Camptodactyly, cleft palate, and club foot (the Gordon syndrome)

A report of a large pedigree

FAHED HALAL AND F. CLARKE FRASER

From the Department of Medical Genetics, The Montreal Children's Hospital, Montreal, Canada

SUMMARY A second family is described in which camptodactyly, club foot, and cleft palate (the Gordon syndrome) is transmitted in a pattern consistent with autosomal dominant inheritance with reduced penetrance and variable expressivity. Penetrance appears to be more reduced in females than in males, and cleft palate is the least frequently manifested trait.

Since Gordon *et al.* (1969) first described the syndrome of camptodactyly, cleft palate, and club foot in a family showing autosomal dominant transmission through 3 generations, no further reports have appeared. We wish to report a family in which the Gordon syndrome shows autosomal dominant transmission through 5 generations. Expression ranges from quite severe camptodactyly, club foot, cleft palate, and undescended testes to no manifestations of the syndrome. So far, the asymptomatic carriers are all females, and the most severely affected members are all males.

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Family history

The proband (Fig. 1, IV.29) was a 45-year-old Caucasian male who consulted us because of a history of cleft palate in his family. He said that his wife, who had recently conceived unexpectedly, was worried about the risk of recurrence of such a deformity. Camptodactyly and club foot were also present in his family, but did not seem to worry them as much. The proband himself had a normal palate, mild camptodactyly of several fingers (Fig. 2 and 3), and moderate talipes equinovarus which had been corrected by home physiotherapy during the

Fig. 1 Pedigree of family.

Fig. 2 Dorsal view of proband's hands, showing camptodactyly of 5th fingers.

Fig. 3 Ventral view of proband's left hand, at limit of extension.

first few months of life. He had also had an undescended left testis which came down 'after a jump' at the age of 6. A sperm count performed recently showed 290 000 sperm/ml with good motility. No other significant features were found at physical examination.

The presence of cleft palate, camptodactyly, and talipes equinovarus in other members of the pedigree are shown in Fig. 1.

Discussion

Although the proband was unwilling to let us examine other members of the family, he was well aware of the

features of the syndrome and well informed about their occurrence in his relatives. We therefore consider the reporting to be accurate with respect to the affected individuals and obligate carriers. However, the large number of individuals with whom the proband had lost contact made it impossible to establish accurate segregation ratios. Nevertheless, the pattern is consistent with autosomal dominant inheritance and the two instances of male-to-male transmission rule out X-linked inheritance. Most of the female carriers in this family were either apparently normal (III.5, 6, 7, 10; IV.19, 25, 31) or mildly affected with camptodactyly of the 5th finger (III.12, 13). In contrast, 16 of the 21 affected males had camptodactyly and club foot; 4 had cleft palate, as well, forming the complete triad (III.4, IV.13, IV.13, V.18). The frequency of cleft palate in affected individuals (4/26 affected, or 4/33 known gene carriers) was lower than in the Gordon family (3/6). Four males had camptodactyly alone. Seven unaffected and 4 affected females transmitted the gene, so that penetrance is much reduced in females. All 3 males who transmitted the gene were affected.

Undescended testes were present in 2 affected males, with subfertility in one and infertility in the other, and this may be another component of the syndrome. The proband in the Gordon family had also had bilateral cryptorchidism which was corrected surgically at the age of 11. Other traits occurring in affected members of our family included stunted growth and pigeon chest in one male (III.1) and scoliosis in another (IV.23).

This family confirms that the familial aggregation of camptodactyly, cleft palate, and club foot constitutes a separate genetic entity and transfers the Gordon syndrome from the 'private' to the 'public' sector. There is considerable variation in expressivity and penetrance is reduced, at least in females. Cleft palate, which was the feature of most concern to the proband in terms of his unborn baby, occurs in minority (roughly 20%) of cases.

Reference

- Gordon, H., Davies, D., and Berman, M. (1969). Camptodactyly, cleft palate, and club foot. A syndrome showing the autosomal-dominant pattern of inheritance. *Journal of Medical Genetics*, 6, 266-274.

Requests for reprints to Professor F. Clarke Fraser, Department of Medical Genetics, The Montreal Children's Hospital, Montreal, Canada H3H 1P3.