Isolated Microtia as a Marker for Unsuspected Hemifacial Microsomia

Ivan J. Keogh, MD, FRCS; Maria J. Troulis, DDS, MSc; Angelo A. Monroy, MD; Roland D. Eavey, MD, SM; L. B. Kaban, MD, DMD

Objective: To determine the proportion of hemifacial microsomia (HFM) in patients with unilateral or bilateral “isolated” microtia.

Design: Prospective cohort clinical study.

Setting: University-affiliated, tertiary referral clinic for patients with microtia.

Patients: One hundred consecutive patients with isolated microtia.

Interventions: All the patients underwent a clinical examination and audiologic evaluation. The OMENS classification system was used to grade the severity of craniofacial features: orbital deformity, mandibular hypoplasia, ear deformity, nerve (cranial nerve VII) involvement, and soft-tissue deficiency. Each anatomical abnormality was graded from 0 (normal) to 3 (most severe) (score range, 0-15).

Main Outcome Measures: The OMENS scores, percentage of patients with isolated microtia and undiagnosed HFM, and isolated microtia as an early clinical marker for HFM.

Results: Forty patients (40%) with microtia were determined to have HFM (31 unilateral and 9 bilateral). Mean patient age was 9.2 years (range, 6 weeks to 41 years), with male predominance (27 males and 13 females). The OMENS scores were less than 5 in 24 patients and 6 to 10 in 16 patients. Thirty patients had cranial nerve deficits, and 37 had mandibular asymmetry. Thirty-seven patients demonstrated conductive hearing loss, and 1 had sensorineural hearing loss.

Conclusions: Isolated microtia served as an early clinical marker for asymmetrical facial growth in 40% of the patients. Isolated microtia and HFM could represent a spectrum of expression of the same developmental phenomenon.


EMIFACIAL MICROsomia (HFM) is a congenital facial deformity involving the structures of the first and second pharyngeal arches: maxilla, mandible, external and middle ear, facial and trigeminal nerves, muscles of mastication, and overlying soft tissue. After cleft lip and cleft palate, HFM is the most common craniofacial malformation.1,2 It varies in phenotypic severity along a spectrum from mild to severe. As the child develops and the face matures, the skeletal deformity progresses and becomes more obvious. If the deformity is bilateral or involves the orbit, the term craniofacial microsomia is sometimes synonymously used. Up to 20% of individuals with HFM have bilateral malformations (craniofacial microsomia) when assessed clinically.1,2 In patients with bilateral HFM, the deformity is always asymmetrical. In the presence of an epibulbar dermoid, vertebral anomalies, and abnormalities beyond the craniofacial region, the term expanded spectrum HFM is the current appropriate term.

Microtia and HFM have similarities such as variable phenotypic expression, asymmetrical nature of the defects, association with seventh nerve palsy, right side preponderance, and male predilection.3 Patients with HFM often have microtia as part of the clinical spectrum; however, patients with microtia do not always have HFM. The clinical expressions of these 2 malformations therefore overlap, yet the relationship remains unclear.2 The clinical visibility of microtia allows early diagnosis and counseling.3 However, HFM, unless severe, often is not clinically obvious to even the trained eye.

In early infancy, plump buccal fat pads conceal mild to moderate mandibular asymmetry. Jaw asymmetry becomes exaggerated during periods of active mandibular growth at 4 to 7 years of age and during the pubertal growth spurt. Early diagnosis allows for early intervention, which may modify growth and provide the patient with a better outcome using contemporary minimally invasive techniques.4 Surgical and nonsurgical ortho-
Pedic manipulations may stimulate growth on the affected side and reduce secondary deformity in adjacent skeletal structures. Late correction, at the stage of skeletal maturity, always necessitates a more major operative procedure, with associated morbidity. Early diagnosis also allows communication among the oral and maxillofacial surgeon, otolaryngologist, orthodontist, and pediatrician. During microtia reconstruction the ear needs to be positioned with the final position of the jaw in mind, so a coordinated approach is ideal. Furthermore, dental correction can be suboptimal if the underlying skeletal asymmetry is not recognized. Early diagnosis allows the physicians to provide better counseling and information to the family regarding prognosis and long-term management issues.

Bennun et al\(^2\) retrospectively examined the relationship between microtia and HFM using clinical photographs and patient records. In this prospective study, we examined a consecutive cohort of patients with a chief concern of obvious “isolated” microtia. The objectives were to evaluate for the coexistence of HFM and to use the OMENS classification system to grade severity.\(^1\)

### METHODS

Patients with the chief concern of microtia were prospectively examined at the Massachusetts Eye and Ear Infirmary. Some patients were examined more than once during follow-up; intervals for follow-up were selected for clinical reasons and not according to a regimented schedule. Patients with a known diagnosis of HFM, chromosome abnormalities, known exposure to teratogens, or a family history of craniofacial abnormalities were eliminated from the study.

The microtia and the degree of auricular deformity were determined as follows: (1) mild: an anatomical auricular difference beyond normal variation, yet most of the cartilaginous pinna, sulcus, and lobule were present, plus the external auditory canal (EAC) was present; (2) moderate: notable anomaly with diminished pinna size (usually one-half width and two-thirds height) with often absent tragus and conchal bowl, significantly diminished sulcus, and lobule usually present, plus the EAC was absent; and (3) severe: the classic peanut- or figure 8–shaped ear appendage without a definable pinna, with absent sulcus, and with rudimentary lobule-like fatty tissue region, plus the EAC was absent.

All the patients underwent an age-appropriate audiologic assessment. Pure-tone audiometry (range, 125-8000 Hz), which included air and bone conduction in decibels, was performed in the older patients. Pure-tone averages were calculated for each ear using decibel results at 0.5, 1.0, and 2.0 kHz. Younger patients were tested using behavioral methods or brainstem-evoked response audiometry. Hearing was defined as normal (0-20 dB), mild loss (21-40 dB), moderate loss (41-60 dB), severe loss (61-80 dB), or profound loss (>80 dB).

Patients were examined by otolaryngologists and by oral and maxillofacial surgeons, not only for auricular morphologic features but also systematically for concurrent facial skeletal deformities such as maxillary or mandibular asymmetry, canting of the occlusal plane, alar bases or gonial angles, malocclusion, maxillary and mandibular dental midline discrepancy, deviation of the chin point, facial nerve weakness, asymmetry in elevation of the palate, and soft-tissue deformities (Figure 1 and Figure 2). On the basis of clinical examination, patients were given an OMENS score of 0 (normal) to 15 (most severe) to describe the physical findings of orbital asymmetry, mandibular hypoplasia, ear deformity, nerve involvement, and soft-tissue deficiency. For example, O\(_1\), M\(_2\), E\(_1\), N\(_0\), S\(_1\)=OMENS score 5 (Table). Patients who had microtia plus any evidence of skeletal deformity with or without facial nerve involvement, asymmetrical elevation of the soft palate, or soft-tissue deficiency were classified as having HFM.

### RESULTS

**HFM AND MICROTIJA**

One hundred consecutive patients with isolated microtia were included in this study. Forty patients were de-
determined to have HFM using the previous criteria (31 unilateral and 9 bilateral). Twenty-seven patients were male and 13 were female. Mean patient age was 9.2 years (range, 6 weeks to 41 years) (1 adult was an age-atypical outlier for the study population). Of the 40 patients with microtia and HFM, 16 had right-sided, 15 had left-sided, and 9 had bilateral deformity. For patients with bilateral microtia, the most severe grade was used for classification. Thirty-two patients had severe microtia, 6 had moderate microtia, and 2 had a mild form of microtia.

HEARING TEST RESULTS

Normal hearing was detected in 2 patients with mild microtia. Unilateral mild conductive hearing loss (CHL) was determined for 17 patients and moderate CHL for 10 patients; the CHL occurred on the ipsilateral side as the microtia in all individuals. Bilateral CHL was detected in 10 patients. One patient exhibited bilateral sensorineural hearing loss, with a right ear pure-tone average of 80 dB and a left ear pure-tone average of 60 dB.

OMENS CLASSIFICATION

Overall OMENS scores for the 40 patients with HFM were as follows: 0-2 (n=0), 3 (n=2), 4 (n=9), 5 (n=13), 6 (n=7), 7 (n=5), 8 (n=2), and 9 (n=2).

Orbit

Five individuals demonstrated an orbital abnormality, and all were classified as grade 1 (inferiorly displaced orbit).

Mandible

Three patients exhibited no mandibular asymmetry and were classified as having HFM based on other concurrent findings (facial nerve deficit and asymmetrical elevation of the palate). Most patients (n=29) exhibited mild mandibular asymmetry (grade 1 mandible findings), and 8 individuals demonstrated grade 2 mandible findings (Figure 3). No patients had a grade 3 mandible deformity.

Ear

All 40 patients had microtia; 32 patients had a severe malformation, 6 demonstrated a moderate malformation, and 2 had a mild ear malformation.

Nerve

Of the 40 patients, 10 exhibited no evidence of facial nerve deficit, 22 had a grade 1 deficit (the frontalis or marginal mandibular branch alone), 5 had a grade 2 deficit (2 branches), and 3 had a grade 3 nerve deficit (all branches).
Of the 40 patients with HFM, 24 did not have a soft-tissue deficiency, and 14 had a mild deficiency. One patient each had a moderate or severe deficiency.

**Soft Tissue**

Of the 40 patients with HFM, 24 did not have a soft-tissue deficiency, and 14 had a mild deficiency. One patient each had a moderate or severe deficiency.

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**Table. OMENS Classification System**

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<th>Grade</th>
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| 0
| Normal orbital size and position |
| 1 | Abnormal orbital size |
| 2 | Abnormal orbital position; an additional arrow denoted the relative position of the affected side (ie, O1 for superior, O2 for inferior) |
| 3 | Abnormal orbital size and position |
| M0 | Normal mandible |
| M1 | The mandible and glenoid fossa are small, with a short ramus |
| M2 | The mandibular ramus is short and abnormally shaped; subdivisions A and B are based on the relative positions of the condyle and the TMJ |
| M3 | Complete absence of the ramus, glenoid fossa, and TMJ |
| E0 | Normal ear |
| E1 | Mild hypoplasia and cupping with all structures present |
| E2 | Absence of external auditory canal with variable hypoplasia of the concha |
| E3 | Malpositioned lobule with absent auricle; lobular remnant usually inferiorly and inferiorly displaced |
| F0 | No facial nerve involvement |
| F1 | Upper facial nerve involvement (temporal and zygomatic branches) |
| F2 | Lower facial nerve involvement (buccal, mandibular, and cervical branches) |
| F3 | All branches of the facial nerve affected |
| S0 | No obvious soft-tissue or muscle deficiency |
| S1 | Minimal subcutaneous/muscle deficiency |
| S2 | Moderate, between the 2 extremes, S1 and S3 |
| S3 | Severe soft-tissue deficiency due to subcutaneous and muscular hypoplasia |

**Abbreviation:** TMJ, temporomandibular joint.

**OMENS-plus** is used to include the expanded spectrum: cardiac, skeletal, pulmonary, renal, gastrointestinal, and limb anomalies.

The midsagittal plane was defined by drawing a vertical line from the crista galli to the anterior nasal spine. These points were chosen based on ease of identification and a high degree of correlation with physical findings. A horizontal line was then drawn at a right angle to the midsagittal plane tangent to the supraorbital rims.

- The mandible was evaluated radiographically.
- The external ear anomalies were categorized based on clinical examination and standardized photographs.
- Facial nerve weakness was categorized on the basis of clinical examination. Other involved nerves were also analyzed, such as the trigeminal N5 (sensory) and hypoglossal N12; the remaining cranial nerves were signified by the appropriate number in superscript.
- Grading soft-tissue deficiency was based only on subcutaneous and apparent muscular deficiency and did not include associated anomalies, such as cranial nerve involvement, macrostomia, or auricular anomalies.

**Concurrent Anomalies**

A cleft lip/palate was seen in 1 patient, a bifid uvula in 2, a skull deformity in 1, and a vertebral deformity in 1.

The results of this study confirm the hypothesis that a significant subgroup (40%) of patients with microtia and clinically isolated ear malformation exhibited underappreciated skeletal and soft-tissue findings consistent with a diagnosis of HFM. Therefore, by using microtia as a clinical marker, the physician can earlier identify patients with HFM, even if the HFM in some patients becomes apparent later in childhood. The HFM can be difficult to recognize in infancy because prominent buccal fat pads tend to mask underlying mandibular asymmetry and because infants have no teeth to assist with examination of jaw position. However, mandibular deformity can become progressively more visible with age owing to asymmetrical skeletal growth plus concomitant thinning of the buccal fat pads. In this series, some patients were diagnosed only after the facial structures had matured from infancy to early childhood.

This study further extends evidence that a relationship exists between ear and jaw defects, first recognized by Thompson (1845) and Virchow (1864). Microtia occurs less commonly than HFM (1:5600),1,2 in approximately 1 patient per 10,000 live births, although microtia is more frequent in specific populations, such as the Navajo and Ecuadorians.6-8

The pathogenesis of microtia, and of HFM, in humans is incompletely understood. Poswillo9 described an animal phenocopy resulting from embryonic hemorrhages from a defective stapedial artery in the region of the first and second branchial arches. Johnston and Bronsky10,11 proposed the hypothesis that a deficiency in neural crest cell migration to the region of the first and second pharyngeal arches explains the pathogenesis of HFM and microtia. More recently (2002), a report of a transgenic mutation of a locus termed Hfm (B1 to B3 on chromosome 10) in a mouse model seems to provide insight into the pathogenesis of HFM.12 In particular, the model supports the hypothesis that at least some of the anomalies associated with HFM have a genetic basis mediated via mesenchymal disruptions and possibly embryonic hemorrhages. Another genetic study suggested a chromosomal locus termed far to be involved. Mice with the...
far mutation demonstrated hemimandibular hypoplasia. A single gene process that could interfere with cartilaginous mandibular development had been postulated previously based on embryonic experiments plus stochastic modeling. Human heritable conditions involve microtia and jaw structures, such as Treacher-Collins syndrome and branchio-oto-renal syndrome. A gene linkage for nonsyndromic microtia has also been discovered separately by one of us for a large kindred that was not part of the present study (R.D.E., unpublished data, 2006). Microtia also results from the teratogenic effects of medications such as thalidomide and high-dose retinoic acid.

In older children who exhibit microtia, a screening mandibular examination can be performed. The central incisors can be examined with the jaw closed to determine whether the teeth and spaces are aligned. In patients with HFM, the lower jaw is shifted to the side of the ear malformation, causing the teeth also to shift toward the microtia. The teeth can demonstrate a jaw cant if a tongue depressor or flat device is clenched between the upper and lower teeth; the depressor should be parallel with the floor in normal jaw anatomy but will be tilted up to the microtia side if the jaw is underdeveloped (Figure 2).

A jaw malformation will not exist in most patients with microtia and may be minor in other patients, so parents can be reassured. At other times, the pediatric dentist and orthodontist must be made aware of the malformation by an experienced oral surgeon because later orthodontic treatment can become frustrating if the underlying jaw anomaly resists standard alignment efforts. For some patients jaw surgery must be performed, and simpler distraction procedures can be timed in concert with facial growth and microtia reconstruction rather than allowing mandibular maturity in adolescence, with the need for more substantial jaw surgical efforts.

In conclusion, isolated microtia served as an early clinical marker for asymmetrical facial growth in 40% of patients. Parents of infants with isolated microtia should be made aware of the possibility of HFM, and a complete craniofacial examination by an oral and maxillofacial surgery team should be part of the patient evaluation. The OMENS classification system is a valuable tool for assessing these patients.

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Correspondence: Ivan J. Keogh, MD, FRCS, Academic Department of Otolaryngology, University College Hospital, Newcastle Road, Galway, Ireland (ivankeogh@oceanfree.net).

Author Contributions: Drs Keogh and Eavey had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. Study concept and design: Keogh, Troulis, Monroy, Eavey, and Kaban. Acquisition of data: Keogh, Troulis, Eavey, and Kaban. Analysis and interpretation of data: Keogh, Troulis, Eavey, and Kaban. Drafting of the manuscript: Keogh, Troulis, Monroy, and Eavey. Critical revision of the manuscript for important intellectual content: Keogh, Troulis, Eavey, and Kaban. Administrative, technical, and material support: Keogh, Eavey, and Kaban. Study supervision: Keogh, Troulis, Eavey, and Kaban.

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REFERENCES