A 4-YEAR-OLD girl, the offspring of first cousins, was referred for evaluation of unusual appearance. She had a history of frequent upper respiratory tract infections. She was of normal intelligence. Her height, weight, and head circumference were all at the 50th percentile for age. The midface was hypoplastic, with depression of the nasal bridge. The nose was small and flat (Figure 1). The distal phalanges of the thumbs and fingers were short and broad. A grade 2-3/6 systolic murmur was heard over the entire thorax and parascapular areas.

Serum calcium, phosphorous, and alkaline phosphatase levels were normal. A chest x-ray film showed striking calcification of the tracheobronchial tree (Figure 2). Stippled epiphyses were present in the shoulders (Figure 3) and knees on skeletal survey. The distal phalanges appeared short on x-ray films of the hands (Figure 4). Results of an audiogram were normal. Echocardiographic examination revealed mild peripheral pulmonary stenosis. The chromosomal analysis revealed a karyotype of 46, XX.

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Denouement and Discussion

Keutel Syndrome

Figure 1. Midface hypoplasia is present with a depressed nasal bridge and small nose.

Figure 2. Striking calcification of the tracheobronchial tree is present on the chest x-ray film.

Figure 3. An x-ray film of the left shoulder demonstrates stippling of the epiphysis.

Figure 4. The distal phalanges of the hands are short and broad.

Keutel et al described a distinct malformation syndrome in 2 siblings who had brachytelephalangism (short, broad distal phalanges), abnormal calcification of cartilages, and multiple peripheral pulmonary stenoses. At least 11 additional cases of this unique syndrome have been reported. The most consistent findings in this disorder are the unusual and diffuse calcification of cartilage and brachytelephalangism, which have been described in all patients. The striking calcifications involve the nose, ears, larynx, trachea, bronchial rings, and costochondral junctions, and at times they result in an outline of these structures. Epiphyseal stippling may occur in the long bones of infants and young children, and cartilaginous calcifications may be found in the vertebrae. The distal phalanges are short and broad, often with a drumstick appearance. The nails may also be short.

Other characteristic features, with slightly more variable occurrence, include midface hypoplasia, peripheral pulmonary artery or pulmonary valve stenosis, hearing loss, and recurrent respiratory infections. The face is long with a depressed nasal bridge and small nose. Cardiac abnormalities have been described in 9 of the 13 patients with Keutel syndrome. The most common cardiac abnormality is peripheral pulmonary artery stenosis. Hearing loss, also commonly present, is usually mixed conductive and sensorineural in origin. The frequent respiratory tract infections may contribute to the conductive hearing loss. Mild mental retardation or borderline intelligence was present in 9 of the 13 previously described patients.

Parental consanguinity is common in Keutel syndrome. Seven of the 9 families who had children with this disorder, including this one, were consanguinous. Inheritance seems to be autosomal recessive.

The striking cartilaginous calcifications are a major feature of Keutel syndrome. Tracheobronchial calcification is rare in children, with only 34 cases reported as of 1992. The disorder most often confused with Keutel syndrome, because of the calcifications, stippled epiphyses, and midface hypoplasia, is Conradi-Hanermann syndrome. Children with this syndrome usually have asymmetric limb lengths and scoliosis. Other children who have been described with tracheobronchial calcifications include a few with associated congenital cardiovascular abnormalities, idiopathic hypercalcemia (Williams syndrome), and children treated with warfarin sodium following mitral valve replacement. Soft tissue calcification in absence of disturbances in calcium metabolism may be found in Noonan syndrome, pseudohypoparathyroidism, and diastrophic dwarfism, all of which can be separated from Keutel syndrome on the basis of physical examination.

Keutel syndrome is rare; however, the presence of tracheobronchial calcifications and stippled epiphyses on radiographic examination, in combination with short, broad distal phalanges and midface hypoplasia, should suggest the possibility of this disorder.

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REFERENCES


The Editors welcome contributions to Pathological Case of the Month, Picture of the Month, and Radiological Case of the Month. Those who wish to contribute should send their manuscripts to Dr Gilbert-Barness (Pathological Case of the Month), Department of Pathology, Tampa General Hospital, University of South Florida, Davis Island, Tampa, FL 33606; Dr Tunnessen (Picture of the Month), American Board of Pediatrics, 111 Silver Cedar Ct, Chapel Hill, NC 27514-1651; or Dr Wood (Radiological Case of the Month), Department of Radiology, Childrens Hospital Los Angeles, 4650 Sunset Blvd, Los Angeles, CA 90027. Articles and photographs accepted for publication will bear the contributor’s name. There is no charge for reproduction and printing of color illustrations.