Syndrome of the month

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Fountain's syndrome: mental retardation, sensorineural deafness, skeletal abnormalities, and coarse face with full lips

JEAN-PIERRE FRYNS
From the Centre for Human Genetics, University of Leuven, Belgium.

In 1974 Fountain[1] was the first to report on the association of mental retardation, deafness, and skeletal abnormalities in four sibs (three boys and one girl). Two of them developed progressive swelling of the lips and in one of them an eroded, granulomatous mass appeared in the lower lip.

In 1987 we examined three moderately to severely mentally retarded males, two brothers and one isolated male patient, with the same association of symptoms.[2]

No other reports have dealt with this apparently autosomal recessively inherited mental retardation-deafness syndrome.[3]

Major clinical findings

MENTAL RETARDATION

All seven patients reported so far were mentally retarded. The degree of mental retardation was variable. Three of the four sibs reported by Fountain[1] were dull to slightly mentally retarded. The fourth sib "died, and in addition to being deaf, dumb, and mentally retarded, had spina bifida". An IQ of 75 was noted for one sib. One of the brothers of the second report[2] was mildly mentally retarded and now works in a sheltered workshop. His brother is severely retarded. The isolated male patient showed severe psychomotor retardation from the beginning and as a young adult now lives in an institute for the severely mentally retarded.

DEAFNESS

In all the reported patients profound deafness was noted between the ages of 15 and 18 months. Audiometry confirmed the presence of profound sensorineural deafness with rudimentary hearing at the lowest frequencies. Vestibular function was normal. Tomography of the pars petrosa of the temporal bones[2] showed congenital anomalies of the spirals of the cochlea; in the youngest the cochlear spirals were replaced by a simple cavity. In the isolated male patient[2] deafness was only suspected at the age of four years, but this late diagnosis was probably related to the severe mental retardation of the boy. Audiometry also confirmed complete sensorineural deafness in this case.

FACIES

Fountain[1] reported granulomatous enlargement of the lips and gums in two of the four sibs. In the adult female sib the swelling of the lip developed after the age of 20 years. There was a progressive, gross, papular, erythematous swelling of the skin of the cheeks, upper lip, and chin and the lower lip was an eroded, granulomatous mass. The adult male sib had a progressive swelling of the upper and lower lips and the lower lip became red.

The oldest brother in the second report[2] had a peculiar, round, coarse face with mild swelling of the subcutaneous tissue, particularly of the cheeks and lips (figs 1 and 2). The facial stigmata were more pronounced in the youngest brother: he had a long and coarse face with a large, open mouth, full, everted lips, high palate, and 'mandibular prognathism'. The swelling of the subcutaneous facial tissues, especially the lower lip, became more evident between the ages of 13 and 26 years. In the third patient, facial plethora was already present at the age of four years, but the oedematous infiltration of the cheeks, lips, and gums with thick, everted lips was only noted when he was re-examined at the age of 17 years.

As already noted by W B Reed in Fountain's report, the oedematous facial changes are very

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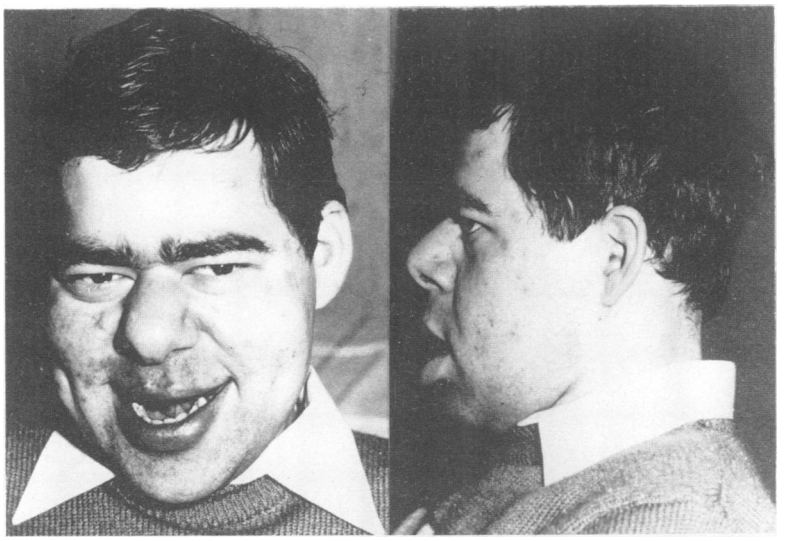

FIG 1 *Long and coarse face and swelling of the subcutaneous tissue, especially of the lower lip (26 years).*

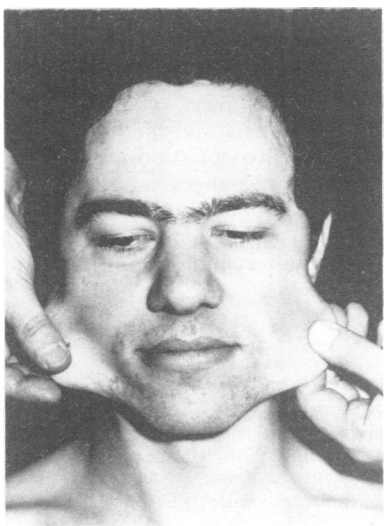

FIG 2 *The facial oedema and the hyperlaxity of the skin.*

similar to those seen in the Melkersson-Rosenthal syndrome,[4] an autosomal dominant condition often manifesting in childhood or youth with swelling, especially of the lips.

SKELETAL ABNORMALITIES

Gross thickening of the calvarium was present on skull *x* rays in three of the four sibs of the first report.[1] Other clinical abnormalities were not mentioned. The three males of the second report[2] had short, stubby hands with broad and short terminal phalanges (fig 3).

X ray examination confirmed the presence of broad, heavy phalanges and metacarpals with thick cortices but without ossification defects. Skull *x* rays showed the same marked thickness of the calvaria in the brothers as present in the sibs reported by Fountain.[1]

Additional features

GROWTH AND PHYSICAL DEVELOPMENT

Birth weight and length were normal in all patients. Adult height, however, was below 153 cm in the sibs reported by Fountain.[1] Adult height and general physical development were normal in the two brothers reported by Fryns *et al,*[2] but the third isolated male patient showed marked growth retardation (143·5 cm) at the age of 17 years.

NEUROLOGICAL FINDINGS

The two brothers of the second report developed infantile spasms after the age of three months, which later became generalised infantile seizures, more severe in the youngest. All patients had general hypotonia, which persisted in adulthood and resulted in secondary scoliotic deformities of the vertebral column.

Heredity

The occurrence of this mental retardation-deafness

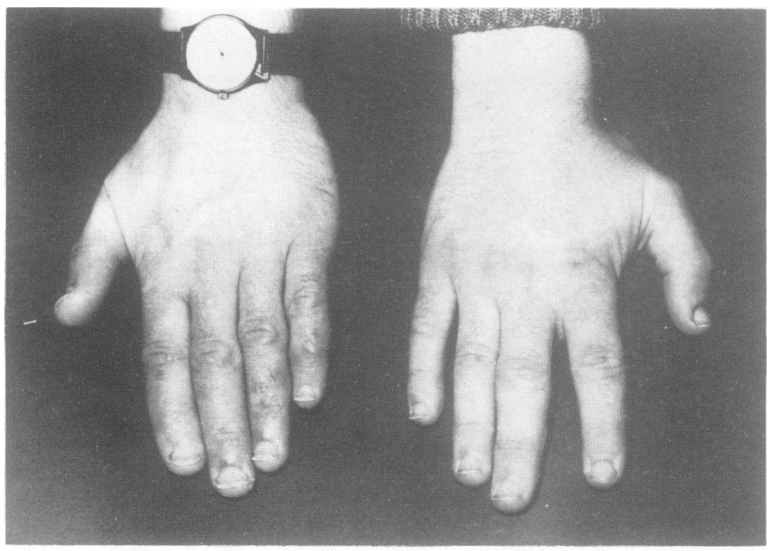

FIG 3 *Broad and plump hands with short, stubby fingers.*

syndrome in sibs of normal parents suggests autosomal recessive inheritance.

Pathogenesis and incidence

Extensive biochemical and metabolic examinations, including calcium, phosphorus, creatine kinase, amino acid chromatography of serum and urine, and urinary excretion of mucopolysaccharides, were normal. Mannosidosis and aspartylglucosaminuria were excluded in all patients.

Other examinations, such as electrocardiography, electromyography, electroencephalography, ophthalmological examination, and peripheral nerve and rectal biopsies, showed no specific abnormalities.[2] As in the Melkersson-Rosenthal syndrome,[4] the pathogenesis of the facial oedema with thickening of the cheeks, lips, and gums remains unknown. Gingival biopsy performed in one patient[1] showed a granulomatous infiltrate marked by large, foamy cells that did not contain fat.

The prevalence of the syndrome is unknown. In Leuven we observed four patients (one additional four year old male after the original description[2]) over a period of 15 years during a genetic-diagnostic evaluation of nearly 500 patients with a combination of mental retardation and deafness.

Differential diagnosis

At a younger age the facial appearance with a round, plethoric face and full lips may suggest the diagnosis of Coffin-Lowry syndrome.[5] The severe sensorineural deafness and the absence of skeletal anomalies, that is, hypoplastic drumstick appearance of the terminal phalanges and dysplasia of the vertebral bodies, which are typical findings in the Coffin-Lowry syndrome, are the most important features in the differential diagnosis of both conditions.

References

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Correspondence to Professor J P Fryns, Centre for Human Genetics, Herestraat 49, B-3000 Leuven, Belgium.