Int. J. Oral Maxillofac. Surg. 2015; 44: 592–598
http://dx.doi.org/10.1016/j.ijom.2015.01.023, available online at http://www.sciencedirect.com

International Journal of Oral & Maxillofacial Surgery

Systematic Review
Craniofacial Surgery

Obstructive sleep apnoea in craniofacial microsomia: a systematic review

C. J. J. M. Caron[1], B. I. Pluijmers[1], K. F. M. Joosten[1], I. M. J. Mathijssen[1], M. P. van der Schroeff[1], D. J. Dunaway[2], E. B. Wolvius[1], M. J. Koudstaal[1]

[1]The Dutch Craniofacial Centre, Department of Oral and Maxillofacial Surgery, Erasmus University Medical Centre, Sophia's Children's Hospital Rotterdam, Rotterdam, Netherlands; [2]The Craniofacial Unit, Great Ormond Street Hospital, London, UK

C.J.J.M. Caron, B.I. Pluijmers, K.F.M. Joosten, I.M.J. Mathijssen, M.P. van der Schroeff, D.J. Dunaway, E.B. Wolvius, M.J. Koudstaal: Obstructive sleep apnoea in craniofacial microsomia: a systematic review. Int. J. Oral Maxillofac. Surg. 2015; 44: 592–598.

Abstract. Children with craniofacial microsomia (CFM) are at risk of obstructive sleep apnoea (OSA). This systematic review provides an overview of the literature on the prevalence of OSA in children with CFM. A search was performed in PubMed, Embase, Cochrane Library, and Web of Science for articles on CFM and OSA. The following data were extracted from the articles: number of patients, patient characteristics, presence of OSA, polysomnography outcomes, and the treatments and outcomes of OSA. We included 16 articles on CFM and OSA, four of which reported the prevalence of OSA (range 7–67%). Surgical treatment was more often described in these patients than conservative treatment. According to the literature, OSA is related to CFM. However, as there have been no prospective studies and few studies have presented objective measurements, no definitive conclusions can be drawn. Prospective studies are needed to determine the prevalence of OSA in patients with CFM.

Key words: craniofacial microsomia; hemifacial microsomia; Goldenhar; obstructive sleep apnoea; systematic review.

Accepted for publication 29 January 2015
Available online 11 March 2015

A common problem in children with a craniofacial anomaly is upper airway obstruction. This obstruction may be related to bilateral mandibular hypoplasia, as is the case in children with Pierre Robin sequence and Treacher Collins syndrome. The prevalence of upper airway obstruction, and more specifically of obstructive sleep apnoea (OSA), in children with Pierre Robin sequence and Treacher Collins syndrome is 12.5% and 46%, respectively.[1,2] In the normal population this is 3–4%.[3–5]

OSA is one of the clinical manifestations of upper airway obstruction and is characterized by snoring, laboured breathing during sleep, apnoea, and excessive daytime sleepiness. Complications of OSA include failure to thrive, pulmonary hypertension, cor pulmonale, and sudden death. Therefore, the accurate diagnosis and identification of risk groups is important.

As is the case for Pierre Robin sequence and Treacher Collins syndrome patients, craniofacial microsomia (CFM) patients have mandibular hypoplasia as a clinical characteristic. CFM is the result of a disturbance in the embryological development of the first and second branchial arches and is characterized by asymmetric underdevelopment of the facial structures, including the mandible, maxilla, ears, soft tissues, and facial nerves.[6,7] CFM is most often regarded as a unilateral malformation, however the facial structures are involved bilaterally in 10% of cases[8,9] and several recent publications have suggested that the contralateral side is abnormal in most cases as well, although not truly hypoplastic.[10,11] The reported incidence rate ranges from 1 in 3500 to 1 in 20,000,[6,12,13] which makes CFM the second most common facial birth defect after

cleft lip and palate. CFM in combination with epibulbar dermoid and extra-craniofacial anomalies, such as heart, renal, and vertebral anomalies, is known as Goldenhar syndrome.[14–19]

The most typical deformity of CFM is mandibular hypoplasia, which occurs in 89–100% of cases.[20] The most commonly used classification of mandibular hypoplasia is the classification of Pruzansky modified by Kaban, in which mandibular hypoplasia is classified into four types.[21,22] In type I, the mandibular ramus and temporomandibular joint (TMJ) are of normal shape but small. In type IIa, the mandibular ramus is abnormal in both size and shape, but the deformed TMJ is adequately positioned. In type IIb, the mandibular ramus and TMJ are abnormal in shape, size, and location. Type III deformity consists of an absent ramus, condyle, and TMJ.

As mandibular hypoplasia increases the risk of airway obstruction, patients with CFM are theoretically at risk of airway obstruction. Several authors have stated that patients with CFM should be screened routinely for OSA.[20,23,24] Nevertheless, the exact prevalence of OSA in CFM and the severity of the pathology on which these statements are based are not mentioned in these expert opinions.

The aim of this review is to provide an overview of the literature regarding CFM and the prevalence and treatment of OSA based on the following key questions: (1) What is the prevalence of OSA in patients with CFM? (2) What are the treatment modalities for OSA reported in patients with CFM? (3) What is known about the follow-up after treatment for OSA in patients with CFM?

Within the group of craniofacial malformation patients, feeding difficulties are often closely related to upper airway obstruction.[25] This topic is described separately in a second article entitled 'Feeding difficulties in craniofacial microsomia: a systematic review'.

Methods

Search strategy

A search of public domain databases was performed to identify articles focusing on CFM and OSA. The search was conducted in the following databases: PubMed, Embase, Cochrane Library, and Web of Science (all searched up to 27 August 2014). In addition, we performed a manual search of secondary sources including references of the articles initially identified. The goal was to identify all studies addressing CFM in relation to OSA.

The following search terms were used: (((facial[tiab] OR face[tiab] OR hemifacial[tiab] OR orbitocranial[tiab] OR facies[tiab] OR cranial[tiab] OR mandibulofacial[tiab] OR otomandibular[tiab] OR craniofacial[tiab] OR faciocranial[tiab] OR hemimandibular[tiab]) AND (microsom*[tiab] OR asymmetr*[tiab] OR dysosto*[tiab] OR dysplasia[tiab] OR anomal*[tiab] OR deformit*[tiab] OR hypoplasia[tiab] OR syndrom*[tiab] OR malformation*[tiab]) OR (treacher collins[tiab] OR goldenhar[tiab]) OR (oculoauriculovertebral*[tiab] OR facioauriculovertebral*[tiab] OR (auriculo vertebral*[tiab]))) AND (airway obstruction*[tiab] OR obstructive airway *[tiab] OR nocturnal apnea[tiab] OR nocturnal apnoea[tiab] OR sleep apnea[tiab] OR sleep apnoea[tiab] OR osa[tiab] OR osas[tiab] OR osahs[tiab])) AND publisher[sb].

Data extraction and analysis

Two investigators (C.J.J.M.C. and B.I.P.) screened the studies independently. All articles on the prevalence and treatment of OSA in patients with CFM were included. Expert opinions were excluded. The full texts of all articles meeting the inclusion criteria and articles for which the abstract was lacking information were obtained.

Articles were graded on quality of evidence using the Oxford Centre for Evidence-Based medicine (CEBM) criteria.[26] Data on the number of patients, patient characteristics such as gender, age, and severity of the CFM, the presence of OSA, polysomnography outcomes, and the treatment and outcome of OSA were tabulated when available.

Results

Craniofacial microsomia and obstructive sleep apnoea

The search retrieved 1385 relevant articles. After removing duplicate articles and including further articles from the manual search of secondary sources, 835 articles were examined based on the title and abstract. A total of 749 articles were then excluded. Of the 86 articles retrieved for further examination, 16 were included in the present analysis (Fig. 1).

What is the prevalence of OSA in patients with CFM?

Six studies and case series on the prevalence of OSA in CFM were found (Table 1). OSA was defined as complete cessation of airflow for more than two breaths or 10 s and hypopnoea as a $\geq$50% reduction in respiratory airflow accompanied by a decrease of $\geq$3% in oxygen saturation (SaO_2),[27,28] with a minimum of 30 episodes of obstructive apnoea in a 7-h sleep period.[29] In some cases OSA was not defined at all.[30–32] The prevalence of OSA in these studies varied from 7% to 67% (Table 2).

Case–control study on the prevalence of OSA in CFM

Cloonan et al.[30] described the prevalence of sleep disordered breathing (SDB) in children with CFM by studying 124 cases and 349 controls. Parents of children with CFM reported that their child snored more often than did the parents of otherwise healthy controls (29% *vs.* 17%). Eighty-four of the 124 cases received a supplementary questionnaire regarding history of airway interventions and the use of polysomnography (PSG) to classify the severity of SDB or OSA. A history of airway interventions was more often reported in children with CFM than controls (14% *vs.* 8%) and more children with CFM had undergone PSG than controls (20% *vs.* 2%). Of the 15 children with CFM who underwent PSG, 10 were diagnosed with SDB/OSA (67%); of the controls, four out of four were diagnosed with SDB/OSA.

Cross-sectional study on the prevalence of OSA in CFM

D'Antonio et al.[31] evaluated the pharyngeal and laryngeal structure and function in patients with Goldenhar syndrome by physical examination, otolaryngological examination, and video-nasoendoscopy. Nine out of 41 patients (22%) reported symptoms of airway obstruction, of whom five (12%) had OSA documented by PSG (Table 2).

Retrospective studies on the prevalence of OSA in CFM

Cohen et al.[27] found a prevalence of OSA of 24%. Upper airway disorders fell within three categories. Patients in category 1 were asymptomatic for airway disorders, patients in category 2 were suspect for intermittent OSA or had experienced a perioperative apnoeic event, and patients in category 3 had a definite history of OSA. Like Cloonan et al.,[30] they found that patients with more severe mandibular and/or extra-craniofacial anomalies had a