dentition cannot yet be evaluated. It is the abnormal nasal configuration at birth that initially suggests the diagnosis. The hypoplasia is severe and largely confined to the nasal alae, leaving a central septum with receding nostrils on either side. The nasal bridge is flat and the whole nose is short.

The pancreatic deficiency is exocrine. Proteolytic, lipolytic, and amylolytic enzymes are reduced or absent. The patient reported by Townes and White received pancreatin with all meals from the age of 4 years. Her symptoms improved but at 12 years she was still below the 3rd centile for height and weight and was mildly retarded.

Despite initial uncertainty (Smith states that the aetiology is unknown and the condition sporadic), autosomal recessive inheritance is now confirmed. Sibs are reported by Day and Israel and consanguinity by Schussheim et al. Mardini et al reported sibs in an inbred family. Microcephaly and mental retardation have been reported in all the patients to date, albeit only mildly in some instances.

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Pericentric inversion of chromosome 1 in an azoospermic man

SUMMARY An azoospermic patient with an inherited inversion of chromosome 1 and a normal Y chromosome is described. The mother of the patient has the same inversion.

Pericentric inversion is not an infrequent structural chromosome anomaly in humans, its occurrence being much more common than paracentric inversion. Pericentric inversion is well known in chromosome 9, but it occurs rarely in chromosome 1. To our knowledge pericentric inversion of chromosome 1 has so far been described in ten cases, but its association with male infertility has only recently been published by Giraldo et al. In the present paper we wish to report a male azoospermic patient carrying a pericentric inversion of chromosome 1, inv(1) (p34q23).

Case report

A 26-year-old patient was referred to our cytogenetic laboratory because of infertility. The patient was born after an uneventful pregnancy; he was the second child of a 20-year-old mother and a 26-year-old father. His birthweight was 3000 g. His mother was not known to have had any miscarriages. He has a 27-year-old healthy brother. At examination his height was 180 cm and his weight was 73 kg. There was no history to suggest damage or inflammation of the testes. In the course of andrological examination, azoospermia was established. We found no other abnormality.

TESTICULAR HISTOLOGY

Histological section of the left testis showed numerous tubules. Most tubules were lined only by Sertoli cells or filled with desquamated and degenerated Sertoli cells. Only a few tubules contained spermatogonia and active spermatogenesis was not found. We did not find tubules with thickened tunica propria or with hyalinisation (fig 1).

CYTGENETIC EXAMINATION

Buccal smears were Y body positive. Chromosome studies were done on peripheral blood lymphocytes, using GAG, QFQ, and CBG banding. All cells showed a modal number of 46 chromosomes including one abnormal chromosome. This abnormal chromosome was identified as a pericentric inversion.

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FIG 1 Testicular tubules lined with Sertoli cells with only a few spermatogonia.

FIG 2 Inv(1)(p34q23) of the proband. (a) G banding, (b) C banding.

FIG 3 Idiogram (Paris Conference, 1971) showing presumptive breakpoints on the chromosome 1 involved in the pericentric inversion.

of a chromosome 1 by G, C, and Q banding. The breakpoints were located at bands p34 and q23. Thus, the karyotype of the patient was 46, XY, inv(1) (p34q23) (figs 2, 3).

The father and brother of the proband had a normal 46,XY karyotype, but the mother’s chromosome 1 had the same pericentric inversion as the proband’s. The karyotype of the mother was 46,XX, inv(1) (p34q23).

Discussion

Pericentric inversion occurs relatively frequently in humans. However, it is rarely associated with phenotypic alteration and its clinical significance has not yet been clarified. Cases published in connection with pericentric inversion of chromosome 1 show variation with regard to the clinical consequences.

As to the role of pericentric inversion in infertility, we have little information as yet. Pericentric inversion of chromosome 9 in cases of spontaneous abortion and infertility has been described by several authors and is regarded by some of them as a factor of reproductive risk. The behaviour of pericentric inversions in man and their effect on the offspring have been reported by Winsor et al and Daniel among others. Dutrillaux and Gueguen observed pericentric inversion of chromosome 7 in a male patient with oligospermia. They explained reduced meiotic activity by “aneusomie de recombinaison”.

A genetic cause for male sterility (oligospermia or aspermia) is usually found in anomalies of the sex chromosomes, particularly in the Y chromosome. Autosomal structural rearrangements or X or Y;autosome translocations may also result in azoospermia.

In our case the sex chromosomes of the patient, including the fluorescent segment of the Y chromosome, seemed to be intact. We did not find any structural aberration in the autosomes except the inversion of chromosome 1 inherited from the mother. Like our patient, three azoospermic brothers reported by Giraldo et al had pericentric inversion of chromosome 1, and Croquette et al, in a carrier of inversion of chromosome 9, found total aspermia but no other phenotypic anomaly. According to Croquette et al, if there is a relationship between sterility and the chromosome anomaly, the inversion does not result only in ‘aneusomie’ but can disturb meiotic chromosome pairing too. Chandley suggested that sterility in autosomal translocation heterozygotes is expressed only in male carriers, because male meiosis seems to be more sensitive to structural chromosome rearrangements than female. Our case, like the cases of Giraldo et al, suggests that this hypothesis might be applied not only to translocation carriers, but inversion carriers too.

It cannot be excluded that the chromosome abnormality and the spermatogenic arrest may be only a chance association of two unrelated phenomena in the same person.

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X long arm deletion with oligomenorrhoea

SUMMARY A 35-year-old female patient with oligomenorrhoea had a deletion of the long arm of the X chromosome. The breakpoint at band q23 caused infertility in spite of excessive pituitary stimulation. The aberrant X chromosome was inactivated in all cells analysed.

Deletion of the long arm of the X chromosome is one of the most frequent structural aberrations of this chromosome. Fertility and reproduction are affected by these chromosomal anomalies.1–3 This paper presents a patient with a deletion of the long arm of the X chromosome and oligomenorrhoea.

Case report

A 35-year-old patient was admitted to the Clinic of Gynaecology and Obstetrics for examination because of oligomenorrhoea. Menarche had occurred at the age of 11, but menstruation was irregular at 1 to 3 month intervals, these intervals becoming progressively longer. Her weight and height were 51 kg and 160 cm, respectively. The breasts were well developed and pubic and axillary hair was normal. Laparoscopy showed that the uterus, fallopian tubes, and ovaries were normal. Gonadotrophins were raised (FSH 76 mIU/ml, LH 68 mIU/ml) and the oestradiol level was high (241 pg/ml). Laparoscopy showed that ovulation was remarkable on the right ovary.

CYTOGENETIC FINDINGS

Forty-two metaphases from peripheral blood were

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![Figure Aberrant X chromosome in situ (RBA banding).](http://jmg.bmj.com/ on June 16, 2017 - Published by group.bmj.com)
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