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

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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]

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