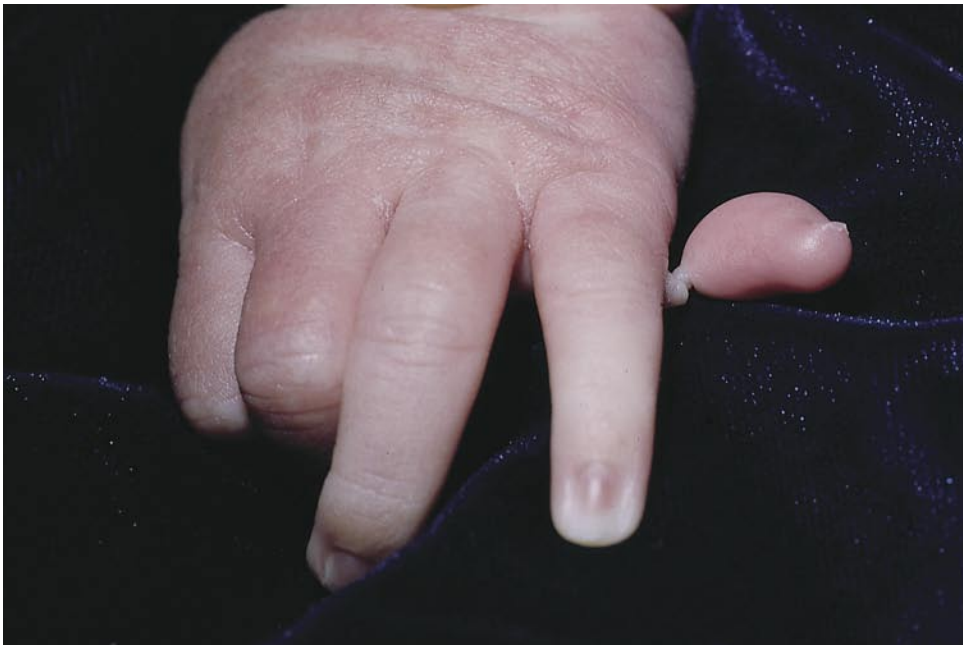
# What Is Your Diagnosis?



The patient has an appendage attached by a thin stalk. What other problems may be associated with this physical finding?

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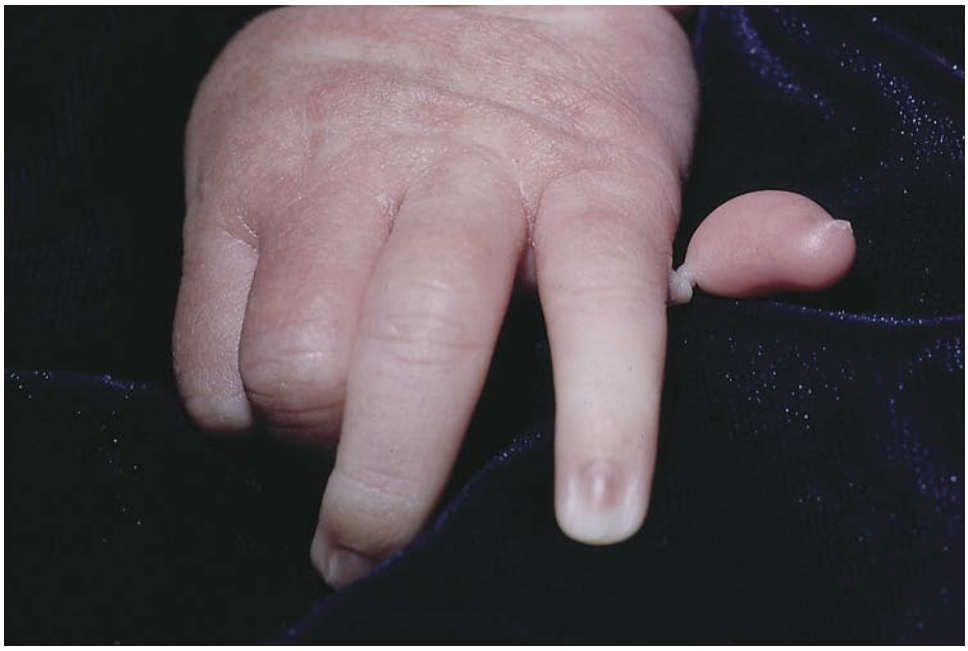
# The Diagnosis: Supernumerary Digit



Supernumerary digits most commonly are noted on the ulnar aspect of the hand. The lesions usually present as small papules at the base of the fifth digit. Histologic evaluation reveals a thick stratum corneum, an unremarkable epidermis, and a fibrovascular core that frequently contains many nerve bundles. The nerve bundles may represent a small amputation neuroma resulting from intrauterine amputation of the digit. Supernumerary digits are differentiated from acquired digital fibrokeratomas by presenting at birth and containing nerve bundles. Occasionally, the digit is formed more completely, containing bone, cartilage, and a nail apparatus. When the digit is connected only by a thin stalk of tissue, as in this case, the digit easily can be removed with surgical scissors.

Polydactyly is the generic term for partial or complete duplication of a digit and represents the most commonly reported hand abnormality, with an incidence of approximately 1 to 2 cases/1000 live births.[1,2] Most cases represent rudimentary supernumerary digits that resemble skin tags or small papules. Completely formed duplicated digits are less common, occurring in approximately 0.014% of live births.[3] Polydactyly can occur as an isolated finding, associated with other limb abnormalities or as part of a syndrome. About 15% of patients with polydactyly have other congenital anomalies.[4] Polydactyly that is not part of a syndrome commonly is inherited as an autosomal dominant trait.[5] Filippi syndrome, characterized by syndactyly, microcephaly, growth retardation, and mental deficiency, is inherited as an autosomal recessive trait.[6] Polydactyly also has been reported in Filippi syndrome.[6] Additionally, polydactyly may be associated with a higher incidence of psychosis.[7]

Limb buds start to develop in the fourth month of intrauterine life. Development of the limbs occurs over approximately 4 weeks. Abnormalities in the normal signals involved in limb development may result in polydactyly. Abnormal signals may be induced by inherited traits or drugs. The fetal

hydantoin syndrome commonly is associated with distal phalangeal hypoplasia; polydactyly also has been reported as a component of this syndrome.[8] All-*trans*-retinoic acid can induce polydactyly in mice.[9]

Preaxial polydactyly is defined as duplication of the digits on the radial aspect of the hand (ie, duplication of the thumb, triphalangeal thumb, or index finger). Postaxial polydactyly involves the ulnar aspect of the hand and is the most common type, especially type B (rudimentary digit). Type A is characterized by completely formed digits.

Postaxial polydactyly may be associated with Dandy-Walker syndrome, which is inherited in an autosomal recessive fashion.[10,11] Components of the McKusick-Kaufman syndrome, postaxial polydactyly, vaginal atresia with resulting abdominal mass, hydronephrosis, and congenital heart disease.[12] Bardet-Biedl syndrome is characterized by postaxial polydactyly, retinal dystrophy, retinitis pigmentosa, obesity, neuropathy, and mental disturbance. Both McKusick-Kaufman and Bardet-Biedl syndromes show some clinical overlap; both are inherited as autosomal recessive disorders.[12] Postaxial polydactyly, absent pituitary, hypoplasia of the cerebellar vermis, and partial ophthalmoplegia have been reported as an inherited trait, possibly related to orofaciodigital syndrome type VI.[13] Hypoplastic nails and postaxial polydactyly may be associated with hypothalamic dysfunction, inherited as an autosomal dominant trait.[14] Postaxial polydactyly also has been reported in trisomy 2p syndrome.[15]

A gene for preaxial polydactyly has been mapped to chromosome 7q36.[16] Preaxial polydactyly can be associated with radial and tibial dysplasia. Townes-Brocks syndrome, characterized by preaxial polydactyly, external ear abnormalities, hearing loss, imperforate anus, renal malformations, and autosomal dominant inheritance, has been localized to chromosome 16q12.1.[17] Preaxial polydactyly also may be associated with the nevus comedonicus syndrome, which also may include skeletal defects, cerebral abnormalities, and cataracts.[18]

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