In their own words: caregiver and patient perspectives on stressors, resources, and recommendations in craniofacial microsomia care

Abstract

This study describes stressors, resources, and recommendations related to craniofacial microsomia (CFM) care from the perspective of caregivers of children with CFM and adults with CFM in order to inform improved quality of healthcare delivery. A mixed method design was used with fixed-response and open-ended questions from an online survey in English. The survey included demographics, CFM phenotypic information, and items about CFM-related experiences across settings. Themes were identified by qualitative analysis of responses to open-ended questions. Respondents (N = 51) included caregivers (n = 42; 90% mothers) and adults with CFM (n = 9; 78% female), who had a mean age of 45±6 years. Most children were male (71%) with an average age of 7±4 years. Respondents were primarily white (80%), non-Hispanic (89%), from the United States (82%), had a college degree (80%), and had private health insurance (80%). Reflecting the high rate of microtia (84%) in the sample, themes centered on the impact of hearing difficulties across settings with related language concerns. Negative social experiences were frequently described and school needs outlined. Multiple medical stressors were identified and corresponding suggestions included: providers need to be better informed about CFM, treatment coordination among specialists, and preference for a family-centered approach with reassurance, empathy, and clear communication. Advice offered to others with CFM included positive coping strategies. Overall, caregivers' and patients' responses reflected the complexity of CFM treatment. Incorporating these perspectives into routine CFM care has the potential to reduce family distress while improving their healthcare.

Key words: craniofacial microsomia; family stressors; family resources; qualitative analysis

Introduction

Craniofacial microsomia (CFM) is a congenital condition associated with underdevelopment of the ear (i.e., microtia) and mandible and has an estimated U.S. birth prevalence of 1 in 3,500-5,600.[1] Other facial features associated with CFM include lateral oral clefts, facial palsy, and eye anomalies. Additionally, CFM may be associated with facial paresis, upper airway obstruction, dysphagia, speech and hearing impairments, extra-cranial anomalies, and learning disabilities.[2-6] Through adulthood, craniofacial surgeries are crucial components of the multidisciplinary care required for CFM.[7] The burden of treatment for CFM has not been well described; however, children with microtia experience teasing and are at higher risk for psychosocial concerns, while their parents have reported difficulties in communication with providers and in their own coping.[8-10] Similarly, studies of individuals with chronic medical demands have identified poor outcomes for well-being, burden on significant others, and ineffective resource use.[11]

Children with CFM and their caretakers also face challenges impacting their quality of life beyond their complex medical care.[12] While little is known about the impact of CFM on families' lives, some evidence suggests parental quality of life may be affected.[12] In regard to school, parents of children with CFM have reported experiencing higher levels of stress.[13] In addition, teachers have described children with CFM as having lower levels of social competence and peer acceptance compared to peers, which mirrors patterns of lower quality of life in school and social domains for children with hearing loss.[14,15] In the broader literature, a health condition including a visible difference has been shown to have a psychosocial impact on those affected and their families.[16] Unwanted attention in the form of staring, questions, comments, and teasing may result in individuals feeling "different" or excluded from their

peers.[17] Such negative social encounters are associated with social anxiety and social avoidance, as well as low self-esteem, poor body image, and depression.[18]

Some findings derived from parent reports have not indicated differences between children with CFM and unaffected peers.[14] Discrepancies have also been identified between child and parent reports of children's psychosocial adjustment.[12] This highlights the importance of gathering multiple perspectives and the complexity of investigating the impact of a condition such as CFM. Furthermore, the patient experience and burden of treatment is known to vary by age, type of intervention, and stage in medical and surgical management.[11] Qualitative research provides an effective means of exploring the multifaceted experiences of both caregivers and patients affected by the understudied diagnosis of CFM. Previous papers have identified a paucity of qualitative approaches in health research, particularly for craniofacial conditions. The question of adjustment to CFM is further suited to qualitative methods due to the relatively rare prevalence of the condition.[19]

The present study used a mixed methods approach to elicit feedback from caregivers of children with CFM, as well as adult patients with CFM. We describe the experiences of individuals with CFM and their caregivers across their home, school, community, and medical settings. In addition, we report on resources participants found helpful, their advice to others affected by CFM, and their suggestions for improving healthcare. The nuanced perspective of patients and their families reflects the growing recognition of the importance of including patient reported outcome measures and integrating their feedback in ongoing quality improvement in clinical practice.[20]

Materials and Methods

Institutional Review Board approval was obtained at Seattle Children's Research Institute. Invitations to participate for adults with CFM older than age 18 years and caregivers of children with CFM were posted on American advocacy and family association websites. Invitation letters were 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. Inclusion criteria were: 1) diagnosis within CFM spectrum, including hemifacial microsomia (HFM), oculo-auriculo-vertebral spectrum (OAVS), microtia, and/or Goldenhar syndrome; 2) CFM-associated features: facial asymmetry, preauricular or facial skin tags, anotia or microtia, aural atresia, lateral oral clefts, and epibulbar dermoid; and 3) fluency in English. Sample images were provided to exemplify craniofacial features. Eligibility was confirmed by two authors reviewing reported phenotypic features, birth history, and healthcare history.

Survey

Data were collected from June 2016 to April 2017 using an online self-report anonymous survey in the REDCap platform.[21] Participants reported demographic characteristics and CFM phenotypic information. Items in fixed response and free text formats asked about difficulties in the home, school, medical, and community settings, along with what they have found helpful and their advice.

Data analysis

Descriptive statistics were used for demographics and categorical variables with Stata version 12. Responses to the 15 open-ended questions were grouped by two authors trained in qualitative methodology in an iterative process into themes.[22] Authors individually coded based on content to capture both the depth and the breadth of the responses, rather than using a theory-

driven analysis.[22] Thematic groupings were compared and initial coding had an average agreement of 94% (range 81% - 100%). Authors then reconciled the thematic groupings together until agreement was reached. Illustrative quotes were selected for each theme and frequency counts calculated.

Results

Participant characteristics

The link was opened by 114 participants and 53 (46%) completed the full survey. Demographic and CFM features provided by 25 of the 61 who did not complete the survey were similar to the study sample, with the exceptions of being only English speakers and reporting fewer major concerns at birth. Two participants who self-identified with Treacher-Collins syndrome were excluded. The sample included 42 caregivers and nine adults with CFM (Table 1), who 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 = 6; range 24-76). Respondents were white (80%) and non-Hispanic (89%). Participants were primarily from the United States (82%), as well as Canada (6%), Central/South American (4%), Mexico, (2%), United Kingdom (2%), and other European countries (4%). Most had a college degree (80%), had private health insurance (80%), and spoke English as their primary language (86%). Individuals with CFM generally received the diagnosis at birth (74%) and the most common diagnosis was microtia (84%), with or without HFM, CFM, and/or Goldenhar syndrome. Common facial features of CFM were microtia (86%), aural atresia (78%), and facial asymmetry (75%).

Home and Community Settings

For those who identified difficulties in the home setting, both caregivers (34%) and adults (29%) reported hearing concerns (Table 2). Caregivers identified stressors in speech difficulties, treatment burden, and their own distress (11% each theme). Adults noted difficulties in social comparisons with others, a lack of understanding by others, and not discussing their diagnosis in the home (14% each theme). Correspondingly, social stigma was noted by caregivers (62%) and adults (57%) to be a primary concern in community settings (Table 3). Hearing concerns were also reported as a difficulty in the community by caregivers (21%). Caregivers noted explaining their child's diagnosis to others (17%) as challenging, as well as accommodating for participating in activities and sports (10%). Adults reported hiding their diagnosis in community settings (29%) and one adult described feeling motivated to disprove negative assumptions made by others based on their diagnosis.

When asked about sources of support, caregivers identified online support websites (71%), their spouse/partners (62%), family (57%), and medical providers (48%) as most helpful (Table 4). They also noted friends (41%) and communicating with others who have similar diagnoses (41%) as supportive. Interestingly, a third identified scientific research and articles as important for coping. Adults with CFM offered advice for children with similar diagnoses, including making informed medical decisions (50%), having self-confidence (50%), reframing their experience when they are teased (25%), and openly communicating about their diagnosis (13%). Caregivers had a dozen themes identified in their advice for new parents, with reassurance that their child is a typical child (34%) given most frequently. They described positive reframing (26%), seeking CFM information and expert care (26%), acceptance (24%), and having patience (13%). Additional advice was to seek online support, normalize the range of parental emotions, and be open about their diagnosis (11% each). They advised teaching their

children coping skills and confidence (8%) and keeping calm (8%). When asked specifically about hearing aid use, about a fifth of caregivers reported children were motivated by the benefit of improved hearing. When asked about ways to help promote hearing aid adherence, caregivers suggested introducing aids at a young age (29%), adjusting for comfort (12%), praising use of aids (12%), practicing coping skills for questions about the aid (8%), and using a reward system (2%).

School Setting

Challenges in school reported by caregivers (Table 5) included being shy (19%), difficulties with attention (19%), and being teased (17%). Reading and writing were identified as academic areas in which some children experienced difficulty (12%). Caregivers (39%) and an adult with CFM (11%) described a range of hearing difficulties at school. Active teasing was identified by caregivers (27%) and adults (56%), as well as passive social exclusion (caregivers 12%; adults 33%). Correspondingly, adults with CFM described trying to hide their difference (22%), experiencing social comparison (11%), and having difficulties in romantic relationships (11%). Caregivers noted problems in accessing and navigating school systems (27%) and in poor teacher communication and understanding (24%).

School-aged children received assistance from schools (Table 6) most commonly through consistent preferential seating (caregivers 57%; adults 33%) and in auditory support services (caregivers 45%). Caregivers also noted additional classroom accommodations (14%). Clear and frequent communication from the school to family was reported by caregivers (33%) and adults (22%). Similarly, creating a proactive approach with peers and support for teasing was provided (caregivers 10%; adults 22%). A portion of caregivers (10%) and adults (67%) reported they received no help from their schools. When asked for suggestions for schools, 53% of caregivers

noted they were satisfied by their school experience. The most offered suggestion by caregivers (21%) and adults (29%) was addressing hearing needs appropriately. Other suggestions were increasing peer awareness of CFM (caregivers 11%; adults 14%), addressing teasing (caregivers 5%; adults 14%), and improving their own understanding of CFM (caregivers 5%; adults 57%). Improved communication with parents was also suggested by caregivers (11%) and adults (29%) wanted to be treated like their peers.

Medical Setting

Difficulties identified in the medical setting (Table 7) by a quarter of caregivers included a lack of knowledge about CFM among their providers. A lack of empathy or understanding (caregivers 19%; adults 40%) and little guidance about CFM treatment (caregivers 16%; adults 20%) were also concerns. Caregivers described difficulties accessing treatment (13%), receiving conflicting medical recommendations (13%), having difficulty coordinating multiple providers (9%), experiencing treatment burden (9%), and lacking insurance coverage (6%). A group of caregivers (19%) and adults (20%) noted no concerns in the medical setting and another group provided positive feedback about their providers (caregivers 9%; adults 20%). Among caregivers, helpful components of their healthcare experience (Table 8) included having a specific treatment, such as hearing aids, provided (30%). They also valued clear communication of treatment plans (21%) and using a patient-focused approach (9%). Similarly, caregivers appreciated when medical staff had a positive attitude (9%), provided reassurance (9%), invested time with them (9%), coordinated their care (6%), and allowed for parental control (6%).

When offering suggestions to improve healthcare delivery (Table 9), caregivers (35%) and adults (50%) wanted improved treatment communication and coordination. Both caregivers (6%) and adults (17%) also suggested patient-centered approaches. Caregivers recommended

having clear and current data about CFM (16%), making appropriate referrals (13%), being more informed about CFM (13%), supporting parent advocacy (10%), and providing support in gaining insurance coverage (6%). Caregivers also wanted parents to be reassured (6%) and care to be more widely geographically accessible (6%). One adult (17%) suggested providers link individuals with CFM together to experience normalization of their diagnosis and experiences.

Discussion

There is a growing recognition among providers that the related features of CFM are better conceptualized as a unified spectrum to allow for coordinated advances in treatment and research.[23] A greater appreciation of the multi-systemic stressors experienced by patients with CFM and their families is a crucial component in improving the provision of healthcare. The present study is one of only a few to comprehensively assess patient and caregiver experiences of CFM and one of the first to utilize a mixed methods approach. The findings provide important insight into the stressors experienced in the home, school, community, and medical settings. It is important to note that within most areas, a substantial subgroup reported that they had not experienced difficulties and may reflect resiliency and effective use of supports and services. Participants also outlined resources that they found helpful and suggestions for the delivery of healthcare.

Participants identified many medical-related stressors, including a perceived lack of knowledge of CFM among health professionals with correspondingly poor treatment coordination and conflicting medical recommendations. Families also reported difficulties accessing appropriate treatment, often related to lack of insurance coverage. Several caregivers and patients described feeling medical providers lacked empathy and understanding. Finally, the burden of ongoing treatment was noted, including frequent appointments, costs, and trying to

address poor coordination of care between providers. Responses reflected identified components of treatment burden, including financial burden, time and travel burden, and healthcare access burden.[11]

To help overcome these challenges, participants wanted health professionals to be better informed about CFM, refer to specialists appropriately, and coordinate care. They reported valuing a patient-focused approach with clearly communicated treatment plans and family involvement in medical decision-making. Caregivers noted the importance of providers demonstrating a positive attitude, providing reassurance, and investing time with them. These suggestions are similar to findings in other populations. For example, patients with a cleft and their caregivers wanted their views to be heard and more treatment coordination and information.[24] Similarly, a qualitative study with patients with chronic conditions described the positive impact of well-coordinated care and feeling respected, listened to, and empowered.[25]

Caregivers' and adults' described multiple ways hearing loss affected them across settings in their daily lives, from tracking each other to safety concerns. As has been well documented, this sample also reported on how children's hearing loss can impact their social and academic functioning and quality of life.[15] Appropriate ENT treatment and follow-up for hearing loss including hearing aids from early childhood can help address these concerns, particularly in language development.[26] As learning difficulties are also known to be a concern, the importance of supporting families in school advocacy was highlighted, such as ensuring children have preferential seating accommodations, auditory support, and FM system use.[3,5] Correspondingly, speech and language difficulties were also described as impacting participants in multiple settings. Beyond linking families to language and speech therapy intervention, research drawn from the fields of cleft lip/palate and hearing loss have identified several strategies for

overcoming these difficulties in the context of social settings and may also be applicable for those affected by CFM.[27,28]

Another common theme was the potential impact of negative social encounters, ranging from curious stares or questions to direct teasing and bullying, which contributed to participants trying to hide their CFM features, feeling excluded, and difficulties in romantic relationships. Parents also described their own distress in witnessing their children's response to teasing and called for schools in particular to address teasing and increase CFM awareness. These patterns are reflected in the visible difference literature.[16] Cognitive Behavioral Therapy techniques and social skills training offer potential strategies to help cope with the impacts of a visible difference and are being trialed in an online format.[18,29-31] Linking families to such interventions may offer valuable support to those affected by CFM. Psychological support, as well as opportunities to meet others affected by CFM, should be offered as standard in a variety of formats and throughout the entire treatment pathway. In addition, public awareness campaigns, such as those promoted by the American Cleft Palate-Craniofacial Association (US), the Cleft Lip and Palate Association (UK), and Changing Faces (UK) may improve social experiences for individuals with CFM and their caregivers.

Psychosocial research in the field of craniofacial conditions has typically focused on the identification of problems and deficits, with less focus on resilience and personal growth. Learning about the supports and coping used by families and incorporating this learning into their routine care has the potential to reduce family distress while improving treatment. For example, in addition to family and friends, many participants identified online support websites, medical providers, others with CFM, and scientific research as sources of support. These responses highlight the vital role healthcare professionals can play in connecting families to

support, as well as directly through providing CFM information, clearly communicating about treatment, and maintaining a positive and reassuring attitude. Advice offered to others affected by CFM included many positive coping strategies, including: reassurance, positive reframing techniques, acceptance, self-confidence, patience, openness about CFM, and seeking medical information and social support.

A study limitation is the small sample that could be due to limited time, interest, internet access, and internet literacy; however, by posting on websites accessed within and outside of the United States, participants came from a more geographically diverse population than previous studies performed in a clinic or hospital setting. Another limitation is the possibility of both negative and positive bias about their experiences based on self-selection of participants. As the majority of respondents were white, non-Hispanic, well-educated individuals who were able to navigate the web-based survey, the sample is not considered representative of the general population. Nonetheless, it would be expected that the issues identified within this higher-functioning sample would be reflected with greater severity among populations that experience more barriers in accessing health information and care. Further qualitative and quantitative research with larger sample sizes, including children and adolescent perspectives, and in other languages and countries will help increase understanding of experiences in multiple cultures.

In conclusion, this study adds to our understanding of the perspective of individuals with CFM and their caregivers' stressors across settings. The range of adaptive coping strategies and supports described and their corresponding suggestions for providers can inform standards for care.

REFERENCES

1. Gorlin RJ, MM Cohen, Hennekam RCM. *Syndromes of the head and neck*. Oxford monographs on medical genetics, no. 42. Oxford, England; New York Oxford University Press; 2001.
2. Cline JM, KE Hicks, Patel KG. Characterization of facial paresis in hemifacial microsomia. *Otolaryngol Head Neck Surg.* 2014;150:188-193.
3. Speltz ML, Wallace ER, Collett, BR et al. Intelligence and academic achievement of adolescents with craniofacial microsomia. *Plast Reconstr Surg.* 2017;140:571-580.
4. Beleza-Meireles A, Clayton-Smith J, Saraiva JM, Tassabehji M. Oculo-auriculo-vertebral spectrum: a review of the literature and genetic update. *J Med Genet.* 2014;51:635-645.
5. Collett BR, Speltz ML, Cloonan YK, et al. Neurodevelopmental outcomes in children with hemifacial microsomia. *Arch Pediatr Adolesc Med.* 2011;165:134-140.
6. Heike CL, Luquetti DV, Hing AV. Craniofacial Microsomia Overview. In: Adam MP, Ardinger, HH, Pagon, et al, eds. *GeneReviews*. Seattle, WA: University of Washington; 2009.
7. Brandsteter KA, Patel KG. Craniofacial microsomia. *Facial Plast Surg Clin North Am.* 2016;24:495-515.
8. Jiamei DC, Jiake Z, Hongxing G, et al. An investigation of psychological profiles and risk factors in congenital microtia patients. *J Plast Reconstr Aesthet Surg*. 2008;61 Suppl 1:S37-43.
9. Johns AL, Lewin SL, Im DD. Teasing in younger and older children with microtia before and after ear reconstruction. *J Plast Surg Hand Surg.* 2017;51:205-209.

10. Johns AL, Im DD, Lewin SL. Early familial experiences with microtia: psychosocial implications for pediatric providers. *Clin Pediatr (Phila)* 2017;PMID 28959893.

11. Sav A, King MA, Whity JA, et al. Burden of treatment for chronic illness: a concept analysis and review of the literature. *Health Expect*. 2015;18:312-324.

12. Khetani MA, Collett BR, Speltz ML, Werler MM. Health-related quality of life in children with hemifacial microsomia: parent and child perspectives. *J Dev Behav Pediatr* 2013;34:661-668.

13. Ongkosuwito EM, van der Vlies L, Kraaij V, et al. Parental stress in parents of a child with hemifacial microsomia: the role of child characteristics and parental coping strategies. *Cleft Palate Craniofac J.* 2016;PMID 27632763.

14. Dufton LM, Speltz ML, Kelly JP, et al. Psychosocial outcomes in children with hemifacial microsomia. *J Pediatr Psychol.* 2011;36:794-805.

15. Roland LC, Fischer K, Tran T, et al. Quality of life in children with hearing impairment: systematic review and meta-analysis. *Otolaryngol Head Neck Surg.* 2016;155:208-219.

16. Rumsey N, Harcourt D. Body image and disfigurement: issues and interventions. *Body Image.* 2004;1:83-97.

17. Thompson A, Kent G. Adjusting to disfigurement: processes involved in dealing with being visibly different. *Clin Psychol Rev.* 2001;21:663-682.

18. Clarke A, Thompson AR, Jenkinson E, Rumsey N, Newell R. *CBT for appearance anxiety : psychosocial interventions for anxiety due to visible difference*. Hoboken, NJ: John Wiley & Sons; 2013.

19. Feragen KB, Stock NM. Psychological adjustment to craniofacial conditions (excluding oral clefts): A review of the literature. *Psychol Health.* 2017;32:253-288.

20. Rosenfeld RM, Wyer PC. Stakeholder-driven quality improvement: a compelling force for clinical practice guidelines. *Otolaryngol Head Neck Surg.* 2017; PMID 29039253.

21. Harris PA, Taylor R, Thielke R, et al. Research electronic data capture (REDCap) - a metadata-driven methodology and workflow process for providing translational research informatics support. *J Biomed Inform.* 2009;42:377-381.

22. Flick U. *An introduction to qualitative research*. Thousand Oaks, CA: Sage; 1998.

23. Heike CL, Hing AV, Aspinall CA, et al. Clinical care in craniofacial microsomia: a review of current management recommendations and opportunities to advance research. *Am J Med Genet C Semin Med Genet.* 2013;163:271-282.

24. Semb G, Brattstrom V, Molsted K, et al. The Eurocleft study: intercenter study of treatment outcome in patients with complete cleft lip and palate. Part 4: relationship among treatment outcome, patient/parent satisfaction, and the burden of care. *Cleft Palate Craniofac J.* 2005;42:83-92.

25. Ridgeway JL, Egginton JS, Tiedje K, et al. Factors that lessen the burden of treatment in complex patients with chronic conditions: a qualitative study. *Patient Prefer Adherence.* 2014;8:339-351.

26. Appachi S, Specht JL, Raol N, et al. Auditory outcomes with hearing rehabilitation in children with unilateral hearing loss: a systematic review. *Otolaryngol Head Neck Surg.* 2017;157:565-571.

27. Havstam C, Laakso K, Lohmander A, Ringsberg KC. Taking charge of communication: adults' descriptions of growing up with a cleft-related speech impairment. *Cleft Palate-Craniofac J.* 2011;48:717-726.

28. Cole EB, Flexer C. *Children with hearing loss: developing listening and talking, brith to six*, 3rd ed. San Diego, CA: Plural Publishing; 2015.

29. Norman A, Moss TP. Psychosocial interventions for adults with visible differences: a systematic review. *PeerJ.* 2015;3:e870.

30. Bessell AV, Brough A, Clarke D, et al. Evaluation of the effectiveness of Face IT, a computer-based psychosocial intervention for disfigurement-related distress. *Psychol Health Med.* 2012;17:565-577.

31. Williamson H, Griffiths C, Harcourt D. Developing young person's Face IT: online psychosocial support for adolescents struggling with conditions or injuries affecting their appearance. *Health Psychol Open.* 2015;2:2055102915619092.