The frequency of mental retardation in hypochondroplasia

Hypochondroplasia is a relatively mild skeletal dysplasia usually with less growth retardation than achondroplasia. It is clearly autosomal dominant and many familial cases have been reported.

One difficulty in genetic counselling is the association between hypochondroplasia and mental retardation. Langer1 has suggested that 20% of cases of hypochondroplasia suffer from mild mental retardation. Hall and Spranger2 reviewed 32 patients in whom there was both clinical and radiological data, and found three out of 32 (9.4%) with mental retardation. The mental retardation appears to be found in both sporadic and familial cases, as Frydman et al.3 reported developmental delay in the son of an affected but intellectually normal mother.

In order to obtain a more accurate figure for counselling, we reviewed the data on hypochondroplasia from the Skeletal Dysplasia Group's Register. There were 27 cases of hypochondroplasia (15 female, 12 male) of whom 11 were familial cases. They had been ascertained through orthopaedic genetic clinics. The age range was 2 to 69 years with a mean of 27 years. Mental handicap was not a feature in any of the 27 cases. This suggests to us that the figures of 10 to 20% frequency of mental handicap may be overestimating the true frequency of mental handicap in this disorder.

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Unusual inheritance of Becker type muscular dystrophy

Tommerup1 has recently pointed out that a family published by us in 1978,2 in which we suggested an unusual inheritance of Becker type muscular dystrophy (BMD), was compatible with the presence of a familial reciprocal X;autosome translocation. At that time there were three generations containing affected subjects including the mother and grandmother of the index case, who also had two brothers with the disease. The affected females had fairly severe symptoms and we proposed that, by chance, the normal X chromosome was inactivated in more cells than in the usual non-manifesting heterozygote. Tommerup1 suggested that a balanced X;autosome translocation would explain this type of non-random X inactivation, and he further stated that this abnormality had been found in at least 20 females with BMD or Duchenne muscular dystrophy.3,4

Chromosome studies were not done in the initial study of the family; however, after exchanging letters with Dr Tommerup, we decided to perform them in order to test the above hypothesis. When the family was contacted again, it was found that the proband now has a male child who is 12 years old and is very likely also affected, as indicated by bilateral calf hypertrophy and abnormally raised serum CK levels found by another physician several months before we examined him.

In spite of this probable male to male transmission of the disease, which eliminates the possibility of it being X linked, we went ahead with the chromosome studies to rule out rare situations, such as the son being a 46,XX male. We studied the proband, his wife, and their affected male child. Peripheral blood samples were obtained and cultured in the usual manner with PHA stimulation. After harvesting, the slides were stained to obtain G bands. Thirty metaphases from each subject were analysed and all three subjects were found to be cytogenetically normal.

We now believe that our family represents an unusual, dominantly transmitted form of muscular dystrophy, rather than an unusual inheritance of BMD, as we could find no similarly published case.

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Haematometra in the Langer-Giedion syndrome

Recently we reviewed a 25 year old woman with the Langer-Giedion syndrome whose case had been diagnosed and reported at the age of 8 years by Koslosski et al.1 in 1977. She was mildly intellectually handicapped and lived at home with her mother. Her general health was fair. She had short stature (132 cm) accentuated by a moderate thoracic kyphosis. Her facies was characteristic of the syndrome but with some overall coarsening (figure). She had multiple exostoses visible and palpable at the wrists and ankles; one on her scapula had required surgical excision. Over the years her mild deafness had not increased but she had two major medical problems. The first was ureteric reflux (noted when she was aged 8 years) with recurrent urinary infections; in the past year she had had her ureters reimplanted in the bladder.

The second problem had been episodic abdominal pain starting about the age of 15 years. At that time she had had some blood loss from the vulva which was interpreted as scanty menstruation but, in retrospect, was more likely associated with haemor-