Mandibulofacial dysostosis: CT findings of the temporal bones


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Received 30 May 1995; revision received 14 June 1995; accepted 4 July 1995

Abstract

Six patients of two families with clinically suspected and genetically proven Treacher Collins syndrome and hearing loss were studied by CT of the temporal bone. The objective of this study was to detect the abnormalities and to show the variation of expression of abnormalities. We found a high incidence of asymmetry in the different ear malformations and a slightly lower incidence of some other classical features, probably due to our patient selection.

Keywords: Mandibulofacial dysostosis; Treacher Collins syndrome; Computed tomography (CT), temporal bone

1. Introduction

Mandibulofacial dysostosis (MFD) is an autosomal dominant genetic disorder. The fully expressed phenotype exhibits characteristic dysmorphic features involving the face, eyes, mandible and ears [1,2]. MFD probably derives from inhibition of the facial structures from the first and second branchial (visceral) arches [3]. The gene for MFD is located on chromosome 5 [4].

We studied two families with an extreme variation in expression of MFD. Marres et al. published clinical, conventional radiological and genetic aspects of this group [5]. So far, only three studies report CT findings of the temporal bone in Treacher Collins patients [1,6,7]. The objective of this study was to detect the abnormalities and to show the variation of expression of abnormalities.

2. Material and methods

Three patients from two families were known to us with MFD. Additional family history revealed several other members with a 'look alike' appearance in both families. A genealogical study resulted in a family pedigree spanning five generations in one family and three generations in the other.

A clinical study was launched in both families and all 59 family members were asked for their cooperation. All patients underwent otorhinolaryngologic assessment; they were tested audiologically by pure-tone audiometry and when a conductive hearing loss was found, impedance audiometry was also performed. Seven patients had a combination of clinically suspected and genetically proven MFD, combined with hearing loss. Six patients underwent CT scanning, in another patient CT was impossible due to a severe kyphoscoliosis. CT scanning was done on a Siemens Somatom DR3. Scan parameters were 1 mm contiguous slices in the axial plane at 120 kVp, 520 mA and a high resolution algorithm. One patient underwent direct coronal scanning with the same parameters; in the other patients coronal images were made by multiplanar reconstruction. All scans were independently reviewed by two radiologists. The following anatomic areas were studied: the mastoid, the external auditory canal, the cavity and ossicles of the middle ear, the inner ear, the course of the facial nerve, and the presence of a bony defect and symmetry.

3. Results

CT was performed on six patients (12 ears). The findings are listed in Table 1. One patient with Treacher Collins syndrome showed no abnormalities on the temporal bone scan (Fig. 1a; case 5). One patient had no external...
Table 1
Findings on high resolution CT of 12 ears of the MFD patients with hearing loss

<table>
<thead>
<tr>
<th>Case Sex</th>
<th>Side</th>
<th>External meatus</th>
<th>Bone cleft</th>
<th>Mastoid pneumatization</th>
<th>Typanic cavity</th>
<th>Attic</th>
<th>Ossicular chain</th>
<th>Course of facial nerve</th>
<th>Asymmetry right/left</th>
<th>Remarks</th>
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<tbody>
<tr>
<td>1 F 49</td>
<td>Right</td>
<td>N</td>
<td>—</td>
<td>Absent</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>+</td>
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<td></td>
<td>Left</td>
<td>N</td>
<td>—</td>
<td>Diminished</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>+</td>
<td>Cholesteatomata</td>
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<tr>
<td>2 M 47</td>
<td>Right</td>
<td>N</td>
<td>—</td>
<td>Absent</td>
<td>Hypoplastic</td>
<td>Slit-like</td>
<td>Abnormal</td>
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<td></td>
<td>Left</td>
<td>N</td>
<td>—</td>
<td>Absent</td>
<td>Hypoplastic</td>
<td>Slit-like</td>
<td>Abnormal</td>
<td>N</td>
<td>+</td>
<td>+</td>
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<tr>
<td>3 M 20</td>
<td>Right</td>
<td>Absent</td>
<td>+</td>
<td>Absent</td>
<td>Hypoplastic</td>
<td>Slit-like</td>
<td>Abnormal</td>
<td>Abnormal</td>
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<td></td>
<td>Left</td>
<td>Absent</td>
<td>+</td>
<td>Absent</td>
<td>Hypoplastic</td>
<td>Slit-like</td>
<td>Abnormal</td>
<td>Abnormal</td>
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<tr>
<td>4 F 69</td>
<td>Right</td>
<td>Absent</td>
<td>+</td>
<td>Gross</td>
<td>Hypoplastic</td>
<td>Slit-like</td>
<td>Abnormal</td>
<td>Abnormal</td>
<td>+</td>
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<tr>
<td></td>
<td>Left</td>
<td>Steep</td>
<td>—</td>
<td>Absent</td>
<td>Hypoplastic</td>
<td>Slit-like</td>
<td>Abnormal</td>
<td>Abnormal</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>5 F 42</td>
<td>Right</td>
<td>N</td>
<td>—</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
<td>N</td>
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<td>—</td>
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<tr>
<td>6 M 19</td>
<td>Right</td>
<td>N</td>
<td>—</td>
<td>Diminished</td>
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<td>N</td>
<td>N</td>
<td>+</td>
<td>Cholesteatomata</td>
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auditory canal at all (Fig. 1b,c; case 3), one had an absent external canal on one side only, on the other side there was an abnormally shaped, steep external auditory canal (Fig. 2; case 4). No patient had inner ear deformities. The so-called slit attic, a narrow epitympanum (hypoplastic tympanic cavity) was found in seven of 12 ears (Fig. 1b,c). Asymmetry was found in four of six cases (Fig. 3a,b). Furthermore, we noted abnormalities in the ossicular chain in six of 12 ears (Fig. 2a), an abnormal course of the facial nerve in four of 12 ears (Fig. 2b), and abnormal mastoid pneumatization in nine of 12 ears (Fig. 1b).

In our series, two patients were operated because of hearing loss and a cholesteatomata was found in two cases.

4. Discussion

The diagnosis of MFD is, in the case of full expression of the syndrome, easily made on clinical appearance. The incidence is estimated as one in 40 000–70 000 live births [8,9].

MFD bears many eponyms of which Treacher Collins syndrome is one of the most frequently used MFD.

Fig. 1. (a) Normal findings on axial CT scan of case 5. Normal middle ear with normal position and configuration of ossicles (o) in the epitympanum (e). There is a normal pneumatization of the mastoid. Normal internal auditory canal (i), cochlea (c), vestibulum (v) and lateral semicircular canal (l). (b) Axial CT scan of the right ear of case 3 shows the slit attic (arrow), atresia of the external auditory canal and absence of mastoid pneumatization. (c) The coronal reconstruction image shows the hypoplastic cavum tympani (arrow) in the coronal plane, a bony cleft (curved arrow) and a lateral course of the descending segment of the facial nerve (open arrow).
The external appearance of the eye is a prominent nose above a redundant cheek with a prominent nose above a redundant cheek. The external appearance of the eye is a prominent nose above a redundant cheek.
ears are absent or malpositioned and hearing is impaired. Whenever only minor stigmata are present, diagnosis and additional genetic counselling become more precarious.

Radiographic findings in MFD were described on plain tomographic [12,13] and CT findings [1,6,7] (Table 2). Recently Pron et al. described CT imaging of the temporal bone with special attention for the external auditory canal and the ossicular chain in relation to audiologic findings [7] (Table 2). Jahrsdoerfer et al. [1] noted three radiographic findings that were unique for the MFD group: (1) absence of mastoid pneumatization, (2) ossicular disjunction and (3) a bony cleft in the lateral aspect of the temporal bone just anterior to the mastoid. This latter feature was found on 3-D reconstructed CT images. Our study revealed a bony cleft, as mentioned above, in 1/4 of the cases. Although the deformities of the ear and face tend to be symmetrical in MFD, in contrast to asymmetric changes in hemifacial microsomia [14], we found in accordance with other authors, considerable asymmetric changes [2,12]. We could not support the symmetry of the external ear and middle ear of Pron et al. Our two families showed asymmetric changes in four of the six cases. Perhaps in cases with a lesser degree of expression than in our patient group, more asymmetric changes are present.

The course of the facial nerve in the classical MFD is abnormal. The degree of aberration appears to depend more on the meatal atresia than on the attic atresia. The nerve follows a more direct path laterally from the geniculate ganglion. This means that the descending part may run more or less horizontally, and is therefore more anteriorly positioned than normal. We found this in four of the 12 ears.

This study of six patients (12 ears) showed the highest rate of normal external ears and normal configuration of the tympanic cavity compared with the other studies in Table 2. All studies show at least 50% of ossicular chain anomalies with an abnormal course of the facial nerve ranging from 33% to 100%. Reduction or absence of pneumatization is a rather constant finding, in 2/3 of cases.

Phelps et al. describe (Table 2) a characteristic slit attic usually containing hypoplastic ossicles. This slit attic could be identified extending in an oblique direction upwards and outwards. Mafee et al. [6] and Pron et al. [7] describe their findings as a hypoplastic epitympanum. We found a typical slit attic or hypoplastic epitympanum in 1/2 of the cases.

Abnormalities of the inner ear are rare and considered to be an incidental finding. The variation of abnormalities of the temporal bone of MFD patients is shown in case 5, with no abnormalities on the temporal bone CT imaging and in case 4, with different and asymmetric findings (Table 1) (Figs. 1, 2a,b,c).

References


