

## Ulnar-mammary syndrome

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In 1975, McKusick<sup>1</sup> first mentioned a syndrome of ulnar ray defects and genital abnormalities, citing a personal communication from A Schinzel (1973), and named it 'Schinzel syndrome' (McKusick No 18145) in this and in all subsequent editions of his catalogue.<sup>2</sup> One year later, Pallister *et al*<sup>3</sup> and Gonzales *et al*<sup>4</sup> described the same syndrome in two families, and the latter authors named the syndrome 'ulnar-mammary syndrome type Pallister'. McKusick<sup>5</sup> adopted this name in his catalogue from 1978 (McKusick No 19145), and since the fifth edition, both codes are found separately in *Mendelian inheritance in man*. Gonzales *et al*<sup>4</sup> found a paper from the 19th century in which another woman with possibly the same syndrome was described.<sup>6</sup> Temtamy and McKusick presented findings and photographs of Schinzel's family in their book *Genetics of hand malformations* and a

detailed report on the original family of Schinzel and Prader<sup>8</sup> is under way. Further families were reported by Hecht and Scott<sup>9</sup> and briefly mentioned by Sherman *et al*,<sup>10</sup> increasing the number of reported cases to 12.

### Major clinical findings

(1) Ulnar ray defects (figs 1 to 3) range from absence of three, two, or one ulnar rays of the fingers to hypoplasia of the distal phalanges of the little fingers and nails with stiff distal interphalangeal joints. Hypoplasia of the little fingers may be combined with incomplete duplication (postaxial hexadactyly) and the extent of the defects is often different between the left and right side. Carpal bones may be absent on the ulnar side, and forearms may be shortened with a hypoplastic or absent ulna, bowed radius, and stiff elbow. Feet may show hypoplasia of the little and fourth toes including the corresponding

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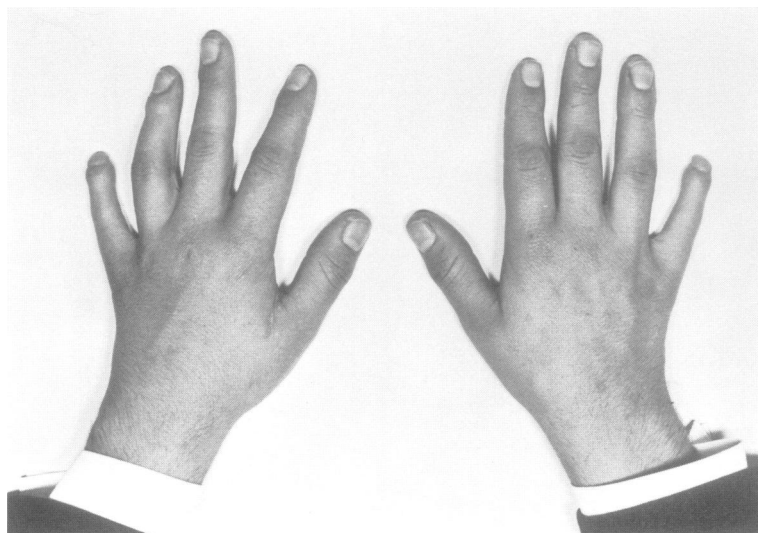


FIG 1 Short little fingers with deformed nails in a 22 year old male.

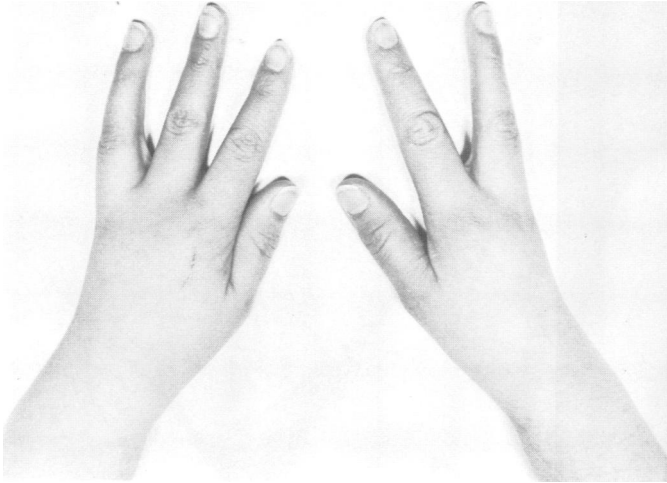


FIG 2 Hands of a 19 year old male with absence of left fifth and right fourth and fifth rays.



FIG 3 Radiograph of the left hand of a 22 year old male with hypoplastic terminal phalanx of little finger.

nails, but major fibular ray defects have not so far been observed.

(2) There is hypoplasia or aplasia of the mammary glands and hypoplasia of the nipples (fig 4), which may remain unnoticed in males. Inability to breast feed probably caused grossly reduced numbers of surviving offspring to affected mothers in earlier centuries.<sup>6</sup>

(3) There is hypoplasia of the apocrine glands, particularly axillary apocrine glands, with reduced body odour. Axillary and body hair is sparse, but pubic hair is normally developed.

(4) There are genital and pubertal anomalies in males. In contrast to females, males with this syndrome are obese and experience delayed growth and skeletal maturation. Puberty and catch up growth occur, but are about five to seven years delayed, sometimes as late as in the third decade. Many, if not most, exhibit hypoplasia of the external genitalia with cryptorchidism, small penis, and small testes persisting through adulthood (fig 5), and libido as well as sperm count are grossly reduced. Thus, fertility is reduced in affected males, and the degree of genital hypoplasia and reduced fertility seem to correlate with the extent of ulnar ray deficiency.<sup>8</sup>

**Other anomalies less commonly associated with the ulnar-mammary syndrome**

These may affect different organs. In patients with severe ulnar ray deficiency and short forearms, the

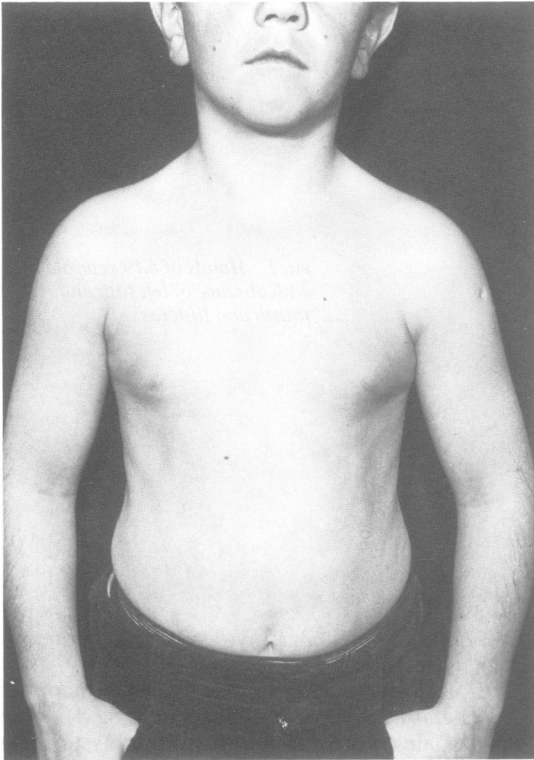


FIG 4 Hypoplastic, underpigmented nipples in a nine year old boy.

humeri, scapulae, clavicles, and pectoralis major muscles may be hypoplastic. Out of 12 patients, one had subglottic stenosis and two had pyloric stenosis, anal atresia/stenosis, imperforate hymen, kidney malformations, inguinal hernias, and hypodontia. Congenital heart defects not investigated in detail were present in another two members of one family.<sup>10</sup>

#### Inheritance and aetiological factors

The ulnar-mammary syndrome is caused by an autosomal dominant pleiotropic gene. Since expression of the ulnar ray defects may be very mild, and hypoplasia of the apocrine glands may remain unnoticed, it is possible that this mutation is present in some families with 'aplasia of the breasts'.<sup>11</sup> The gene action is still unknown, the gene is unmapped, and no defect in endocrine metabolism is known so far.



FIG 5 Hypoplastic genitalia in a 19 year old male.

#### Diagnosis

The diagnosis of the ulnar-mammary syndrome should be considered in any patient with ulnar ray defects plus other congenital anomalies and normal intelligence. Anomalies of the apocrine glands and mammary glands may remain unrecognised before (and in males even after) puberty, and external genitalia and puberty are normal in females. Autosomal dominant inheritance confirms the diagnosis, and on thorough clinical examination of further family members additional carriers of the gene with mild expression may be detected.

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