to thank Drs William Stewart and Dana Brasfield for the referral. The technical assistance of Karen Weaver is gratefully acknowledged. This study was partially supported by Project 905, Grant No MCH-011001-30-0, HSA, from the Department of Health and Human Services.

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Requests for reprints to Dr Patricia N Howard-Peebles, Laboratory of Medical Genetics, University Station, Birmingham, Alabama 35294, USA.

Familial pericentric inversion of chromosome 13 resulting in duplication 13q22→qter

SUMMARY A 10-year-old male with a recombinant (13)dup q chromosome is reported. The recombinant chromosome originated from a maternal pericentric inversion which was present in two of his four normal sibs. A segregation analysis of 60 pregnancies from which one of the parents was a carrier of inv(13) showed a significant predominance of males among the viable offspring.

Cases with duplication of a distal 13q segment have led to the detection of familial pericentric inversions.

Received for publication 8 October 1981

FIG 1 Family pedigree including years of birth.

FIG 2 Proband at 10 years of age.
A complex chromosome rearrangement resulting in trisomy 15q22→qter

Patricia N Howard-Peebles, Paula R Scarbrough, Joe Sharpe, Wayne H Finley and Sara C Finley

doi: 10.1136/jmg.19.3.224

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