male genitalia and secondary sex characteristics were normal. FSH plasma levels were raised (19.4 IU/l), but LH (11 IU/l) and prolactin (7.0 µg/l) were normal and plasma testosterone level was decreased. No information on sperm count was obtained.

Biopsies from the left and right testicles showed similar features: a decrease in diameter of the seminiferous tubules with dystrophic lesions. Numerous spermatogonia and pachytene spermatocytes only were seen but no secondary spermatocytes.

Chromosome investigations were carried out on peripheral blood cultures. PHA stimulated lymphocytes were examined after R and C banding. All 37 R banded metaphases examined showed a reciprocal translocation t(Y;1)(q21;p13) (fig 1a, b). C banding confirmed that the breakpoint in the Y chromosome had occurred within the heterochromatic segment (fig 1c). The proband is being treated for Hodgkin's disease and refused further investigations. His relations also refused to have a blood sample taken.

Y:acrocentric translocations appear not to affect the phenotype and they may be transmitted as a chromosome variant through both males and females. There was no increased incidence of infertility or spontaneous abortions in the families of the patients described by Smith *et al*¹ and they may be associated with a normal Y. These translocations apparently involve only heterochromatic material consisting of highly repeated DNA sequences which are not transcribed.

Y:non-acrocentric translocation is commonly associated with primary infertility and, in those cases where family studies were carried out, the abnormality had arisen *de novo*. The mechanism leading to infertility in these translocations is not known, but abnormal segregation at meiosis may be involved in maturation arrest. The translocated Y induces an abnormal sex vesicle and two opposing results are possible. Either the Y segment not included in the sex vesicle is not normally inactivated, or the autosome translocated onto the Y is partly inactivated by a spreading effect.²

Our case is the second report of Y;1 translocation. The first one³ described a four month old male infant with t(Y;1)(q11;q21) who showed severe psychomotor retardation, infantile spasms, and hypotonia. These clinical abnormalities, like the Hodgkin's disease in our case, may be coincidental to the chromosomal rearrangement.

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Distal trisomy 14q

At least 20 cases of trisomy for the distal part of 14q have been reported.¹⁻³ The main clinical signs of this trisomy are common to all types of autosomal imbalance. They include prenatal growth retardation, physical and psychomotor retardation, wide sutures and fontanelles, a prominent forehead, hypertelorism, a protruding upper lip, a high arched palate, anomalous ears, micrognathia, heart defects, and cryptorchidism. Because of this, it is difficult to diagnose distal trisomy 14q clinically. The phenotypic heterogeneity might be caused by both the genetic composition of the long arm of chromosome 14 and genetic heterogeneity in the cases described. The present report describes a case of trisomy 14q24→q32 resulting from a paternal insertion (4;14).

The proband was the product of a term first pregnancy of unrelated healthy 25 year old parents. Her birth weight was 2370 g and length 45 cm. The following clinical features were noted at birth: haematoma of the eyelids, dystonia, opisthotonus, spasticity and hyperreflexia of the lower extremities.

Severe physical and psychomotor retardation was evident at nine months. Her weight was 5000 g, length 61 cm, and

FIG 1 The patient at nine months

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FIG 2 Partial karyotype of the proband (top row) and her father (middle and bottom rows). Note the insertion of $14q24 \rightarrow q32$ into $4p14$.

head circumference 43 cm. She was excitable and unable to sit or stand. She had dystonia of the muscles and rigidity of the femoral joints. Physical examination revealed a prominent occiput, widely open fontanelles (the anterior fontanelle was 4×4 cm), a prominent forehead, narrow palpebral fissures, sparse hair, eyelashes, and eyebrows,

micrognathia, prominent tented upper lip (fig 1), and a high palate. X ray showed normal skull, spine, and bones. The bone age was retarded and corresponded to five months. There was no evidence of cardiac abnormality. Concentrations of thyroxin, TSH, and growth hormone were normal.

G banded chromosome analysis from lymphocytes showed a $4p+$ chromosome in the proband and a balanced translocation between chromosomes 4 and 14 in the father (fig 2). This was interpreted as an insertion translocation of the distal part of $14q$ into the short arm of chromosome 4. The breaks appeared to be in bands $14q24$, $14q32$, and $4p14$. The karyotypes of the proband and her father were: $46,XX,-4,+der(4),dir\ ins(4;14)(p14;q24q32)pat$ and $46,XY,dir\ ins(4;14)(p14;q24q32)$ respectively. The mother's chromosomes were normal.

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