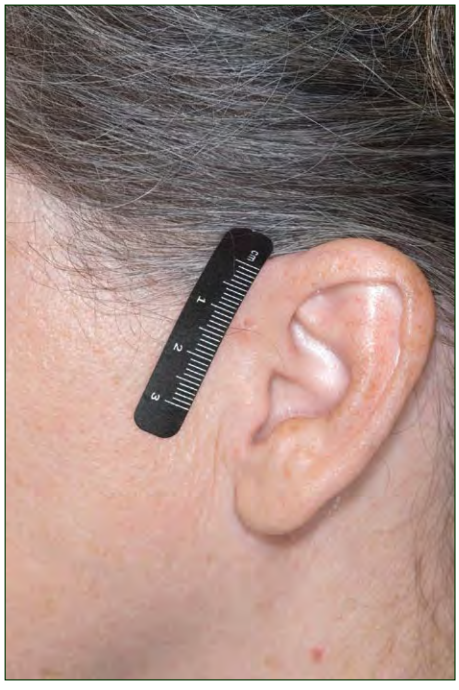
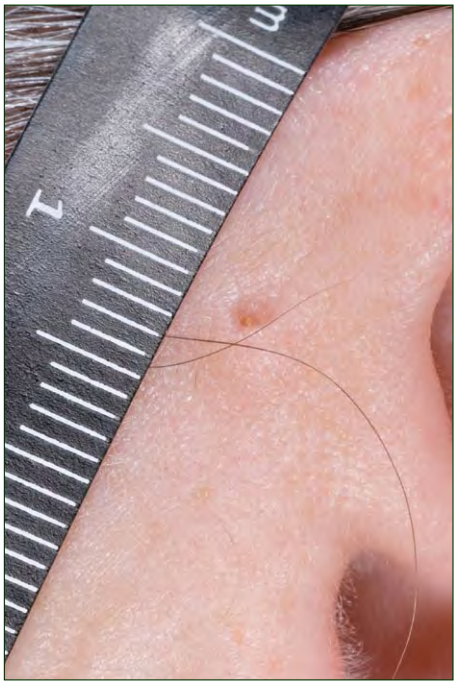
What Is Your Diagnosis?





A 49-year-old woman seen in clinic for a skin examination was noted to have 3-mm wide depressions in her preauricular region bilaterally. On review of systems, she denied problems with her hearing or vision and did not report other systemic concerns. She stated that the depressions sometimes produced an exudate of less then 1 mL that was white and without odor. The sites were not tender, erythematous, and no exudate was appreciated during the clinic visit. The patient reported that they never bothered her. She stated that she was born with these findings and they were present in everyone in her family. The patient also denied family history of congenital or later development of hearing or vision problems.

PLEASE TURN TO PAGE 278 FOR DISCUSSION

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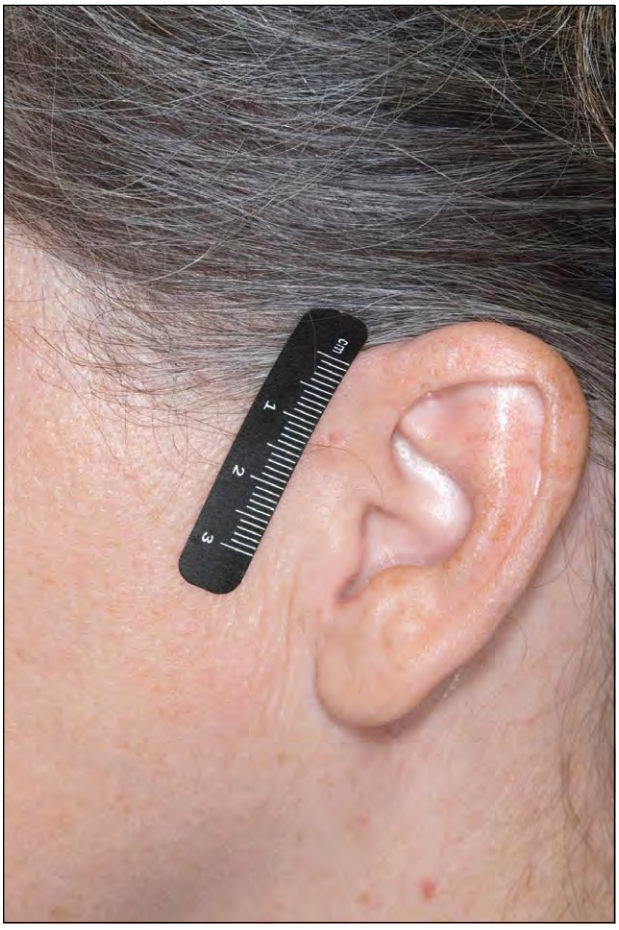
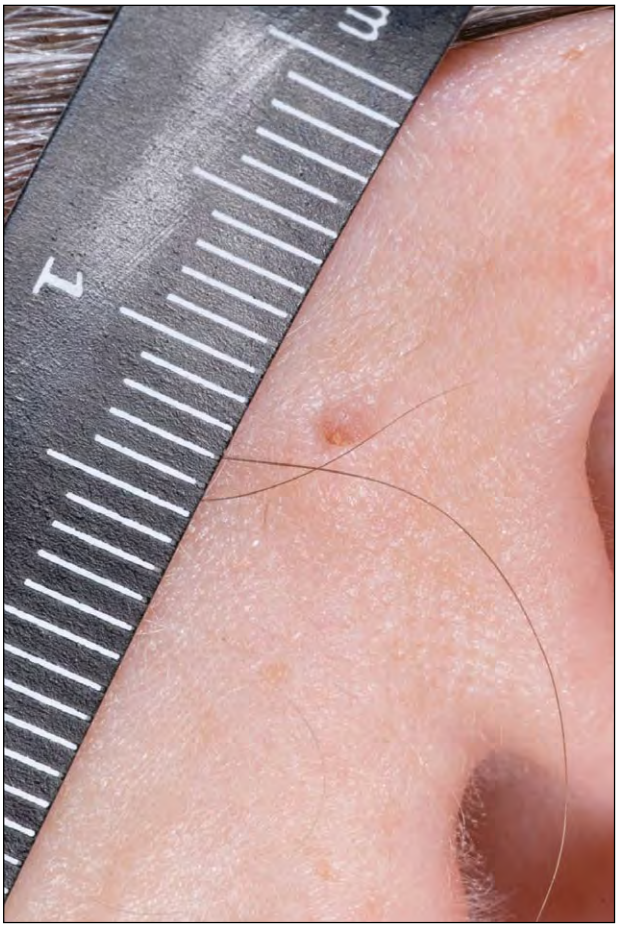
The authors report no conflict of interest.

The Diagnosis: Preauricular Pit





Preauricular pits, also known as preauricular sinuses, were first described by Heusinger in 1864.[1] This physical finding is the most common minor congenital abnormality of the ear and is thought to be a result of defective fusion of the 6 auricular hillocks during development.[1,2] They can occur either sporadically or in an inherited fashion and have an incidence of 0.1% to 0.9% in the United States.[3] The pits are small openings or dells in the skin, often located just anterior to the ascending helix.[4] Although they are more frequently seen on the right side, bilateral presentations are common in familial cases, as noted in our patient. Preauricular pits generally are isolated occurrences but have been linked with a number of congenital syndromes (Table).

Although biopsy is not required, histologic examination of the pit reveals stratified squamous epithelium in the lining of the duct with marked hyperkeratosis and parakeratosis. The duct can sometimes contain sebocytes or sebaceous glands, sweat glands, or hair follicles.[4]

Preauricular pits generally are asymptomatic but can become secondarily infected. Signs of infection include pain, surrounding erythema, and drainage. Staphylococcal species are the most common pathogens in infected pits. Others include *Proteus*, *Streptococcus*, and *Peptococcus* species.[4,5]

In most cases, preauricular pits have little significance apart from their cosmetic appearance. Their presence, however, warrants a thorough physical examination to look for any other craniofacial abnormalities or features of an associated congenital syndrome. The association with both congenital and noncongenital syndromes also emphasizes the importance of a detailed patient history and physical examination, practicing extra vigilance for findings associated with these syndromes and refreshing one's

Syndromes With Preauricular Pits

Syndrome[3]	Inheritance/Gene Defect	Sample Symptoms
Branchiootorenal syndrome	Autosomal dominant/*EYA1*	Structural defects of outer, middle, or inner ear; sensorineural, conductive, or mixed hearing impairment; lateral cervical fistulas, cysts, or sinuses; renal anomalies; nasolacrimal duct stenosis
Branchiootoureteral syndrome	Autosomal dominant	Bilateral sensorineural hearing impairment, duplication of ureters, bifid renal pelves
Branchiootic syndrome	Autosomal dominant/*SIX1*	Brachial anomalies, hearing loss, no renal abnormalities
Branchiootocostal syndrome	Autosomal recessive	Bilateral commissural lip pits, brachial fistula, rib anomalies
Tetralogy of fallot and clinodactyly	Autosomal dominant	Characteristic facies, tetralogy of fallot, clinodactyly of fifth finger
Steatocystoma multiplex	Autosomal dominant or sporadic/*KRT17*	Pilar cysts, facial steatocystoma
Rare syndrome of bilateral defects	Male to male transmission	Bilateral cervical sinus and brachial sinus, malformed auricles, bilateral hearing impairment
Deafness, preauricular sinus, external ear anomaly, and commissural lip pit syndrome	Autosomal dominant	Commissural lip pits, pinna dysplasia, mixed or conductive hearing impairment
Cat's-eye syndrome	Sporadic/*CECR2*	Iris colobomas, imperforate anus with possible fistula, down-slanting palpebral fissures, congenital heart abnormalities
Incomplete trisomy 22 mosaicism	46,XX/47,XX+22 (female)	Complex congenital heart defect, membranous anal atresia without fistula, distal limb hypoplasia, hypotonia and delayed development
Complete trisomy 22	Maternal origin of extra chromosome	Low-set ears, broad nasal bridge, macroglossia, cleft palate, micrognathia, hypoplastic genitalia

Abbreviations: *EYA1*, eyes absent homolog 1 (Drosophila); *SIX1*, sine oculitis–related homeobox 1 homolog (Drosophila); *KRT17*, keratin 17; *CECR2*, cat eye syndrome chromosome region, candidate 2.

knowledge of these syndromes. Appropriate testing should then follow. Although in the past the role of renal and hearing tests for isolated preauricular pits were controversial, routine testing is no longer recommended.[4,6,7] If a sinus becomes secondarily infected, surgical excision is an option to prevent future complications.

One of the more common syndromes associated with preauricular pits is branchiootorenal syndrome. This syndrome is caused by a defect in the eyes absent homolog 1 (Drosophila) gene, *EYA1*. It is inherited in an autosomal-dominant fashion and its features include preauricular pits; structural defects of the ear (outer, middle, or inner); and sensorineural, conductive, or mixed hearing impairment. Renal abnormalities can range from hypoplasia to complete absence of the kidneys. The patients also can have lateral cervical cysts, nasolacrimal duct stenosis, or fistulas.[3,4] Other notable syndromes are included in the Table.

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