BMJ Open

Characterising the speech phenotype in individuals with craniofacial microsomia: a scoping review protocol

Sara Kinter ,[1,2,3] Katelyn Kotlarek,[4] Anna Meehan,[1,5] Carrie Heike[1,2,5]

To cite: Kinter S, Kotlarek K, Meehan A, *et al*. Characterising the speech phenotype in individuals with craniofacial microsomia: a scoping review protocol. *BMJ Open* 2023;**13**:e069233. doi:10.1136/bmjopen-2022-069233

► Prepublication history and additional supplemental material for this paper are available online. To view these files, please visit the journal online (http://dx.doi.org/10.1136/bmjopen-2022-069233).

Received 15 October 2022
Accepted 15 February 2023

[1]Craniofacial Center, Seattle Children's Hospital, Seattle, Washington, USA
[2]Pediatrics, Division of Craniofacial Medicine, University of Washington, Seattle, Washington, USA
[3]Center for Child Health, Behavior, and Development, Seattle Children's Research Institute, Seattle, WA, USA
[4]College of Health Sciences, Division of Communication Disorders, University of Wyoming, Laramie, Wyoming, USA
[5]Center for Clinical and Translational Sciences, Seattle Children's Research Institute, Seattle, WA, USA

Correspondence to
Dr Sara Kinter;
sara.kinter@seattlechildrens.org

ABSTRACT

Introduction Asymmetric mandibular hypoplasia, microtia, tongue and laryngeal anomalies, and soft palate and facial nerve dysfunction are clinical features observed in children with craniofacial microsomia (CFM). Despite involvement of all these structures in hearing and speech, there is limited evidence reporting speech outcomes in this population. Systematic reviews of clinical and surgical interventions related to CFM have been published, but no methodological review of speech outcomes exists. This scoping review will summarise what is known about speech production in individuals with CFM as well as illustrate gaps in the existing body of literature that will guide future research.

Methods/analysis This review will follow the methodological framework for scoping reviews first reported by Arksey & O'Malley and revised by Levac and others. Databases searched will include Ovid MEDLINE, EMBASE, CINAHL, PsycINFO and grey literature. Articles reporting any parameter of speech production in individuals with CFM will be considered for inclusion. Articles published in a language other than English will be excluded. Articles will be screened in three stages: (1) title review, (2) abstract review and (3) full text review. Ten per cent of articles will be rescreened by a second reviewer. Reference lists will be hand reviewed to identify additional relevant articles. Data charting will capture article metadata, study population and design, CFM diagnostic criteria, speech outcome measurement and key findings. The Preferred Reporting Systems for Systematic Reviews and Meta-Analyses Protocols-Extension for Scoping Reviews checklist will guide reporting of results. Descriptive analysis and data visualisation strategies will be used.

Ethics and dissemination Institutional review board approval is not required for a scoping review, as it does not directly involve human subjects. Results will be disseminated through peer-reviewed publication as well as conference presentation.

STRENGTHS AND LIMITATIONS OF THIS STUDY

⇒ Protocol designed by a multidisciplinary investigator team to ensure comprehensive review of the literature related to speech production in individuals with craniofacial microsomia.
⇒ Search strategy designed to be sensitive rather than specific, allowing for comprehensive description of publication trends in the investigation of speech production in this population.
⇒ Because translation services were not an available resource, only articles published in English or already translated into English were included, potentially resulting in exclusion of evidence published in a language other than English.
⇒ This scoping review focuses specifically on speech production characteristics directly related to structure and function of the vocal tract, thus the review may not fully capture literature related to the link between language, cognition and hearing on speech sound production.

INTRODUCTION

Craniofacial microsomia (CFM) is the second most common congenital condition related to anatomy of the head and face after cleft lip/palate, occurring in between 0.4 and 17 per 10 000 births worldwide with variable prevalence across regions and dependent on ascertainment criteria.[1–5] CFM results from disruption in the embryological development of the first and second branchial arches, leading to variable asymmetric underdevelopment of structures, which often include the ear, mandible, temporomandibular joint, facial nerve and facial soft-tissue and musculature.[6–8] Many children with CFM also demonstrate atypical structure of the larynx, soft palate and tongue.[9–13] The manifestation of these anatomic differences in an individual child is highly variable, with some having isolated microtia/atresia, while others may have more complicated phenotypes including cleft palate, mandibular hypoplasia and a tracheostomy. All craniofacial structural characteristics of the CFM spectrum are involved in speech production. The larynx, which encompasses the vocal folds, is responsible for voice production via approximation and coordination of the vocal folds with exhaled air from the lungs below. Superiorly, the soft palate controls the amount of nasal resonance and oral pressure present during speech production; elevation of the soft palate directs sound orally, decreasing

Table 1 Inclusion and exclusion criteria based on Population-Concept-Context framework

Category	Description
P-Population	Children, adolescents and young adults with CFM/Goldenhar/oculo-auriculo-vertebral syndrome/microtia and atresia
C-Concept	Anatomic differences that overlap with structures used to produce intelligible/acceptable speech. Studies should report phenotype findings including those related to ears, mandible, larynx, pharynx, cleft palate, velopharyngeal insufficiency, unilateral hemiparalysis, or facial nerve dysfunction. Potential outcomes include speech reception or discrimination threshold, articulation/phonological disorder, velopharyngeal dysfunction/insufficiency and voice disorder.
C-Context	The language is limited to English and readily available English translations. Articles using any type of study design, including case reports/series, will be included to capture both breadth and heterogeneity of research in this area.

CFM, craniofacial microsomia.

nasal resonance and increasing oral pressure. The tongue is responsible for place and manner characteristics of speech sounds (articulation) and for controlling resonance to differentiate vowels. However, there is limited evidence as to the prevalence, aetiology and phenotype of speech disorders in individuals with CFM despite these known anatomical differences.

While there are widely accepted standards of care related to structural versus functional speech disorders in children with other craniofacial differences (such as cleft lip and palate,[14 15] comparable standards do not yet exist for children with CFM. Further, the burden of speech disorders in individuals with CFM is not well understood. Speech-language pathologists with craniofacial-specific training are an important part of the care team for individuals with CFM due to the complex nature and multifaceted origin of speech abnormalities in this population. However, clinicians need evidence to support an assessment protocol that includes monitoring by a speech-language pathologist trained in this area. Communication to stakeholders regarding the burden of speech disorders in CFM may increase awareness to craniofacial providers that manage the medical care of these individuals. The first step in the development of speech-related standards of care is to understand how speech production manifests in individuals with CFM.

The purpose of this scoping review is to provide a summary of research on speech outcomes in children with CFM, guide providers in improving management of these children and identify gaps in the literature where additional evidence is needed to improve the standard of care. Results have the potential to identify gaps in scientific knowledge of speech disorders associated with CFM, generate recommendations for future studies and inform the future standard of care for evaluation and management by the craniofacial speech-language pathologist as part of a multidisciplinary care team.

METHODS AND ANALYSIS

Scoping reviews, unlike systematic reviews, are designed to summarise evidence within a broader topic area and consequently require fewer limits on included study designs. Because the association between a diagnosis of CFM and speech disorders is a relatively novel area of investigation, the methodological framework for scoping reviews originally described by Arskey & O'Malley[16] and refined by Levac *et al*[17–20] was considered appropriate to answer the proposed research questions. The final protocol was registered prospectively in the Open Science Framework on 9 September 2022 (osf.io/npr94). Data collection relating to study design and confounding control will provide some insight into the current level of evidence. Results will be reported using the Preferred Reporting Systems for Systematic Reviews and Meta-Analyses Protocols – Extension for Scoping Reviews (PRISMA-ScR).[21]

Stage 1: identifying the research questions

Our primary objective is to summarise the literature pertaining to speech sound production in children and adolescents with CFM. Specific questions include:

1. Which parameters of 'speech production' have been studied?
2. What approaches to study design have been undertaken?
3. How is speech production/disorder defined and measured?

Stage 2: developing a search strategy

A broad systematic review of the literature that includes all articles reporting characteristics of speech production, including articulation, resonance, voice and motor coordination that involve individuals with CFM will be conducted. There are no planned restrictions on study design or year of publication, as our goal is to obtain a broad perspective of what has been published relating to speech production in children and adolescents with CFM. Only articles published in English or already translated into English will be included in the review. The search will include Ovid MEDLINE, EMBASE, CINAHL, PsycINFO, OpenDissertations and Google Scholar. The search strategy was developed in consultation with a librarian and will include terms for CFM (CFM OR OAVS

OR Goldenhar OR hemifacial microsomia OR microtia) AND speech (speech OR voice OR resonance OR velopharyngeal insufficiency). The detailed search strategy for OVID Medline is shown in online supplemental table 1. This search will be translated for other databases and grey literature, as appropriate. Investigators will review reference lists by hand to identify additional relevant articles once the full-text review is complete.

Stage 3: study selection

Inclusion/exclusion criteria based on the Population-Concept-Context framework[22] are described in table 1.

All references identified using the search strategy will be managed with duplicates removed by EndNote reference manager software. Article review will occur in three phases. Each phase will involve two independent reviewers and a training process designed to further clarify inclusion/exclusion criteria through discussion of disagreements. Specifically, rereview of 10% of randomly selected articles will be repeated until at least 90% agreement for inclusion and exclusion is reached. Any disagreements will be discussed between researchers and arbitrated by a third reviewer, as necessary.

Following deduplication, articles will be divided into two groups for independent title review by a single reviewer for each group (SK and AM). Based on title review only, articles will be marked for inclusion, exclusion or undecided. In the second phase of study selection, remaining titles will be divided into two groups. Abstracts for articles in each group will be independently reviewed (SK and KK) and again marked for inclusion, exclusion or undecided. Finally, remaining articles will again be divided into two groups for full-text review by the same independent reviewers. The title and article selection process will be managed using Rayyan.[23] Full-text review will be tracked using Research Electronic Data Capture (REDCap)[24] and summarised using a PRISMA-ScR[21] flow diagram.

Stage 4: charting the data

Study data will be collected and managed using REDCap electronic data capture tools hosted at the University of Washington.[24 25] REDCap is a secure, web-based software platform designed to support data capture for research studies, providing, (1) an intuitive interface for validated data capture; (2) audit trails for tracking data manipulation and exposure procedures; (3) automated export procedures for seamless data downloads to common statistical package; and (4) procedures for data integration and interoperability with external sources. Two reviewers (SK and KK) will independently extract data into a REDCap project designed specifically for this scoping review in the following areas:

1. Study metadata: authors, title, publication year, journal and country of publication.
2. Study design/methodology: design architecture, primary and secondary aims/objectives.
3. Study population: CFM case definition, sample size, comparison group (if applicable) and age range.
4. Analyses and outcomes: speech outcome, outcome measurement, type of analysis undertaken, methods for confounding control and key findings.

Reliability of data extraction will be determined following independent review of the first five included studies. Consistency of data extraction will be determined following rereview of 10% of included studies. For any papers with unclear or missing data, we will make one attempt to contact corresponding authors via email for clarification.

Stage 5: synthesising, summarising and reporting results

A descriptive analysis will be completed to summarise characteristics of speech production reported in the literature, how CFM is defined, and the study designs used to generate evidence. Data visualisation strategies will be used, including the PRISMA-ScR diagram for reporting the study selection process. Exploratory analysis may be initiated, if deemed appropriate. It is anticipated that the most useful summary of results will be met through descriptive analysis as well as qualitative discussion.

Patient and public involvement

Patients will not be directly involved.

DISSEMINATION AND ETHICS

Because this study involves data extracted from published studies, formal ethics approval is not indicated. Our findings will be submitted to a national conference of multidisciplinary researchers in children with craniofacial conditions for presentation and will be published in a peer-reviewed journal following the reporting standards for scoping reviews (PRISMA-ScR).[21]

Acknowledgements The authors would like to thank Sue Groshong, MLIS, who worked closely with the authors to design and conduct the literature search.

Contributors SK conceived of the presented idea. SK, KK, AM and CH contributed to the design of the protocol. SK initially drafted the manuscript and all authors participated in manuscript revision.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Disclaimer The views expressed in this article are the authors' own and not an official position of the institution.

Competing interests None declared.

Patient and public involvement Patients and/or the public were not involved in the design, or conduct, or reporting, or dissemination plans of this research.

Patient consent for publication Not applicable.

Provenance and peer review Not commissioned; externally peer reviewed.

ORCID iD

Sara Kinter http://orcid.org/0000-0001-6017-4529

REFERENCES

1 Paul MA, Opyrchał J, Knakiewicz M, *et al*. Hemifacial microsomia review: recent advancements in understanding the disease. *J Craniofac Surg* 2020;31:2123–7.
2 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;163C:271–82.
3 Barisic I, Odak L, Loane M, *et al*. Prevalence, prenatal diagnosis and clinical features of oculo-auriculo-vertebral spectrum: a registry-based study in Europe. *Eur J Hum Genet* 2014;22:1026–33.
4 Junaid M, Slack-Smith L, Wong K, *et al*. Epidemiology of rare craniofacial anomalies: retrospective western australian population data linkage study. *J Pediatr* 2022;241:162–72.
5 Luquetti DV, Heike CL, Hing AV, *et al*. Microtia: epidemiology and genetics. *Am J Med Genet A* 2012;158A:124–39.
6 Poswillo D. The aetiology and pathogenesis of craniofacial deformity. *Development* 1988;103 Suppl:207–12.
7 Grabb WC. The first and second branchial arch syndrome. *Plast Reconstr Surg* 1965;36:485–508.
8 Carvalho GJ, Song CS, Vargervik K, *et al*. Auditory and facial nerve dysfunction in patients with hemifacial microsomia. *Arch Otolaryngol Head Neck Surg* 1999;125:209–12.
9 Chen EH, Reid RR, Chike-Obi C, *et al*. Tongue dysmorphology in craniofacial microsomia. *Plast Reconstr Surg* 2009;124:583–9.
10 Shprintzen RJ, Croft CB, Berkman MD, *et al*. Velopharyngeal insufficiency in the facio-auriculo-vertebral malformation complex. *Cleft Palate J* 1980;17:132–7.
11 Luce EA, McGibbon B, Hoopes JE. Velopharyngeal insufficiency in hemifacial microsomia. *Plast Reconstr Surg* 1977;60:602–6.
12 Funayama E, Igawa HH, Nishizawa N, *et al*. Velopharyngeal insufficiency in hemifacial microsomia: analysis of correlated factors. *Otolaryngol Head Neck Surg* 2007;136:33–7.
13 D'Antonio LL, Rice RD, Fink SC. Evaluation of pharyngeal and laryngeal structure and function in patients with oculo-auriculo-vertebral spectrum. *Cleft Palate Craniofac J* 1998;35:333–41.
14 American Cleft Palate-Craniofacial Association. Parameters for the evaluation and treatment of patients with cleft lip/palate or other craniofacial anomalies. *Cleft Palate Craniofac J* 1993;30.
15 D'Antonio LL. Evaluation and management of velopharyngeal dysfunction: a speech pathologist's viewpoint. *Probs Plast Reconstr Surg* 1992;2:85–111.
16 Arksey H, O'Malley L. Scoping studies: towards a methodological framework. *Int J Soc Res Methodol* 2005;8:19–32.
17 Daudt HML, van Mossel C, Scott SJ. Enhancing the scoping study methodology: a large, inter-professional team's experience with arksey and O'malley's framework. *BMC Med Res Methodol* 2013;13:48.
18 Levac D, Colquhoun H, O'Brien KK. Scoping studies: advancing the methodology. *Implementation Sci* 2010;5.
19 Colquhoun HL, Levac D, O'Brien KK, *et al*. Scoping reviews: time for clarity in definition, methods, and reporting. *J Clin Epidemiol* 2014;67:1291–4.
20 Peters MDJ, Marnie C, Tricco AC, *et al*. Updated methodological guidance for the conduct of scoping reviews. *JBI Evidence Synthesis* 2020;18:2119–26.
21 Tricco AC, Lillie E, Zarin W, *et al*. PRISMA extension for scoping reviews (PRISMA-scr): checklist and explanation. *Ann Intern Med* 2018;169:467–73.
22 Peters MDJ, Godfrey CM, Khalil H, *et al*. Guidance for conducting systematic scoping reviews. *Int J Evid Based Healthc* 2015;13:141–6.
23 Ouzzani M, Hammady H, Fedorowicz Z, *et al*. Rayyan-a web and mobile app for systematic reviews. *Syst Rev* 2016;5:210.
24 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;42:377–81.
25 Harris PA, Taylor R, Minor BL, *et al*. The redcap consortium: building an international community of software platform partners. *J Biomed Inform* 2019;95:103208.

Supplementary Table 1: Ovid MEDLINE search strategy.*

#	Ovid Medline and Epub Ahead of Print, In-Process, In-Data-Review & Other Non-Indexed Citations, 1946 to Current
1	Facial Asymmetry/ or Goldenhar Syndrome/ or (goldenhar* or otomandibular dysostos* or otomandibular syndrome* or ((branchial* or first or pharyngeal*) adj3 arch syndrome*) or ((crani* or face? or facies or facial* or orbitocranial*) adj3 asymmetr*) or ((craniofacial* or hemifacial*) adj3 microsomia*) or ((facioauriculovertebral or facio auricul* or lateral facial or moeschler clarren* or oculoauricul* or oculo auricul* or oral mandibular auricular) adj3 (disease* or dysplasia* or sequence* or spectrum* or syndrome*)) or (unilateral* adj3 hypoplas*)).tw,kf.
2	exp Central Nervous System/ab or (anarthri* or aphoni* or apractic* or apraxi* or articulat* or cluttering* or communicat* or disarthric* or disarticulat* or dysarthos* or dysarthr* or dysfluen* or dysphoni* or dysglossia* or dyslali* or dyspraxi* or fluen* or hemiparaly* or hemipegi* or hoarseness* or hypernasal* or hyperphoni* or hyperrhinolali* or hyponasal* or hypophoni* or leptophoni* or logopath* or logopedi* or microphoni* or misarticulat* or mute* or mutism* or neurodevelop* or neurolog* or oral* or pals* or paralali* or paraly* or pareses or paresis or paretic* or phonat* or pronounc* or (resonance* not magnetic resonance*) or rhinism* or rhinolali* or rhinophoni* or speech* or stammer* or stutter* or trachyphoni* or verbal* or vocal* or voice* or ((absen* or los#) adj3 voice) or (central nervous system* adj3 (abnormal* or anomal* or defect* or deficien* or disease* or disorder* or dysfunct* or fail* or inadequa* or incompeten* or insufficien* or malform*))).jw,mp.
3	Velopharyngeal Insufficiency/ or ((palata or palatal* or palate* or palatine* or palatonasal* or palatopharyn* or palatum or pharyn* or velopalat* or velopharyn*) adj3 (abnormal* or anomal* or defect* or deficien* or disease* or disorder* or dysfunct* or fail* or inadequa* or incompeten* or insufficien* or malform*)).tw,kf.
4	Congenital Microtia/ or (anotia* or microtia* or small ear? or ((auricul* or ear? or mandibul*) adj3 (aplasia* or hypoplas*))).tw,kf.
5	((aural* or auricul* or ear*) adj3 atresi*).tw,kf.
6	exp Infant/ or exp Child/ or Child, Hospitalized/ or Adolescent/ or Adolescent, Hospitalized/ or Young Adult/ or (pediatric* or paediatric* or child* or newborn* or neonat* or baby or babies or infan* or preschool* or toddler* or kindergarten* or ((nursery or primary or elementary or middle or high or age*1) adj school*) or teen* or preteen* or preadolescen* or adolescen* or youth* or youngster* or boy* or girl* or juvenile* or young adult* or college*).tw,kf,so,jw.
7	1 and (2 or 3) and 6
8	2 and (4 or 5) and 6
9	(7 or 8) and english.la.

* Field codes: jw = journal word; kf = keyword heading word; la = language; mp = title, abstract, original title, name of substance word, subject heading word, floating sub-heading word, keyword heading word, organism supplementary concept word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms; so = source; tw = text word (title or abstract)