Original investigations

The Coffin-Lowry syndrome
A study of two new index patients and their families

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Abstract. Two adult, mentally retarded males with the typical features of the Coffin-Lowry syndrome are reported. Further family investigation led to the same diagnosis in a 2.5-year-old male cousin, and to the identification of five female carriers, with variable clinical expression of this X-linked inherited mental retardation syndrome.

Key words: Coffin-Lowry syndrome – X-linked mental retardation – Dysplasia syndrome

Introduction

X-linked mental retardation, which is responsible for the excess of males among the mentally retarded [13] constitutes a heterogeneous group of different entities. After the discovery [7] and the confirmation of the fragile X syndrome [11], medical interest in this subject has increased during the past few years. Within this group of disorders the Coffin-Lowry syndrome constitutes a rare, clinically recognisable entity, with partial expression of the clinical stigmata in female carriers [4].

Report on the families

Family I

S.J., a male (III1, Figs. 1–4), was the eldest in a family of four children, two boys and two girls. His psychomotor develop-
ment was retarded from the beginning and he only started to walk at 24 months. As a child he was admitted to a special institute for the mentally retarded and examined during a systematic study of the mentally retarded.

At his present age of 35 years his length is 152 cm, his weight 70 kg and his head circumference 56 cm. His face is characteristically coarse with heavy bowed eyebrows, hypertelorism and down-slanting of the somewhat narrowed palpebral fissures. Other facial anomalies included large ears, a big nose with a broad and flattened nasal tip and a large open mouth with full everted lips (Fig. 2). Skeletal deformities were severe with thoracolumbar kyphosis, pectus carinatum and calcaneovalgus deformity of the feet. His hands were large and soft (Fig. 3) with tapered fingers and a hypothenar crease. Radiographic examination of the hands showed hypoplastic, drum-stick type terminal phalanges (Fig. 3); in the spine dysplasia of the thoracolumbar vertebrae was seen (Fig. 4).

The mother (II.), aged 58 years, has a round face with prominent brows, a broad nasal bridge with thick septum and full, everted lips. Her hands show typical tapering of the fingers and a transverse crease on the hypothenar eminence. Her mental state is difficult to evaluate due to a cerebrovascular accident some years ago, but family members describe her as mentally retarded. She had three healthy brothers (II.,) and one sister (I.). Her sister has been hospitalised in a psychiatric hospital since the age of 20 years due to psychotic behaviour and slight mental retardation. Her face shows prominent brows and a very lax skin with “peau de pêche” appearance. Her hands and fingers are normal.

The eldest sister (II.) could not be examined but photographs reveal small stature and coarse facial features like her mother, with typical tapering fingers. She is slightly mentally retarded, and has three children, two boys and one daughter. The youngest son (IV.) was born after a normal pregnancy and delivery. The birth weight was 2900 g, the length 49.5 cm. At the age of 2 months surgical correction of an inguinal hernia was performed. Soon after birth severe developmental delay was noted. At 2 years his length and weight were below