An Inherited Deformity

Alexander K. C. Leung, MBBS, FRCP, FRCP(UK&Irel), FRCPCH

A 10-month-old girl presents with a deformity in her right foot. The deformity has been noted since birth. Her 24-year-old mother has a similar deformity in both feet.

What is your diagnosis?

This girl has postaxial polydactyly. Polydactyly refers to a congenital abnormality characterized by the presence of an extra digit. The condition is classified as preaxial if the great toe is duplicated, postaxial if the fifth toe is duplicated, and central if the second, third, or fourth toe is affected.

Approximately 50% of cases are bilateral and two-thirds of the bilateral cases are symmetrical.

Polydactyly can be an isolated finding or might be present as part of a generalized disorder. When present as an isolated finding, polydactyly is more common in Asian and Black individuals than Caucasian individuals. Approximately 50% of cases are bilateral and two-thirds of the bilateral cases are symmetrical. The condition is slightly more common in girls than boys. Polydactyly may be inherited as an autosomal dominant trait with variable penetrance. Polydactyly can be associated with Trisomy 13, Carpenter syndrome, Ellis-van Creveld syndrome, Meckel-Gruber syndrome, Rubinstein-Taybi syndrome, Bardet-Biedl syndrome, Grebe syndrome, Pallister-Hall syndrome, Townes-Brocks syndrome, Greig cephalopolysyndactyly syndrome and Orofaciodigital syndrome.

Untreated polydactyly may cause problems with shoe-fitting and angular deformity of the toe in addition to cosmetic concerns. Amputation of the extra digit is often necessary.

Dr. Leung is a Clinical Associate Professor of Pediatrics, University of Calgary, Calgary, Alberta.