Auditory and Facial Nerve Dysfunction in Patients With Hemifacial Microsomia

Gerard J. Carvalho, MD; Caroline S. Song, MD, MPH; Karin Vargervik, DDS; Anil K. Lalwani, MD

Background: Hemifacial microsomia (HFM) is a common craniofacial disorder characterized by a wide spectrum of anomalies, including conductive hearing loss due to external and middle ear deformities. However, the prevalence of sensorineural hearing loss (SNHL) as well as facial nerve dysfunction is underappreciated.

Objective: To determine the frequency of auditory and facial nerve dysfunction and its relationship to more severe forms of bilateral HFM.

Design: Retrospective medical record review to characterize the clinical severity of HFM and the prevalence and nature of the associated auditory and facial nerve dysfunction.

Setting: Center for Craniofacial Anomalies at the University of California, San Francisco, Medical Center.

Patients: Ninety-nine pediatric patients with HFM evaluated at the University of California, San Francisco, Medical Center.

Main Outcome Measures: The prevalence of SNHL and facial nerve dysfunction in this patient population and any associations between these 2 characteristics.

Results: Hearing loss was present in 74 (75%) of 99 patients, with a conductive component in 73 patients. Sensorineural hearing loss was present in 11 patients (11%), with mixed hearing loss in most patients. Fourteen patients required rehabilitation with auditory amplification. Nearly a quarter of the patients (22 [22%] of 99) had facial nerve dysfunction, but only 1 patient had facial palsy on the same side as the SNHL. There was a statistically significant association between having auricular abnormalities and conductive hearing loss or SNHL (P = .30 and .80, respectively). However, there was no statistically significant association between bilateral HFM and the occurrence of either SNHL or facial paralysis, nor was there an association between auditory and facial nerve dysfunction.

Conclusions: Sensorineural hearing loss and facial nerve dysfunction are common in HFM. These findings have important implications in the treatment of patients with HFM.


Hemifacial microsomia (HFM) has been estimated to occur in 1 in 5600 live births, perhaps making it the most significant asymmetric craniofacial disorder.1-3 Hemifacial microsomia is also known by various other names, including Goldenhar syndrome, facoauriculovertebral sequence, oculoauriculovertebral dysplasia, and first and second branchial arch syndrome. Hemifacial microsomia is clinically heterogeneous, with a wide spectrum of anomalies, including ocular, auricular, mandibular, facial nerve, and soft-tissue abnormalities.4-9 External and middle ear anomalies associated with conductive hearing loss (CHL) are common since most patients have abnormal development of embryologic structures derived from the first and second brachial arches. Mild-to-severe ear findings have been reported, including flattened helical rim of the pinna, preauricular skin tags, microtia, external auditory canal atresia, osseous malformations, and anotia.8,10-12 Sensorineural hearing loss (SNHL) is thought to be uncommon in HFM and remains underappreciated.12 Failure to diagnose SNHL in children with HFM may have serious consequences, including significant sensory deprivation and consequently delayed speech and language acquisition.13-15 The aims of this study were to determine the prevalence of clinical features with particular reference to hearing loss, facial nerve dysfunction, and other facial anomalies in a case series of 99 patients with HFM and to explore possible associations between specific clinical characteristics.
RESULTS

PREVALENCE MEASURES OF HFM CHARACTERISTICS

The ages of patients with HFM ranged from 2 months to 18 years. There were 46 male patients (47%) and 53 female patients (53%). The right side was affected in 39 patients (39.4%), the left side was affected in 30 patients (30.3%), and 30 patients had bilateral disease (30.3%) (Figure 1). Fifteen patients had ocular abnormalities (15%), including coloboma and orbital asymmetry (Figure 2). Seventy-one patients had mandibular anomalies (71.2%), and 91 had auricular anomalies (92.0%), including microtia, ear tags, and ear pits (Figure 2).

Hearing loss was noted in 74 (75%) of the 99 patients with HFM (Figure 2). Conductive hearing loss was present in 73 (74%) of the 99 patients (74%); SNHL was present in only 11 (11.1% [95% confidence interval, 5.68-19.0]) of the 99 patients with HFM. The Table outlines the profile of patients with SNHL. Interestingly, in the majority of patients with SNHL, SNHL coexisted with CHL (10 [91.0%] of 11), leaving only 1 patient with SNHL alone. Sensorineural hearing loss tended to be a unilateral disease (8 [73%] of 11); only 3 patients had bilateral SNHL. Rehabilitation with auditory amplification for severe hearing loss was required in 18.9% (14 of 74) of patients.

Nearly a quarter of the patients with HFM (22 [22%] of 99) had facial nerve dysfunction; none of these cases were bilateral and only 1 patient had facial palsy on the same side as the SNHL (Figure 2). The severity and nature of facial nerve dysfunction (eg, facial nerve grading scale of House and Brackmann16) could not be determined from this retrospective record review.

TESTS OF ASSOCIATION

Statistical analysis using Fisher exact test revealed associations between certain characteristics among the study sample of 99 patients with HFM. Twice as many patients with auricular abnormalities had CHL compared with those without auricular abnormalities (P = .03). There was also a statistically significant association between the presence of auricular abnormalities and SNHL (P = .04). However, there was an absence of significant relationships when several other clinical characteristics were considered. There was no association between SNHL and facial nerve dysfunction (P = .45) or between auricular abnormalities and facial nerve dysfunction (P = .37). Furthermore, there was no association between having the
more severe form of bilateral (as opposed to unilateral) HFM and having either SNHL (P = .30) or facial nerve dysfunction (P = .80).

**COMMENT**

Hemifacial microsomia is a common craniofacial disorder that most otolaryngologists will encounter in their clinical practice. Of the many different names used to describe this disorder, HFM is probably the most widely used term, which unfortunately and erroneously conveys a unilateral disease process. While unilateral disease predominates, bilateral involvement in HFM is common and was seen in 30.3% of the patients in our series.\(^4\)\(^8\) The presence of bilateral disease suggests that the term HFM may be inaccurate or that patients with bilateral disease may in fact possess a different syndrome. The clinical phenotype in HFM is quite variable and can involve the face, eye, ear, mouth, central nervous system, skeletal system, and renal system.\(^1\)\(^4\)\(^5\)\(^8\)\(^9\) In our series, ear involvement was the most common, although the ear involvement was frequently present with malformations of the mandible and orbit. There was no clear sex preference or particular dominance of facial asymmetry to either the right or left side.

The severity of involvement of the ear can be highly variable.\(^5\)\(^11\)\(^17\)\(^18\) External ear abnormalities may be as mild as a flattened helical rim or as severe as complete absence of the auricle; stenosis or atresia of the external auditory canal can also be seen. Since the ossicles are also derived from the first and second branchial arches, abnormal development of the middle ear ossicles may be present in HFM. Consequently, CHL as a result of external and middle ear malformations is common. Not surprisingly, a positive association between auricular abnormalities and CHL was present in our series, with CHL being twice as common in children with ear malformations.

While clinicians are aware of the association between HFM and CHL, the incidence of SNHL is less appreciated. In our series, SNHL was present in 11% of the affected children (95% confidence interval, 5.68-19.0) and is much higher than the incidence of 0.001% to 0.004% of congenital SNHL or 3% to 4% seen in patients with craniofacial syndromes. Other investigators\(^5\)\(^9\) reported an incidence ranging from 6% to 14%. Our study was conducted in a craniofacial anomalies clinic that uses a multidisciplinary approach to these children’s health care; this setting should reduce the potential bias toward an increased prevalence of hearing loss that may be seen in a purely otologic setting. Interestingly, in the present series, SNHL was much more common in children with auricular abnormalities. Further studies are needed to confirm this finding and determine if there is a regional disturbance during development that may provide an explanation for the association between auricular malformations and SNHL.

It is clear from our work and that of others that the incidence of SNHL in HFM is significantly higher than expected. Unfortunately, there may be a significant delay in the diagnosis of SNHL. The diagnosis of HFM may not be made until late infancy or early childhood since recognizable features such as facial asymmetry may be subtle or inapparent in a newborn infant. Therefore, these children will not meet the criteria for the high-risk registry for hearing loss and most will not be screened for hearing loss at birth. We strongly recommend a complete audiologic evaluation of every child in whom the diagnosis of HFM is being considered.

Facial nerve dysfunction was present in 22% of patients in our study and reaffirms that facial nerve dysfunction is a common feature of HFM. This finding is similar to the 22% to 25% prevalence reported in several other studies.\(^8\)\(^19\)\(^20\) However, care should be taken to differentiate tree facial nerve dysfunction from facial muscle weakness that results from a primary deficiency of the mesoderm of the branchial arches.\(^4\)\(^8\) Given the retrospective nature of this study, it is difficult to differentiate between neuropathy and myopathy.

The occurrence of either SNHL or facial nerve dysfunction was not seen more frequently in patients with bilateral HFM. This finding is surprising because one would expect a higher incidence of regional embryologic disturbance in what can be considered a more severe phenotype of HFM. The low prevalence of coexisting SNHL and facial nerve dysfunction (1 patient of 99)

<table>
<thead>
<tr>
<th>Patient No.</th>
<th>Ear</th>
<th>Severity of Sensorineural Hearing Loss</th>
<th>Conductive Hearing Loss</th>
<th>Amplification</th>
<th>Facial Palsy</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Right</td>
<td>Profound</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>2</td>
<td>Right</td>
<td>Moderate to severe</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>3</td>
<td>Right</td>
<td>Moderate</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>4</td>
<td>Right</td>
<td>Moderate</td>
<td>Yes</td>
<td>No</td>
<td>Yes (left side)</td>
</tr>
<tr>
<td>5</td>
<td>Left</td>
<td>Mild</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>6</td>
<td>Left</td>
<td>Moderate to severe</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>7</td>
<td>Left</td>
<td>Mild to severe, sloping</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
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<tr>
<td>8</td>
<td>Left</td>
<td>Mild to moderate</td>
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<td>No</td>
<td>No</td>
</tr>
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<td>9</td>
<td>Bilateral</td>
<td>Right: severe</td>
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<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>10</td>
<td>Bilateral</td>
<td>Right: mild to moderate</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>11</td>
<td>Bilateral</td>
<td>Right: severe</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
</tr>
</tbody>
</table>

**Sensorineural Hearing Loss in Patients With Hemifacial Microsomia**
seen in our study differs from that of Bassila and Gold-
berg, in which 6 of the 50 patients had coexisting SNHL and some facial nerve dysfunction. It is possible that our results differ due to the random variation that exists among different study samples. Ultimately, we observed no statistically significant association between SNHL and facial nerve dysfunction. Further analytic studies that specifically focus on facial and auditory nerve dysfunction may help determine if there is a possible association between the seventh and eighth nerve dysfunction observed in patients with HFM.

CONCLUSIONS

Auricular abnormalities, CHL, SNHL, and facial nerve dysfunction are common in HFM. There should be a high index of suspicion for SNHL and all children diagnosed as having HFM should undergo complete audiologic evaluation. A significant number of children with HFM may require auditory amplification for severe hearing loss.

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Reprints: Anil K. Lalwani, MD, Laboratory of Molecular Otology, Department of Otolaryngology–Head and Neck Surgery, 533 Parnassus Ave, Room U490A, San Francisco, CA 94143-0526 (e-mail: lalwani@itsa.ucsf.edu).