Case reports

this may be so. In any event, further reports of cases with these chromosome abnormalities will be of great importance in establishing these two possible new syndromes.

The authors wish to express their thanks to Dr Yanagisawa, Department of Paediatrics, Yamaguchi University School of Medicine, for information on the family. Thanks are also due to Dr Matsui, Kanagawa Children’s Medical Center, for cord-blood culture. This study was aided in part by a grant from the Ministry of Health and Welfare.

YASUO NAKAGOME and HIDEO KOBAYASHI
Department of Human Genetics, National Institute of Genetics, Mishima and Department of Obstetrics and Gynecology, Kawasaki City Hospital, Kawasaki, Japan

REFERENCES

Stub thumbs

Summary. A case of familial brachydactyly is reported.

Bell (1951) and Temtamy and McKusick (1969) have classified the different types of brachydactyly due to maldevelopment of the phalanges or metacarpals. It may occur as an isolated phenomenon or as part of a syndrome. Type D brachydactyly or 'stub thumbs' is characterized by shortening and broadening of the terminal phalanges of the thumb and big toes. It is usually an isolated finding but has been reported in association with cardiac arrhythmias (Tabaznick, 1965).

Received 22 July 1974.

Case report

A 49-year-old manual worker was admitted to the Cardiff Royal Infirmary with a history of myocardial ischaemic pain. His stay was uneventful but he demonstrated a marked abnormality of both thumbs. They were bulbous and shortened. The nail was broad (Fig. 1). There was no abnormality of his toes. Radiology of the hands showed a shortening of the terminal phalanx in the thumbs only (Fig. 2).

His family, including reportedly the whole of the second generation, demonstrated this abnormality (Fig. 3). II.1, II.2, and II.3 had lost contact with the rest of the family and it is not known whether their progeny were affected. There were no other congenital abnormalities noted in the family.
The husband of III.2 (III.1), a fit middle aged male, was found to have a shortening limited to the thumb of his left hand (Fig. 4). His family was in no way related to that of the propositus and to his knowledge he was the only member with a short thumb.

Two of the offspring of III.1 and III.2 were unaffected, the third (IV.1) showed a left stub thumb and she has recently produced a daughter (V.1) with normal thumbs.

**Discussion**

Bilateral stub thumbs occurring as an isolated defect are a well-demonstrated abnormality with an autosomal dominant mode of inheritance. Previous papers have shown the penetrance to be variable but in the pedigree demonstrated there is no evidence of lack of penetrance although IV.1 is only unilaterally affected.

Unilateral brachydactyly of this type has been described by Sayles and Jailer (1934). All the members of generation II were allegedly affected. This seems unlikely but unfortunately only three sibs could be examined personally. However, they were quite positive that all their generation had short thumbs.

In this kindred an unrelated spouse III.1 also showed a unilateral short thumb which showed minor clinical differences from the others in that although the terminal phalanx was affected it was not as markedly broadened nor was the nail of the same shape. The fact that IV.1 has an unaffected daughter V.1 is proof that she cannot be homozygous even if the traits demonstrated by her parents were allelic. The clinical similarity of IV.1 to the rest of the family suggests that she has inherited her trait from her mother III.2.

The family recognized the defect as being inherited and carefully inspected every newborn for abnormal thumbs; however, they rightly assumed that possession of brachydactyly of this type is usually of no clinical importance.

**A. B. Davies (M.B. M.R.C.P.)**

S.H.O. Department of Respiratory Medicine, Hammersmith Hospital, Ducane Road, London W12.
Autosomal recessive oculopharyngeal muscular dystrophy

Summary. Oculopharyngeal muscular dystrophy is known as a rare autosomal dominant disease. A family is reported suggesting that there may be genetic heterogeneity in oculopharyngeal muscular dystrophy and that in some families the mode of inheritance may be autosomal recessive.

Progressive dystrophy of external ocular muscles (ocular myopathy) was reviewed by Kiloh and Nevin (1951). In 1962, Victor et al described and gave the name of oculopharyngeal muscular dystrophy to a familial disease of late life characterized by dysphagia and progressive ptosis of the eyelids. Their report included a sporadic case and a family which demonstrated an autosomal dominant mode of inheritance. Since then several families and some single case reports have been published and the nosological entity of oculopharyngeal muscular dystrophy has been accepted as a hereditary disease due to an autosomal dominant gene (Hayes et al, 1963; Schotland and Rowland, 1964; Teasdale et al, 1964; Bray et al, 1965; Aarli, 1969; Graf, 1971; Penchazadeh and Teasdale, 1971; Szobor, 1973).

The purpose of this report is to present a family suggesting that there may be genetic heterogeneity in oculopharyngeal muscular dystrophy and that in some families the mode of inheritance may be autosomal recessive.

Case reports

The proposita, case 1 (VI.10, Fig. 1), was the youngest in her sibship. The family is Ashkenazi Jewish of Hungarian origin. The parents of the proposita are both first cousins once removed and third cousins. An older sister, case 2 (VI.9), is also affected. The rest of the family is reported to be without muscle disease except one distant relative (V.3) who is said to have a disease similar to that of the two sisters. This relative is herself the product of first-cousin marriage. She lives abroad and unfortunately details of her condition were not available. Both parents and the eldest brother of the proposita were personally examined. The father and mother were found to be in good health and without muscle weakness at the age of 75 and 70, respectively. The three brothers were healthy at ages 47, 46, and 40 years.

Case 1. (VI.10) is a 37-year-old woman, mother of three healthy children. From the age of 34 she noticed mild bilateral ptosis, and gradually she developed difficulty in looking forward. There were no other complaints. The patient was admitted to the Neurology Department for investigation. She used to wrinkle her forehead and extend her neck in an attempt to look forward. She never had double vision. Neurological examination revealed bilateral symmetrical ptosis (Fig. 2) down to the midpupillary level. The facies was expressionless. Eye movements were limited in all directions but especially upwards. The patient was not able to raise her eyes above the horizon but there was no limitation in convergence. The rest of the neurological examination revealed bilateral drop foot and the ankle reflexes could not be elicited. There was no myotonia or muscle fibrillation. The gag reflex was reduced on both sides. Chest radiography was normal and the thymus was not enlarged. EEC and ECG were normal. CK and LDH levels were normal. EMG of the lateral recti of both eyes showed potentials of low amplitude and short duration and no fibrillation was observed, indicating a primary muscle disease. EMG of the orbicularis oculi and of the frontalis muscle