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A 31-year-old white man presented with a 1×2 -mm asymptomatic pit anterior to the left helix.

What is your diagnosis?

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The Diagnosis: Preauricular Pit



Preauricular pits (also called sinuses) are a relatively common finding, occurring in 1% to 10% of the population, depending on race.1 They are inherited as an autosomal dominant trait with incomplete penetrance and variable expression and are commonly associated with commissural pits of the lip. The auricle begins to develop in the fourth week of gestation. Preauricular pits form because of failure of auditory tubercles to fuse with the first and second branchial arches surrounding the first branchial cleft, specifically the cartilages of Meckel.² Ectodermal tissue is entrapped in the mesodermal layer of the first branchial arch of the external ear. Sinuses may be uni- or multilocular and frequently end in a cystic structure deep along the external pinna. All preauricular

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pits are external to the temporalis fascia and never involve the tympanic membrane or external auditory meatus.³

Preauricular pits have been linked to internal malformations, the most significant of which is an association with renal anomalies. By performing renal ultrasonography on otherwise healthy and asymptomatic children with preauricular pits, investigators in Canada found that 4.3% of the children examined exhibited some type of renal anomaly, including a single hypoplastic kidney, small bilateral kidneys, and hydronephrosis secondary to vesicoureteric reflux. In the general population, the prevalence of renal anomalies is only 1%.4

Branchio-oto-renal syndrome (also known as ear pit-deafness syndrome) was first described by Melnick et al⁵ in 1976. This syndrome is transmitted in an autosomal dominant fashion and combines preauricular sinus, branchial anomalies, and renal dysplasia. In addition to preauricular pits, the branchial anomalies may present as cervical

sinus tracts. Another syndrome linking preauricular pits, commissural lip pits, and congenital conductive/mixed deafness has been reported in the Netherlands.⁶ The authors studied an extended family and linked the presence of ear and lip pits with deafness.

Preauricular pits also have been associated with trisomy 22 mosaicism.⁷ In these cases, the affected individual exhibits multiple anomalies, such as microcephaly, prominent epicanthal folds, micrognathia, cleft palate, hypotonia, long slender fingers, and/or congenital heart defects. Expression of the anomalies is variable.

Controversy exists as to whether asymptomatic preauricular pits should be treated. Most will never cause symptoms, but a minority become infected or discharge caseous material.³ Although most authors agree that a symptomatic pit requires surgery, some argue that asymptomatic pits also should be excised.⁸ The optimal treatment strategy is controversial. Prasad et al⁹ described various surgical techniques, as well as how to define the sinus tract intraoperatively to prevent inadequate excision and recurrence. Some surgeons use a metal probe, some advocate the use of methylene blue to define the sinus tract, and others have used microscopes or 2.5-power loop magnification.

The presence of a preauricular sinus should alert clinicians to the possibility of associated internal anomalies, including both renal and auditory problems. Appropriate review of systems, family history, noninvasive screening tests such as ultrasound and audiometry, interview and examination of family

members, and genetic counseling should be offered to the patient if warranted. Surgical options for treatment should be explained carefully.

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