"What's wrong with my baby's ear?"

Jerzy K. Pawlak, MD, MSc, PhD; and T. J. Kroczak, BSc

Mark, almost two-years-old, was born with a grossly abnormal left external ear, absent of external auditory meatus (Figures 1 and 2), a minor deformity of the right external ear, along with an opening in the roof of his mouth.

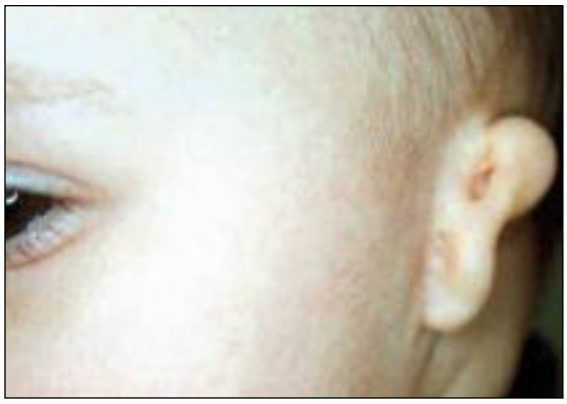

Figure 1. Abnormal left external ear.

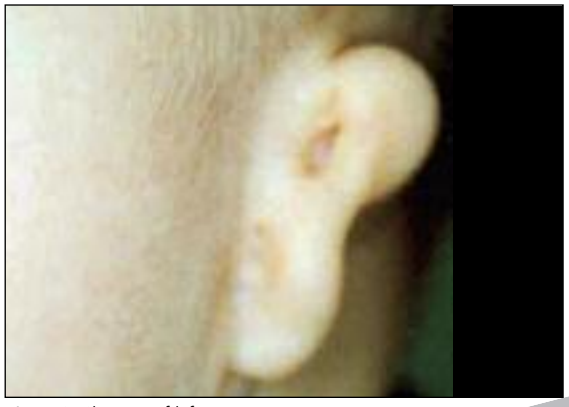

Figure 2. Close up of left ear.

Medical history

Mark's medical history is unremarkable:

- Both his parents are young and healthy
- He is the firstborn of a normal delivery
- His mother was healthy during her pregnancy
- His family history is unremarkable

Physical examination

A physical examination reveals nothing significant, except those facial abnormalities described above.

What's your diagnosis?

a) Franceschetti syndrome (complete)
b) Treacher Collins syndrome (incomplete)

This condition is characterized by hypoplasia of the facial bones, especially the zygoma and the mandible.

Dr. Pawlak is a General Practitioner, Winnipeg, Manitoba.

Mr. Kroczak is a Second Year Medical Student, University of Manitoba, Winnipeg, Manitoba.

Answer: B

Treacher Collins syndrome (incomplete)

Treacher Collins syndrome (a form of mandibulofacial dysostosis or Franceschetti's syndrome) is a highly complex disease process.

Etiology

The basic etiology of Treacher Collins syndrome is unknown, but it is generally thought to be inherited as an autosomal dominant-pattern trait with variable penetrance. One known cause of this syndrome is a mutation in the TCOF1 gene, at chromosome 5-q32-q33.1. The protein coded by this gene is called treacle and has been hypothesized to assist in protein sorting during particular stages of embryonic development, particularly that of the structures of the head and face.

Treacher Collins syndrome is generally thought to be inherited as an autosomal dominant-pattern trait with variable penetrance.

Characteristics

Treacher Collins syndrome is characterized by hypoplasia of the facial bones, especially the zygoma and the mandible. Facial clefting causes this hypoplastic appearance, with possible deformities or deficiencies of the:

- ear,
- orbital region,
- midface and
- lower jaw regions.

The clinical appearance is a result of the zygoma (malar bone) failing to fuse with the maxilla, frontal and temporal bones.

Degrees of involvement

Highly variant degrees of involvement (complete, incomplete and abortive forms) can be seen, but common facial features may include:

- Hypoplastic cheeks, zygomatic arches and mandible
- Microtia with possible hearing loss
- High arched or cleft palate
- Macrostomia (*i.e.*, abnormally large mouth)
- Slanting eyes
- Colobomas
- Increased anterior facial height
- Malocclusion (*i.e.*, anterior open bite)
- Small oral cavity and airway with a normal-sized tongue
- Pointed nasal prominence

Treatment

Treatment consists of testing for and treating any hearing loss so that a child can perform at a normal level in school (individuals with Treacher Collins syndrome usually have normal levels of intelligence). Plastic surgery can address the receding chin and other defects. Dx