

Orthodontic considerations for patients presenting with Goldenhar Syndrome. A long-term case report

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Introduction: The correction of facial asymmetry in patients presenting with Goldenhar Syndrome can be challenging due to the complexity of the orthodontic and surgical procedures required, the psychosocial considerations of the patient and the risk of relapse.

Materials and methods: A Caucasian male with Goldenhar Syndrome originally presented at age 8.2 years with concerns relating to a poor bite. He had previously undertaken surgery for the removal of pre-auricular skin tags on the right side. With ongoing skeletal growth, the facial asymmetry became more obvious and caused the patient to become socially withdrawn. Multidisciplinary treatment involving orthodontics and orthognathic surgery to correct the mandibular asymmetry was delayed until facial growth had slowed. The correction involved the use of an asymmetrical bilateral sagittal split osteotomy and an advancement genioplasty followed by a dermal fat graft and Medpor® onlay to the right mandibular body and angle in addition to a refinement genioplasty.

Results: Follow-up has revealed a partial return of a buccal openbite illustrating the risk of occlusal relapse.

Conclusion: The case report illustrates the complexity of the orthodontic and surgical management of facial asymmetry, the psychosocial considerations of the patient and the risk of relapse. Multidisciplinary management is essential in the management of Goldenhar Syndrome.

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Introduction

Goldenhar syndrome (GS) (OMIM 164210 + 141400) was first described in 1952 and is part of the wider craniofacial microsomia spectrum.¹ Although craniofacial microsomia is relatively rare, it is the most common congenital craniofacial anomaly after cleft lip and palate. The condition is associated with abnormal development of the first two branchial arches, resulting in incomplete development of the ear, nose, soft palate, lip and mandible. In addition, defects within the vertebrae, heart and central nervous system can also exist.² Although the craniofacial involvement is highly variable facial

asymmetry is a common feature which becomes more noticeable with continuing facial growth. Traditionally, GS was considered as a unilateral facial malformation associated with a classic triad of mandibular hypoplasia, ear and/or eye malformation and vertebral anomalies. However, in 10% to 36% of cases there is bilateral involvement, but one side is almost always more severely affected than the other.³

The incidence of GS is estimated to range from 1:3500 to 1:5600 live births, with a male-female ratio of 3:2.⁴ A plethora of other names for the condition has been used including oculo-auriculo-vertebral spectrum, hemifacial microsomia, oculoauriculovertebral

dysplasia, facioauriculovertebral sequence, first and second branchial arch syndrome, Goldenhar–Gorlin syndrome, lateral facial dysplasia, unilateral craniofacial microsomia, otomandibular dysostosis, facio-auriculo-vertebral dysplasia, facio-auriculo-vertebral malformation complex and craniofacial microsomia.⁵

Currently the aetiology of GS remains unknown. The syndrome is thought to be due to sporadic mutations in SF3B2, the gene encoding subunit 2 of the splicing factor 3b protein complex; however, an autosomal dominant pattern has also been shown.⁶ Alternative theories include vascular perturbation and/or neural crestopathy during a critical time of embryogenesis.⁷ For example, experimental evidence suggests that occlusion of the stapedia artery during development results in Goldenhar syndrome-like features.² Abnormal development of cranial neural crest cells, caused by a range of environmental or maternal factors, may also contribute to abnormal development of the first and second branchial arches.⁸

The clinical presentation is variable but often the extra-oral features include marked facial asymmetry secondary to hypoplasia of the mandibular ramus and condyle as well as the maxilla, cleft lip, epibulbar dermoids of the eye, colobomas of the upper eyelid, microtia, or possible anotia and pre-auricular skin tags. In 50% of cases, patients suffer combined conductive and sensorineural hearing loss.⁹ Intra-oral features of GS can include canting of the occlusal plane,⁵ often a unilateral cross-bite, congenitally missing mandibular teeth as well as macrostomia and a narrowed palate.⁸

Although a classification of GS is problematic due to the heterogenous variation in presentation and multisystem involvement, the OMENS¹⁰ and Kaban's modification¹¹ of Pruzansky's original three types of mandibular deformity¹² are often used. The OMENS classification assesses the degree of involvement of the Orbits, Mandible, Ears, Nerves (VII) and Soft tissues in which each component is scored 0 to 3 depending upon the severity of the defect. The classification proposed by Kaban assesses the mandibular anatomical anomalies within four types (Type I: hypoplastic temporomandibular joint, Type IIa: hypoplastic and abnormal shape of the mandibular ramus, condyle, and temporomandibular joint, Type IIb: the mandibular ramus is hypoplastic and markedly abnormal in form and location,

being medial and anterior, Type III: an absence of the mandibular ramus, Type IV: mandibular body hypoplasia). Those patients with a functioning TMJ (Type I or IIa) do not require TMJ reconstruction and can undergo mandibular reconstruction through conventional osteotomy or distraction if there is sufficient bone stock for distractor placement. Those presenting with a non-functioning TMJ (Type II b or III) require either a costochondral graft and/or a total joint replacement.

As with other craniofacial anomalies, the rehabilitation of a patient with GS is co-ordinated and staged within a multidisciplinary team. It is managed by surgical, in addition to non-surgical practitioners and the treatment plan is considered depending on individual requirements, particularly regarding the timing of therapeutic interventions. Supportive multidisciplinary management, including specialists from paediatric cardiology, audiology and ophthalmology units is required. Treatment of the facial disharmony requires the participation of many other disciplines including plastic surgeons, otolaryngologists, oral and maxillofacial surgeons as well as orthodontists.¹³

Patient co-operation throughout the treatment process is essential to obtain facial symmetry and there is a high burden of care. Affected patients and their families often experience a range of emotional and psychological issues associated with the abnormalities and the associated treatment.¹⁴ As with other craniofacial anomalies, orthodontic intervention is often related to improving staged surgical outcomes and places the orthodontist as a key figure within the MDT due to the extended duration of multiple orthodontic treatment interventions. A patient presenting with Goldenhar Syndrome who has been followed from age 8 years to early adulthood (23 years old) is reported, with reference to the orthodontic management and treatment challenges.

Case history

Initial assessment

The GS patient first presented at the orthodontic clinic in the mixed dentition (8.2 years of age) with concerns relating to a poor bite and facial asymmetry. He had previously been seen at a craniofacial clinic multiple times and had undertaken ophthalmological surgery to his right eye as well as surgical removal of

pre-auricular skin tags on the right. His left atlanto-axial joint was fused, and there was asymmetry to his skull base. Extra orally there was a skeletal Class II relationship indicated by a retrognathic mandible, an increased Frankfort Mandibular Plane Angle (FMA)

and an increased lower face height (LFH). There was obvious facial asymmetry as the chin deviated to the right, along with a lack of soft tissue bulk and scarring associated with the previous removal of the right side pre-auricular skin tags (Figure 1A).

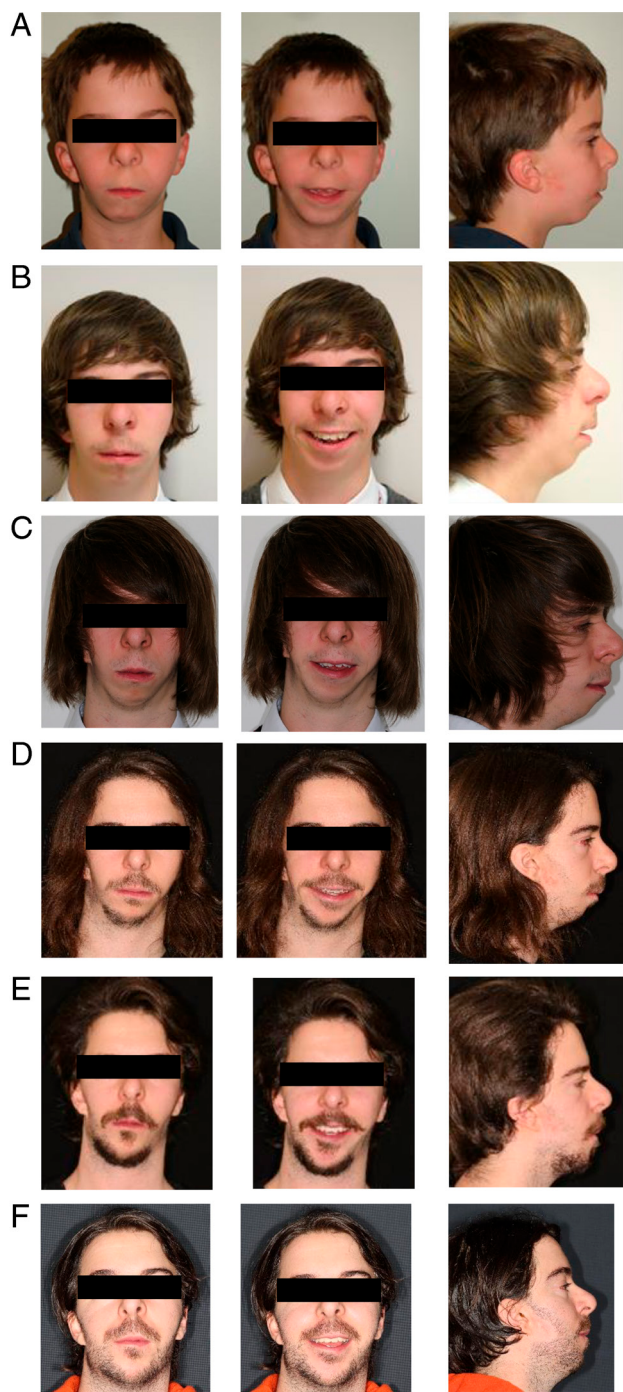


Figure 1. Extra oral photographs. (A) Patient at age 8.2 years. (B) Patient at age 15.5 years. (C) Pre-orthodontic treatment (17.3 years). (D) Presurgical (19.2 years). (E) Post orthodontic treatment and bilateral sagittal split osteotomy (BSSO) and rotational advancement genioplasty (20.2 years). (F) Follow up (post dermal fat graft/Medpor® onlay and refinement genioplasty) at 23.4 years.



Figure 2. Intra-oral photographs. (A). Mixed dentition (aged 8.2 years). (B) Permanent dentition (aged 15.5 years). (C) Pre-orthodontic Tx (aged 17.3 years). (D) Pre surgical occlusion (aged 19.2 years). (E) Post orthodontic treatment and bilateral sagittal split osteotomy (BSSO) rotational advancement with genioplasty (aged 20.2 years). (F) Follow up (aged 23.4 years).

Intraorally, there was a Class I malocclusion in the mixed dentition with proclined, spaced upper incisors and mildly crowded lower incisors. There was a 3 mm overjet, a 4 mm anterior openbite that extended to include the left deciduous first molars. Paradoxically, there was a midline shift to the left in the lower arch which changed the molar relationship towards Class III on the right and Class I on the left in association with a unilateral buccal crossbite on the left side (Figure 2A). Radiographic findings included

a small rudimentary right condyle (Kaban Type IIa) and reduced bone height to the lower mandibular right border and angle. As there was a good range of mandibular movements and no limitation to normal function, no active orthodontic treatment was offered at this time.

The patient and parents were reassured that although comprehensive orthodontic treatment in combination with surgery would likely to be required once facial

growth had slowed or ceased, it was unlikely that the skeletal mandibular asymmetry would be progressive, and that growth of the affected side would likely parallel that of the unaffected side.¹⁵

Treatment plan and progress

Orthodontic clinical records were collected at 15.5 years of age when the parents voiced concerns relating to the patient's social development and lack of peer engagement. He had developed a hair style to hide the facial asymmetry and scarring on the right side and had become socially withdrawn. His facial and intraoral features had changed little other than the eruption of the permanent dentition (Figures 1B, 2B, 4A–C). There was a slight reduction in the anterior openbite, although the buccal openbite was more extensive on the left and associated with the development of an obvious mandibular occlusal cant on that side. As he was experiencing rapid skeletal growth, a decision was made to further delay the start of orthodontic treatment. However, in preparation for future asymmetric mandibular surgery, it was decided to proceed with the extraction of the lower right second permanent molar (the 3rd molar was absent) to improve the amount of accessible bone stock in that region (Figure 3A). In addition, a referral was arranged to provide psychological support.

At 17.3 years of age the patient returned with his family expressing ongoing concerns relating to his social development. As skeletal growth appeared to have slowed, a collection of treatment planning records was undertaken (Figures 1C, 2C, 4D). Surgical and orthodontic treatment objectives were determined to address the chief concerns relating to facial asymmetry and occlusal discrepancies. Specific treatment objectives included (1) mandibular asymmetry correction and advancement, (2) openbite closure, overjet correction and dental midline correction, (3) soft tissue augmentation surgery (Medpor[®] onlay and dermal fat graft).

The patient commenced upper and lower fixed appliance treatment (0.022" MBT prescription) in preparation for asymmetrical mandibular advancement surgery. The orthodontic treatment involved the removal of the lower first premolars to allow decompensation of the proclined lower incisors (Table IA) and for the relief of mild lower anterior crowding. Both arches were levelled and aligned (Figures 2D, 3B, 4E and Table IB), prior to

undertaking a bilateral sagittal split osteotomy and an asymmetrical mandibular advancement to correct the overjet, dental midlines and obtain a Class I canine and a molar Class III relationship. An advancement genioplasty of 8 mm was also undertaken to improve chin symmetry and projection, along with the

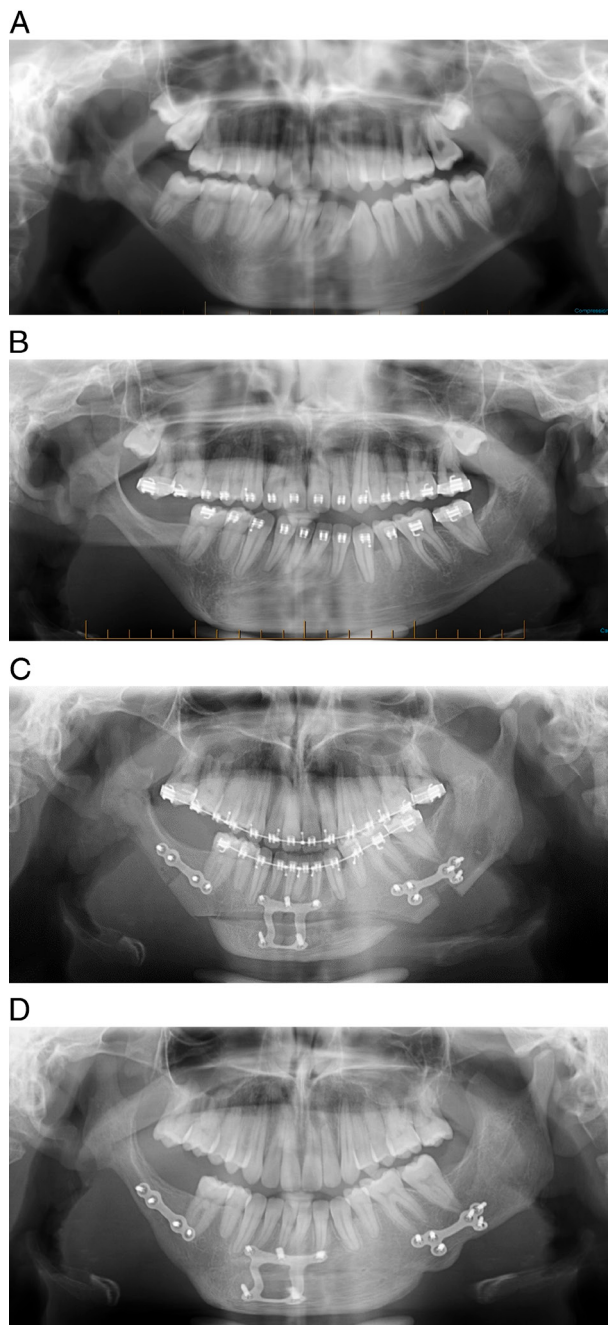


Figure 3. OPG images. (A) Prior to removal of the lower right second molar to improve accessible bone stock for future asymmetric mandibular surgery. (B) Pre surgery. (C) Immediately post-surgical BSSO differential advancement and genioplasty. (D) Post orthodontic treatment (prior to Medpor[®] onlay and refinement genioplasty).

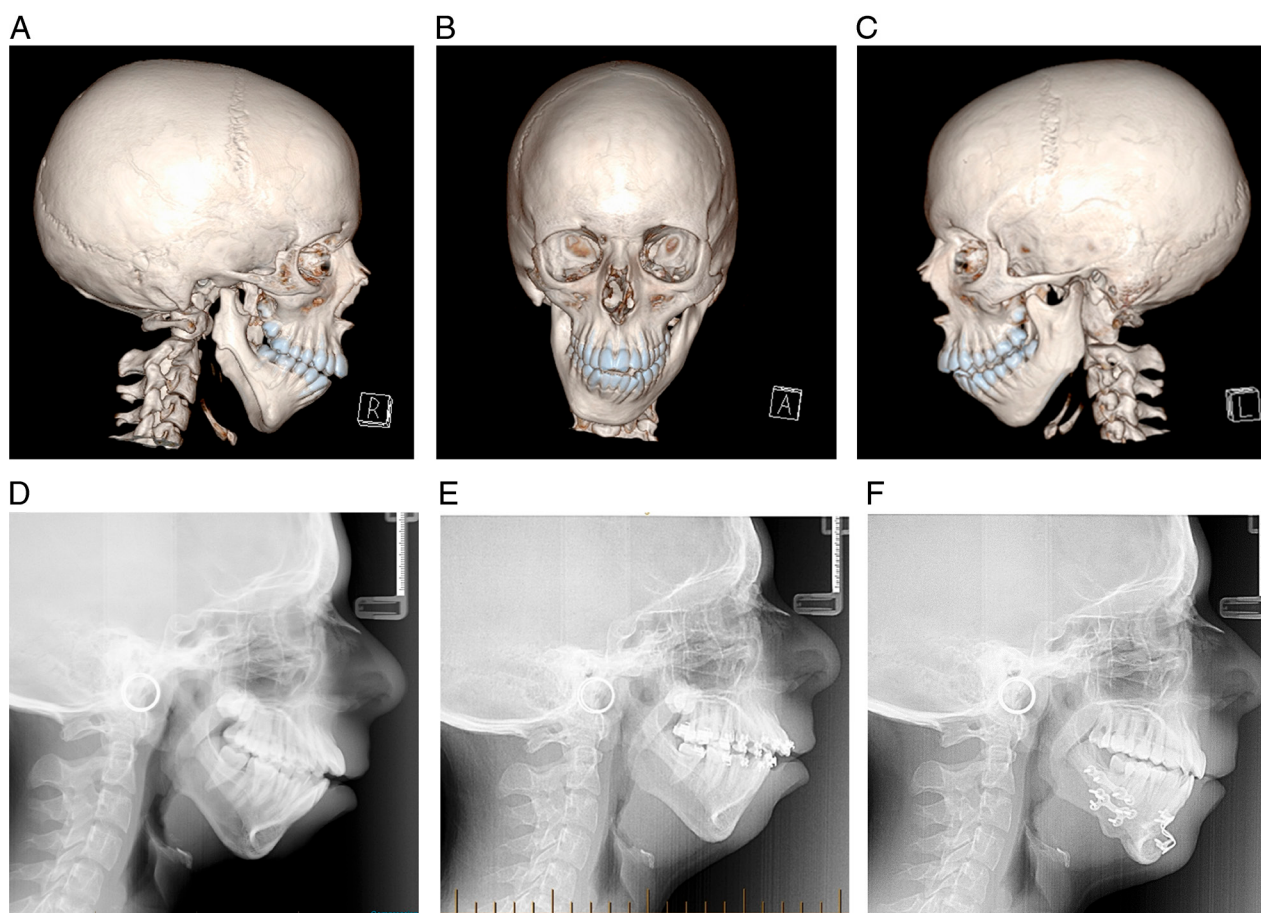


Figure 4. Pre and post-surgical radiographs. (A) 3D imaging Right-side. (B) 3D imaging Frontal. (C) 3D imaging Left side (aged 15.5 years). (D) Pre orthodontic lateral cephalogram (aged 17.3 years). (E) Pre surgical lateral cephalogram (aged 19.2 years). (F) Post-surgical rotational bilateral sagittal split osteotomy (BSSO) advancement with genioplasty (aged 20.2 years of age).

removal of the upper 3rd molars (Figure 3C). Post-surgery interarch elastics were required to settle the occlusion on the left side and to maintain the centreline correction. The fixed appliances were removed (age 20.2 years) and vacuum-formed retainers inserted, initially to be worn full time and then after 4 months, night wear only (Figures 1E, 2E, 3D, 4F and Table IC).

Twelve months following orthodontic treatment, the patient (age 21.2 years) undertook revision surgery which included a right mandibular Medpor[®] onlay implant and right facial dermal fat graft to address the deficient soft tissue bulk on that side. In addition, a modification was made to the previous genioplasty due to a relapse in chin symmetry.

Treatment outcome and subsequent changes

Two years following the revision surgery (aged 23.4 years) the patient expressed positive comments

regarding the treatment outcome but did raise concerns relating to the development of an openbite and a return of a minor soft tissue deficiency on the right side. The arch alignment was maintained by good retainer wear, but there was an anterior openbite on the left side that extended from the maxillary left central incisor to the maxillary left first molar. Although the midlines remained corrected there were signs of dentoalveolar compensation as the lower incisors angulated to the left along with relapse of the mandibular occlusal cant on the left side (Figures 1F, 2F).

Discussion

The case presentation prompts discussion regarding the treatment planning dilemmas in which patients present with a facial asymmetry well before skeletal maturity has been achieved and prior to the possibility of definitive skeletal surgery.

Longitudinal facial growth is an important variable in the MDT decision-making process for the timing and sequence of treatment with integrity of the glenoid fossa, condylar head and ascending ramus taken into consideration.¹⁶ However, the key determinant of MDT management relates to the functional status of the TMJ. Those with a Kaban Type I or IIa deformity with a functioning TMJ do not usually require TMJ reconstruction and this allows for mandibular reconstruction through a ramus osteotomy (+/- genioplasty). Differing surgical procedures depend upon OMENS characteristics and can include surgical correction of skeletal and soft tissue asymmetry as well as auricular anomalies. Mandibular skeletal surgery is often timed once most of the facial growth has ceased and can be carried out with conventional surgery, although distraction of the mandible is possible if there is a sufficient bone volume for distractor placement and large movements are required.¹⁷ At times, maxillary surgery is also undertaken when compensatory changes have occurred at the maxillary level.¹⁸

Earlier orthodontic intervention employing asymmetric functional appliances while in the mixed dentition, remains controversial.¹⁹ There is speculation that reduced mandibular growth leads to vertical maxillary deficiencies on the ipsilateral side producing an occlusal cant, buccal openbites and increased mandibular asymmetry. When undertaken, the goal of asymmetric functional appliance treatment often includes centring the mandible, maximising the growth potential of the maxilla and mandible on the affected side and limiting the amount of compensatory dentoalveolar change experienced.¹⁹ However, this treatment has a high burden of care due to unpredictable outcomes and, often, an extended duration of the functional appliance wear is required to achieve and maintain the desired changes.²⁰

The correction of occlusal cants by differential levelling using fixed appliances and/or surgical interventions can be challenging, as can pre-surgical arch co-ordination in which asymmetrical skeletal movements are required. While virtual surgical planning has allowed greater precision of planned surgical movements to be undertaken, arch co-ordination to achieve a well interdigitated occlusion can be challenging when often post-surgical orthodontic movements are also required to enhance overall stability.¹⁶

Table 1. Lateral cephalogram analysis: (A) Pre-treatment, (B) Pre-surgery, (C) Post treatment

Variable	A	B	C
SNA (°)	78.2	78.9	79.2
SNB (°)	67.5	68.9	72.9
ANB (°)	10.7	10.0	6.3
Wits (mm)	+2.3	+1.1	-1.4
MMA (°)	44.0	44.8	39.4
UI/Max (°)	121.9	119.1	110.8
LI/Man (°)	96.2	84.2	84.9
UI/LI (°)	106.5	101.9	128.4
LFH (%)	58.5	60.2	58.6
Holdaway (°)	25.3	31.6	20.5

A revision genioplasty and soft tissue augmentation surgeries including Medpor[®] onlays and dermal fat grafting highlight the requirement for additional corrective surgery following the mandibular advancement procedure to achieve desired aesthetic outcomes.²¹ The need to carry out a realignment genioplasty may suggest that the patient experienced further mandibular growth although the return of the occlusal cant to the lower arch was likely related to shortening of the LHS condylar/ramus due to remodelling and/or dental relapse.²² The dental arches were levelled prior to surgery using 0.019 x 0.022-inch stainless steel arch wires and vertical elastics were required post-surgery to settle the occlusion, particularly on the left side. However, complete closure of the RHS open bite post-surgery was not fully achieved and remains. Additional orthodontic treatment in the future and the possible use of temporary anchorage devices (TADS) for vertical anchorage may be required to re-establish occlusal contact with on-going retention supervision. This will be dependent upon the patient's wishes and any further deterioration in the openbite.

A recent Finnish study has highlighted the increased need for psychiatric support in both childhood and adolescence as well as in adults affected by craniofacial microsomia.²³ Many patients have hearing and speech impairments accompanying the craniofacial malformations which can trigger social biases associated with poor acceptance by peers.¹⁴ This can lead to complicated social interactions including those with teachers and other caregivers.²⁴

This case report illustrates some of the psychological issues experienced by an adolescent while waiting for facial growth to cease before undertaking definitive corrective treatment.

Conclusions

The present case report illustrates the complexity of the orthodontic and surgical management of facial asymmetry, the psychosocial considerations of the patient and the risk of relapse. The longitudinal nature of the patient's dental records provides an insight into the orthodontic considerations for a patient presenting with facial asymmetry and anticipation of potential relapse. Like other craniofacial anomalies, the present case report highlights the requirement for multidisciplinary care in the management of Goldenhar Syndrome.

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Conflict of interest

The authors declare that there is no conflict of interest.

Declaration of conflicting interests

The authors declare no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

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