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Signs in dysmorphology

Question mark ears and post-auricular tags

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Abstract

Questions mark ears are an easily recognisable but uncommon malformation of the external ear. They can be found as an isolated malformation and are a pathognomonic sign of the auriculo-condylar syndrome. An additional unique sign in this syndrome may be present in the form of post-auricular tags. Such malformations should prompt further investigation for other signs of the auriculo-condylar syndrome.

Keywords: Question mark ears; Post-auricular tags; Auriculo-condylar syndrome; Constricted ears; Congenital cleft auricle

In our clinical genetics clinic, we recently saw a four-week-old boy with question mark ears, post-auricular tags, micrognathia and a small mouth. This prompted us to search the literature for this particular combination. The term ‘‘question mark ears’’ proved to be a specific diagnostic search term.

The term ‘‘question mark ear’’ was first proposed by Cosman et al. as a major auricular malformation [1,2]. He described two unrelated patients with a striking ear anomaly that gave the external ear an appearance like a question mark. The most distinct feature of this ear anomaly is the congenital cleft or constriction of the lower auricle at the junction of the helix and the lobule [7], also known as congenital auricular cleft or constricted pinna [4]. The upper helix can be prominent. In some cases, the ear lobule is completely detached from the helix, in other cases there is only a mild constriction. The anomaly can be unilateral or bilateral and may be asymmetric (Figs. 1 and 2) [7]. Vincent et al. first described this distinct ear anomaly in 1961 in

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a young girl and her father [13]. Several other familial cases have been reported. One of these cases concerned a discordant ''apparently identical twin'' [11,2].

The term ''question mark ear'' is not listed as a search criterion in the Winter-Baraitser Dysmorphology Database [14], nor as a trait in the POSSUM database [8]. Inserting the term in the Online Mendelian Inheritance in Man (OMIM) database leads to only one hit: the auriculo-condylar syndrome (ACS).

The term ''post-auricular tags'' used in an OMIM search also only gives ACS as a diagnostic option. To our knowledge, post-auricular tags have not been described in other syndromes or as isolated findings, in contrast to the more common pre-auricular tags. Therefore, the post-auricular tags should be considered part of the primary question mark ear malformation. Post-auricular tags are located behind the pinna and are composed of skin and sometimes cartilage (Fig. 3). They should be distinguished from Quelprud's nodules, which are small post-auricular cartilaginous nodules arising from the posterior surface of the pinna present in nearly one-third of the population without other ear malformations [6].

The auriculo-condylar syndrome has been described several times in genetic and plastic surgery literature. It was first described by Uuspää in 1978 as a combination of bilateral external ear deformity and hypoplastic mandible in three related patients [12]. Jampol et al. reported a family in which several members had question mark ears, hypoplasia of the condyle of the mandible, abnormalities of the temporomandibular joint (TMJ) and microstomia [5]. Autosomal dominant transmission was suggested. There is marked intra-familial variability, with obligate carriers sometimes lacking external abnormalities. In 2005, Storm et al. reported 20 individuals with ACS [10]. A number of features were added to the phenotypic spectrum of the syndrome, including micrognathia, a round facial appearance with prominent cheeks, stenotic ear canals and hearing loss. Associated complications arising from the condylar and

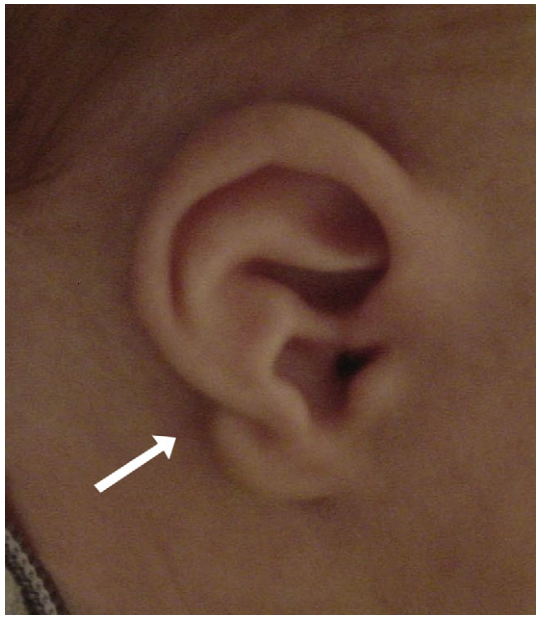

Fig. 1. Question mark ear (right ear) with congenital cleft (arrow) of the lower auricle in a four-week-old boy. Also note the prominent antihelix with a distorted course and interruption of the stem of the antihelix. The crus helix is prominent and fuses with the antihelix.

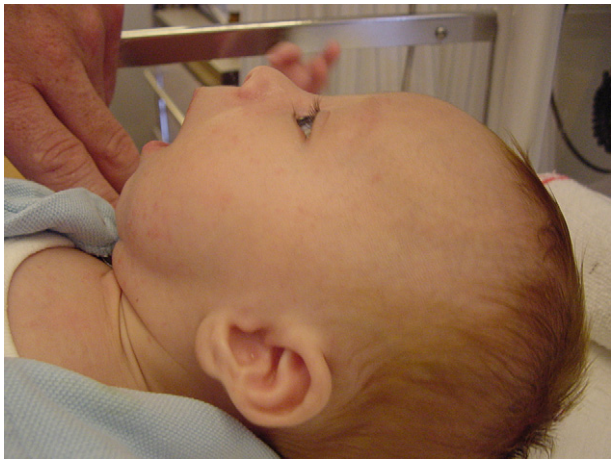

Fig. 2. A milder form of question mark ear on the left side and micrognathia in a four-week-old boy with auriculo-condylar syndrome.

oral abnormalities are glossoptosis, upper respiratory problems, apnoea and orthodontic problems [10]. Priolo et al. described a boy with several of the above mentioned clinical features of ACS, but also hypotonia and mild developmental delay [9]. A synonym for the syndrome used in the Winter-Baraitser Dysmorphology Database and POSSUM database is ‘‘prominent and constricted ears- malformed condyle of the mandibula’’ [8,14].

Formation. The auricle of the ear arises in the fifth week of gestation by the fusion of six hillocks that originate from the first and second branchial arches. The hillocks that will form the lower auricle arise from the second, or hyoid arch. The cleft in the lower auricle in a ‘‘question mark ear’’, just above the lobule, can be explained by an arrest of fusion of the fifth

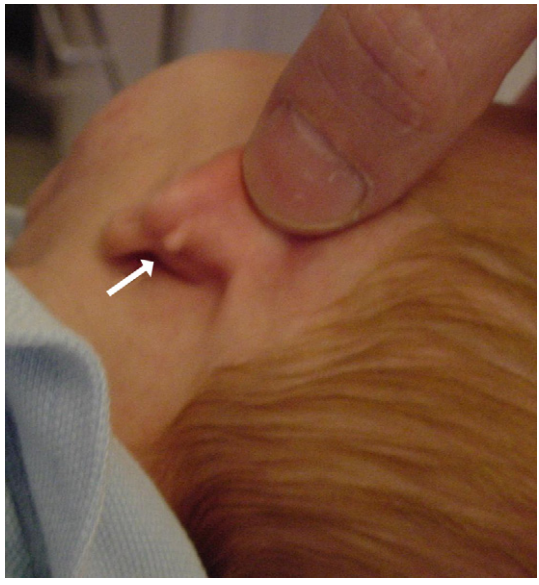

Fig. 3. Post-auricular ear tag (arrow) in a four-week-old boy.

(helical) and sixth (lobular) hillock [1,7]. The time of fusion arrest is thought to determine the severity of the cleft. The reason for the accompanying mandibular and condyle hypoplasia in ACS is unknown. Both structures arise from the first branchial arch.

Question mark ears have been described as an isolated, and sometimes unilateral finding, in several patients [1]. However, in these patients, no radiological studies were done to look for hypoplasia of the mandibular condyle. They might therefore represent a milder form of ACS without other external malformations.

Question mark ears have, to our knowledge, only been described once in a syndrome other than ACS. Erlich et al. reported a mother and daughter with question mark ears and micrognathia. They classified the patients as having the dysgnathia complex [3]. However, these two cases show a marked resemblance to ACS patients, and they may have been misclassified.

The differential diagnosis for ACS includes other syndromes caused by abnormal development of the first and second branchial arches, like Treacher-Collins syndrome, oculo-auriculovertebral syndrome, the dysgnathia complex, and isolated microtia. When necessary, the heart, kidneys and spine should be evaluated to differentiate ACS from other first- and second- branchial arch related disorders [10]. There is a high variability in the severity of the ear malformation, even within families. In extreme forms it can be difficult to recognize the malformed ear as a question mark ear. In these cases, a family history with more typical question mark ears could help to distinguish this type of ear malformation.

We believe that the ''question mark ear'' is an easily recognisable anomaly and an excellent diagnostic trait for the auriculo-condylar syndrome. Unfortunately, the term is not included as a search criterion in most dysmorphology databases.

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