

## **TITLE PAGE**

### **Healthcare and Psychosocial Experiences of Individuals with Craniofacial Microsomia:**

#### **Patient and Caregivers Perspectives**

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**Running title:** Healthcare and Psychosocial Experiences in Craniofacial Microsomia

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## **SUMMARY**

**Objective:** Craniofacial microsomia (CFM) is primarily characterized by underdevelopment of the ear and mandible, with several additional possible congenital anomalies. Despite the potential burden of care and impact of CFM on multiple domains of functioning, few studies have investigated patient and caregiver perspectives. The objective of this study was to explore the diagnostic, treatment-related, and early psychosocial experiences of families with CFM with the aim of optimizing future healthcare delivery.

**Methods:** Forty-two caregivers and nine adults with CFM responded to an online mixed-methods survey. Descriptive statistics and qualitative methods were used for the analysis.

**Results:** Survey respondents reported high rates of subspecialty evaluations, surgeries, and participation in therapies. Some participants reported receiving inaccurate or incomplete information about CFM and experienced confusion about etiology. Communication about CFM among family members included mostly positive messages. Self-awareness of facial differences began at a mean age of three years and teasing at mean age six, with 43% of individuals four years or older reporting teasing. Teasing often involved name-calling and frequent reactions were ignoring and negative emotional responses. Participants ranked “understanding diagnosis and treatment” as a top priority for future research and had the most questions about etiology and treatment guidance.

**Conclusions:** The survey results on the healthcare and psychosocial experiences from birth through adulthood of individuals with CFM reinforce the need for ongoing psychological assessment and intervention. Healthcare provision could be improved through establishing diagnostic criteria and standardized treatment guidelines, as well as continued investigation of CFM etiology.

**Keywords:** craniofacial microsomia, hemifacial microsomia, microtia, oculo-auriculo-vertebral spectrum, psychosocial experience, teasing

## INTRODUCTION

Craniofacial microsomia (CFM) is a congenital condition primarily characterized by underdevelopment of the ear (i.e. microtia) and mandible. Other facial features associated with CFM include lateral oral clefts, facial palsy, and eye anomalies. Additionally, CFM may be associated with upper airway obstruction, dysphagia, speech and hearing impairments, and anomalies of the spine, kidneys, heart and central nervous system.<sup>1</sup> CFM has an estimated U.S. birth prevalence of 1 in 3,500-5,600, leading to approximately 1,100 infants born in the U.S. with CFM annually.<sup>2</sup> Medical and surgical treatments can be complex and individuals with CFM require longitudinal evaluations by multiple specialty providers. Interventions often occur across key developmental phases in a patient's life from infancy through adulthood.

Despite the fact that CFM is the second most common congenital facial condition of patients treated by craniofacial teams, established diagnostic criteria for CFM do not exist. This is, in part, due to the wide range of phenotypic variability associated with the condition and the fact that the etiology is unknown for most patients. Children with CFM may be labeled as having hemifacial microsomia (HFM), oculo-auricular-vertebral spectrum (OAVS), or Goldenhar syndrome, among other terms. In addition, individuals with isolated microtia are considered to represent the mild spectrum of CFM.<sup>3</sup>

The diagnosis of a craniofacial condition can be a difficult experience for children and their caretakers, who together are likely to face challenges related to and beyond their medical care.<sup>4</sup> In previous studies, some parents have associated the diagnosis of a craniofacial condition with a negative impact on their emotional health and overall quality of life.<sup>5,6</sup> Parents of children with microtia have reported either a severe (35%) or moderate (46%) emotional family impact, feelings of shock, guilt, and depression.<sup>7</sup> Although some children's self-reports have not

identified differences from their peers, parental perceptions of children with CFM indicate lower physical, social, and school functioning, which may reflect their own stress and concern for their children.<sup>8,9</sup> Teachers have also rated children with CFM as having more behavioral problems at school, particularly social difficulties.<sup>10</sup>

Patients with craniofacial differences may experience appearance-related teasing and stigma.<sup>10</sup> Observers may react to person with a visible facial difference by staring, whispering, asking intrusive questions, or making inappropriate comments, which can induce embarrassment, aggression, emotional distress, and/or social withdrawal among those affected and their families.<sup>11-13</sup> In a sample of patients with Crouzon syndrome, 66% of patients reported being teased at some time, compared to 20% of those in a matched control group.<sup>14</sup> Among children with microtia, rates of teasing vary by age from 30% to 100% and teasing started at a mean age of four years, with some experiencing lower rates of psychosocial concerns after ear surgery.<sup>7,15,16</sup> Teasing by peers has been associated with a higher risk for depression, social difficulties, and aggression in children with microtia.<sup>7,17</sup>

Despite the potential burden of care and impact of CFM on multiple domains of functioning, little is known about its psychological effects and what patients and parents understand about the components of CFM, its etiology, and prognosis. Qualitative research provides an ideal means of exploring the experiences of caregivers and adults with an understudied diagnosis like CFM.<sup>18,19</sup> The purpose of the current study was to explore the diagnostic, treatment-related, and early psychosocial experiences of individuals with CFM and their caregivers using a mixed method approach.

## **METHODS**

## ***Participants***

Institutional Review Board approval was obtained for all study procedures at Seattle Children's Hospital. Individuals with CFM older than 18 years of age and adult caregivers of children with CFM were invited to participate via advocacy and family association websites. Invitation letters were also sent to families treated for CFM at Seattle Children's Hospital (SCH) and flyers were distributed in clinic at SCH and Children's Hospital Los Angeles (CHLA).

Inclusion criteria were: 1) diagnosis within CFM spectrum, including HFM, OAVS, microtia, and/or Goldenhar syndrome; 2) the presence of CFM-associated features: facial asymmetry, preauricular or facial skin tags, anotia or microtia, aural atresia, lateral oral clefts (i.e. macrostomia), and epibulbar dermoid; and 3) fluency in English. Sample images were provided to exemplify each of the craniofacial features. Reported phenotypic features, birth history, and healthcare history were reviewed to confirm eligibility.

## ***Survey***

Data was collected between June 2016 and April 2017 using an online, one-time, self-report anonymous survey in the REDCap platform.<sup>20</sup> The survey included: demographic characteristics, CFM phenotypic information, and use of health services, beliefs and communication about CFM, and perceptions of teasing. Participants ranked issues in importance relating to CFM and future research. Items included both fixed-response options and free text.

## ***Data analysis***

Descriptive statistics were used for demographics, phenotype, health history, and life experience categorical variables. We completed analyses in Stata version 12 (StataCorp, 2010).

Two authors trained in qualitative methodology grouped responses to each of the 13 open-ended questions into themes in an iterative process.<sup>21</sup> Each author individually coded responses, allowing for multiple themes to be identified from each response based on content.<sup>22</sup> This approach captured both the depth and the breadth of the responses and is “a data- rather than theory-driven process, enabling the researcher to describe and summarize the data in its entirety.”<sup>23</sup> The thematic groupings were then compared and initial coding had an average agreement of 92% (range 82% - 100%). Both authors then reconciled the thematic groupings until agreement was reached. Finally, frequency counts were calculated and illustrative quotes selected for each theme.

## **RESULTS**

### ***Participant characteristics***

Of the 114 participants who opened the study link, 53 (46%) completed the survey. Demographic and CFM features provided by 25 of the 61 who did not complete the survey were similar to those who finished the survey, except that partial respondents reported fewer major concerns at birth and were all primarily English speakers. Responses from two participants self-identifying as having Treacher-Collins syndrome were excluded. The remaining 42 caregivers and nine adults with CFM completed the survey in an average of 38 minutes. Most caregivers were mothers (90%) with male children (71%) who had a mean age of 7 years (SD 4, range 0-17). Most adults with CFM were female (78%) and the mean age was 45 years (SD 16, range 24-76). Most respondents were white (80%), non-Hispanic (89%), living in the United States (82%), had a college degree (80%), had private health insurance (80%), and spoke English (86%; Table 1). Most individuals with CFM received the diagnosis at birth (74%). The most common

diagnosis was microtia (84%), with or without HFM, CFM, and/or Goldenhar syndrome. The most common facial features of CFM were: microtia (86%), aural atresia (78%), and facial asymmetry (75%; Table 2).

### ***Healthcare services***

The majority of individuals with CFM had been seen at a craniofacial clinic (77%). An average of 7.7 (SD 3.7) specialists were consulted, most often audiology (98%), otolaryngology (73%), plastic surgery (71%), and dentistry (67%; Table 2). A majority (80%) reported some degree of hearing impairment and all of them had used a hearing aid at some time. A total of 67% of individuals with CFM had received a form of therapy, most commonly speech and language therapy. Eighty percent of individuals with CFM had at least one surgery (mean 4 surgeries, range 1-30; Table 3).

### ***Initial communication about diagnosis***

Most caregivers learned about the diagnosis from a pediatrician (46%), delivering physician (27%), or delivery nurse (17%). Caregivers endorsed feelings of concern/anxiety (79%), surprise/shock (64%), sadness (64%), guilt (55%), and confusion (31%) in response to the diagnosis. Common themes in communication about the diagnosis at birth included specific diagnoses provided (38%), delayed or inaccurate diagnoses (27%), and little or no information at birth (24%). Some caregivers (11%) sought additional information sources and others (11%) did not recall what was said (Table 4). In contrast, adults with CFM recalled the first communication about the diagnosis was from a parent (67%), plastic surgeon (11%), orthodontist (11%), or independent research (11%, data not shown).



### ***Etiology Beliefs***

Caregivers and adults with CFM often reported being unsure about the cause of the diagnosis (31% and 38%, respectively; Table 5). Some respondents attributed it to random occurrence (caregivers: 15%, adults: 25%). Others suspected a genetic component (caregivers: 21%, adults: 25%), with 15% of caregivers noting a familial inheritance pattern. Several respondents attributed CFM to a circulation issue (caregivers: 18%, adults: 13%) or other medical issues during pregnancy (caregivers: 13%, adults: 25%). Some caregivers attributed causality to their own medication use (15%) or exposure to environmental toxins before or during pregnancy (13%). A few parents identified religious explanations (5%). Finally, one mother (3%) and one adult with CFM (13%) noted that no one is at fault or to blame for the diagnosis.

### ***Family Communication about CFM***

Caregivers communicated to their children about CFM in a variety of ways (Table 6), with frequent themes including positive language and reassurance (36%). Both caregivers and adults with CFM reported communication centered on ‘being born with the diagnosis’ (21% and 38%, respectively). Family members sometimes used labels for ear anomalies, such as “quiet ear” and “miracle ear” (12%), along with normalization (15%), religious themes (12%), advice (9%), and literary resources (6%). Communication also involved medical (9%), hearing (9%), and future treatment-related (18%) information. One caregiver and one adult reported discussing CFM in terms of an accident during pregnancy. Adults with CFM reported more negative

communication experiences, including no family discussion, significantly delayed information provided outside the family, label of birth defect, or punishment from God (each 13%).

### ***Self-awareness, Responses from Others, and Teasing***

Individuals with CFM first noticed their diagnosis at a mean age of 3.3 years (SD 1.4) as reported by caregivers and 3.2 (SD 3.6) years as reported by adults with CFM. Parents reported their children first perceived others may be looking at them differently at a mean age of 3.9 years (SD 1.9). Children first reported experiencing teasing at a mean age of 6.4 years (SD 2.0), as reported by caregivers and 6.3 years (SD 2.5) by self-report of adults. While caregivers reported teasing for 43% of children who were 4 years of age or older, all adults with CFM, but one, acknowledged teasing during their childhood. Peak ages of teasing were reported in early childhood (>5 years) and elementary school (6-10 years), with the mean age of 9.0 (SD 2.5) when they were most teased. Teasing occurred most often at school or daycare (92%) followed by community settings (39%). The person teasing was usually a classmate (75%) or a child in the community (67%), although adults in the community had also teased at times (25%, Table 7).

While some caregivers felt their children were too young to notice people staring (25%), other parents (10%) and adults (25%) reported that other people do not notice their diagnosis. Some parents who did observe staring noted their children did not care (13%) and adaptive coping responses were described by caregivers (26%) and adults (38%). Caregivers with CFM (13%) and adults (38%) described feeling shy and gave examples of negative emotional responses. While caregivers (6%) and an adult (13%) reported responding by ignoring, a caregiver (3%) and adults (25%) also described that noticing others' reactions reminded them of their CFM diagnosis. Some caregivers (6%) noted their children have variable responses based

on the situation. The remaining caregiver themes (3% each) described seeking parental assistance, parents offering advice, reduction in teasing after surgery and using long hairstyles to cover ears (Table 8).

When asked what is said when teased, most caregivers (56%) and adults (83%) reported name calling (Table 8). Both groups (caregivers: 22%, adults 17%) also noted questions about their CFM-related differences and nonverbal negative reactions of mimicking or running away. One mother reported that her child experienced social exclusion (11%) and another distinguished teasing about his appearance from teasing related to his social miscues related to hearing loss (11%). The most frequent reaction to teasing reported by caregivers (45%) and adults (67%) was ignoring the teasing. Negative emotional responses were also reported by caregivers (27%) and adults (33%). Some parents noted that the child sought adult assistance (27%), while others were not sure about the child's reactions to teasing (27%). Both adaptive coping (18%) and aggression (9%) was reported by caregivers and one adult described becoming withdrawn (16%).

### ***Priorities for Future Research***

The most important topic for future research identified by caregivers and adults with CFM was “understanding diagnosis and treatment.” Caregivers also ranked “hearing concerns” as highly important. Among adults with CFM, teasing, social concerns and communication about diagnosis by healthcare providers were the second and third most important concerns. When asked about which topic they wanted more information, etiology was ranked highly by caregivers (41%). Treatment guidance and prognosis was also important to caregivers and adults (15% and 50%, respectively). Some parents felt well informed (15%) and others had no questions (7%); however, a variety of other topics were identified, including recurrence rates

(11%) and clarifying CFM features present in the child (7%). Caregivers (7%) described wanting to know how to better help their children and understand their experience with CFM. Adults wanted timely and accurate information (33%) and caregivers described learning more as a child develops (4%). One caregiver expressed interest in normed data (4%) and another wanted to see progress in prenatal diagnoses (4%, Table 9).

## **DISCUSSION**

This study explored the healthcare and psychosocial experiences of caregivers and individuals with CFM, with the goal of addressing gaps in the literature and identifying opportunities to advance clinical care, research, and psychosocial support for this population. We used a mixed-methods design including both caregivers and adult patients drawn from an international online sample.

The survey results demonstrate the substantial lifetime health impact of CFM with high rates of subspecialty evaluations, surgeries, and participation in therapies. Despite frequent contact with healthcare providers, about a quarter of respondents reported receiving limited information or an incorrect diagnosis, which may have contributed to anxiety, sadness, shock, guilt, and confusion. This corresponds with respondents' ranking "understanding diagnosis and treatment" as a top priority for future research. Miscommunication may be exacerbated by the interchangeable use of multiple terms, such as CFM, hemifacial microsomia, Goldenhar syndrome, and OAVS, while some providers consider these diagnoses to be distinct entities. In fact, none of these diagnoses are associated with well-established and specific diagnostic criteria. In addition, there is a lack of consensus if microtia without other features should be included under the CFM diagnosis. The use of multiple labels might be a barrier for exchanging information between families, patients, and healthcare providers. The unknown etiology in the

majority of cases, with both genetic and non-genetic potential risk factors, could make this issue even more confusing. Stronger collaboration between scientific communities and patient organizations can help clarify anticipatory guidance related to all ages of CFM medical care management recommendations and treatment options.

Despite caregivers' reports of their own challenges, they described family communication about CFM as generally positive and reassuring. However, there were several negative messages and avoidance of family discussion reported by the adults with CFM. This may reflect a generational difference between the two groups, selection factors, or a positive response bias by caregivers. Most individuals with CFM first noticed their diagnosis at age three years. Teasing onset was usually at about age six with a peak around age 6-10 years. For children over age four, teasing was reported in 43%, which included name-calling and mimicking. All adults with CFM, but one, reported teasing during their childhood and the most common response was ignoring the teasing, with about a third of children and adults describing negative emotional responses. These data reinforce the need for early and ongoing assessment of psychosocial well-being in individuals with CFM and their families with interventions as indicated.

Study limitations include a lack of complete sample representativeness, since the majority of respondents were white, non-Hispanic, well-educated individuals with private health insurance. This is likely a function of internet literacy and time and interest to complete surveys. However, given the paucity of this type of CFM research, we believe this is a necessary first step in better understanding patients' and parents' experiences and we could expect that the issues are similar or more severe for individuals more likely to experience barriers to accessing health information and care. As an exploratory study, we limited participation to those older than 18 and fluent in

English. Another limitation was a relatively small sample size; however, we enrolled participants from a more geographically representative population from multiple clinics than the few prior studies that have used retrospective chart review or enrollment through clinics to identify participants. Another potential limitation was self-report of diagnosis. To address this, images were provided for each CFM feature and language was used from previous studies on CFM. Responses were also reviewed for consistency with reported features, clinical history, and treatments. Further research in other languages and countries will help to understand experiences in multiple cultures. Future research should also include children and adolescent perspectives to help improve their care.

### **Conclusions**

The detailed information provided by parents and adults illustrates the intricacy and challenges of their CFM healthcare and psychosocial experiences from the moment of diagnosis through adulthood. The need for ongoing psychological assessment and intervention for patients and families was highlighted in their responses. Families expressed strong interest in understanding CFM etiology, diagnostic criteria, and treatment guidelines. Stronger collaboration between scientific communities, families, and patient organizations can help explain expectations related to all ages of CFM medical care management and treatment.

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