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Case Report

Modification of Dentofacial Growth Associated with Goldenhar Syndrome

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The rare developmental defect, Goldenhar syndrome is characterized by complex craniofacial and dentofacial anomalies. Here we describe the successful orthodontic treatment of a 5-year-old Japanese Goldenhar syndrome patient with mild facial asymmetry, right microtia, right-side hearing loss, and tongue-thrusting by a modification of dentofacial growth using a non-surgical orthopedic treatment approach. Improvement of the vertical discrepancies on the affected side and canted occlusal plane as well as mandibular deviation were achieved with a functional orthopaedic approach. Stable and acceptable occlusion were obtained over the 32-month post-retention period. A non-surgical orthodontic treatment approach offers satisfactory facial aesthetic outcomes in Goldenhar syndrome.

Key words: Goldenhar syndrome, orthodontic treatment, functional appliance

Goldenhar syndrome (hemifacial macrosomia or HFM; OMIM® [Online Mendelian Inheritance in Man] #164210) is a rare congenital defect characterized by complex craniofacial abnormalities associated with a broad spectrum of defects such as vertebral, cardiac, renal, central nervous system and gastrointestinal malformations [1,2]. Goldenhar syndrome is also known as oculo-auriculovertebral syndrome (OAVS), arising from defects in the first and second branchial arches of the first pharyngeal pouch, the first branchial cleft and the primordia of the temporal bone [1,2]. The incidence of this disorder ranges from 1:3,500 to 1:5,600 live births, with a male-to-female ratio of 3:2 [3,4].

Although the majority of Goldenhar syndrome cases (85%) are unilateral, bilateral anomalies are found in 10-33% of the cases [1,5]. The right side is more frequently affected than the left, at a ratio of 3:2 [6]. Regarding the pathogenesis of the syndrome, most cases are sporadic, but autosomal dominant, autosomal recessive, and multifactorial modes of inheritance have been suggested [7-11].

The most recognizable features of Goldenhar syndrome is the presence of facial abnormalities. This symptom varies from mild asymmetry of the face to a severely underdeveloped facial half with orbital anomalies [5]. Microtia and auricular tags are found in 100% of the cases. Approximately 50% of the cases have combined conductive and sensorineural hearing loss [12]. One of the most common craniofacial defects in Goldenhar syndrome is unilateral hypoplasia of the mandible on the affected side. This syndrome also affects dentofacial structures, inducing features such as a cleft lip and palate, tongue cleft, unilateral tongue hypoplasia, a highly arched palate, hypoplasia of the maxillary and mandibular arches, micrognathia, gingival hypertrophy, supernumerary teeth, enamel and dentine malformations, and delayed tooth development. Some patients exhibit hypoplastic development

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of the facial expression muscles and asymmetric development of the mastication muscles on the affected side. Agenesis of the salivary glands or salivary fistulas and velopharyngeal insufficiency have also been observed [1, 2, 13-15].

Treatment for the craniofacial and dentofacial anomalies associated with Goldenhar syndrome greatly depends on the severity of the deformity and the needs of the individual patient. In less severe cases, growth modification treatment using functional appliances has been reported to provide better functional and aesthetic outcomes. A functional appliance is used for growth modification procedures that are aimed at intercepting and treating jaw discrepancies. Although the major effect of a functional appliance is dentoalveolar changes, the reported mandibular skeletal changes are from 1 to 2 mm [16].

Functional appliance therapy enhances growth modification and the stretching of soft tissues, which leads to improvements in mandibular morphology and facial form [17]. Such therapy can also restore the function of the temporomandibular joint (TMJ) and the growth capacity of the condyle [18]. However, there are few reports regarding solitary non-surgical treatments of patients with Goldenhar syndrome.

In this case report, we describe a successful orthodontic treatment via a noninvasive approach in a Goldenhar syndrome patient who demonstrated mild facial asymmetry with a straight facial profile, canted occlusal plane, microtia and a tongue-thrusting habit.

Case Report

A Japanese boy (5 years and 4 months old) who had been diagnosed with Goldenhar syndrome was referred to the outpatient dental clinic of Okayama University Hospital. The patient's chief complaint was an asymmetric facial profile. Pretreatment facial photographs showed right microtia and mild facial asymmetry from the frontal aspect, with a straight profile from the lateral aspect. He also had unilateral hearing loss, on the right side. The mandible was deviated to the right side. The right ramus was reduced in height compared to the left side, but normal morphologic characteristics of the ramus were present (Fig. 1A). The condyle, sigmoid notch and glenoid fossa were normal in size and shape with a functioning TMJ and no significant symptoms of TMJ disorder.

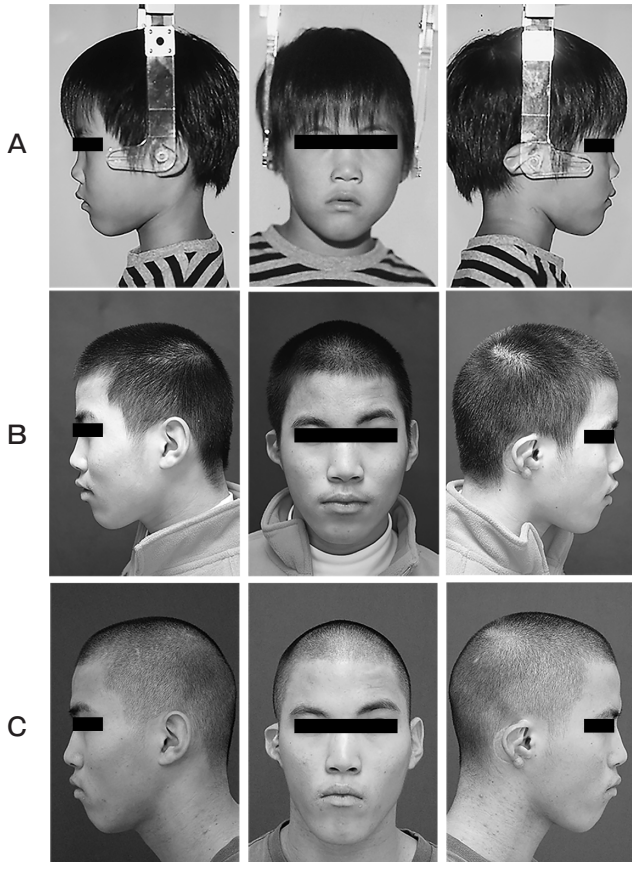

Fig. 1 Facial photographs. A, Pretreatment (5 years, 4 months.); B, Post-treatment (14 years, 9 months.); C, Post-retention (17 years, 6 months.).

Based on these findings, we suspected that the patient's case should be classified as the grade I category of the Pruzansky-Koban classification [19]. Intraoral photographs showed that the maxillary dental midline almost coincided with the facial midline; however, the mandibular midline was shifted 3.0 mm to the right (Fig. 2A). A unilateral crossbite combined with anterior and posterior crossbite were also seen on the right side in the intercuspal position. Decreased overbites of −1.6 mm with Angle Class I molar relationships on both sides were observed. A dental panoramic tomogram confirmed all of the permanent teeth except the third molars (Fig. 3A).

A cephalometric analysis of the patient showed a skeletal Class I jaw relationship: A point-nasion-B point angle (ANB), 5.4°; sella-nasion-A point angle (SNA), 84.7°; sella-nasion-B point angle (SNB), 79.3°. The analysis also revealed a trend toward a high mandibular plane angle (Frankfort mandibular-plane angle

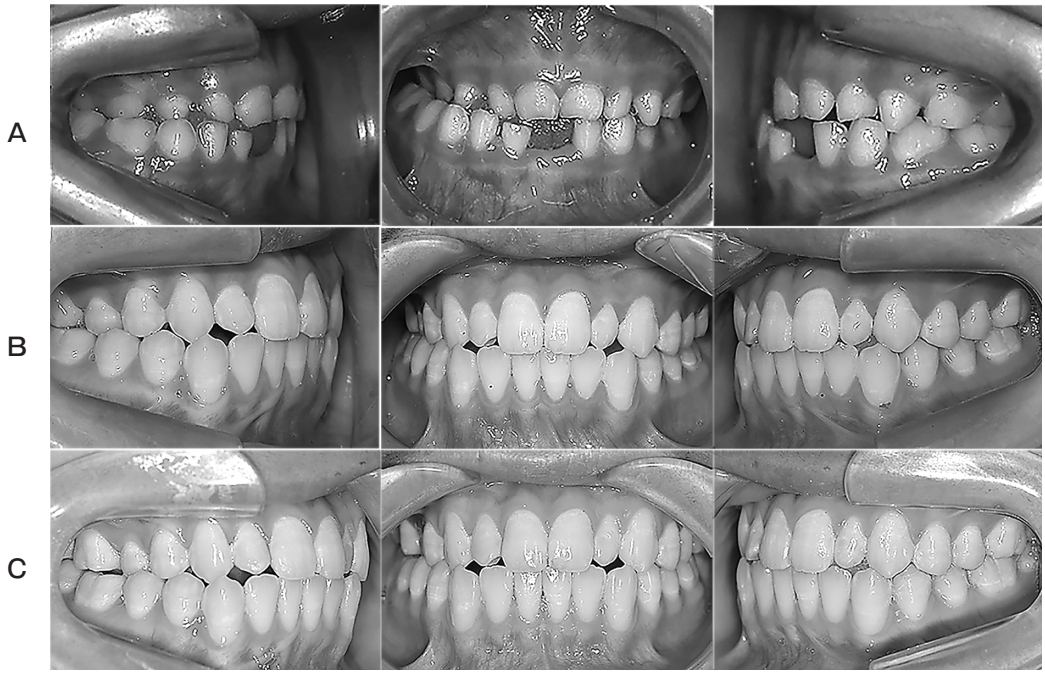

Fig. 2 Intraoral photographs. A, Pretreatment (5 years, 4 months.); B, Post-treatment (14 years, 9 months.); C, Post-retention (17 years, 6 months.).

[FMA], 36.0°) compared with Japanese norms [20]. Although the maxillary incisors were lingually inclined, the mandibular incisor angle was within normal ranges (U1-FH, 100.7°; L1-Mp, 88.0°) (Fig. 4A, Table 1). Posteroanterior cephalometric tracing demonstrated that the lack of vertical development on the affected side had produced a canted occlusal plane (Fig. 5A). The patient showed no significant symptoms of TMJ dysfunction.

Based on these findings, we diagnosed the patient with skeletal Class I, Angle Class I malocclusion, a high-mandibular-plane-angle, facial asymmetry caused by Goldenhar syndrome. The objectives for the treatment were to correct the transverse and vertical skeletal discrepancies, improve the facial aesthetics, correct the dental midline discrepancy due to the deviated mandible, and create functional and aesthetic occlusion.

Subsequent to the first stage of auricular reconstruction (Fig. 6A, B), an expansion plate was placed in the maxillary arch to improve the patient's narrowed maxillary arch for 4 months. When the patient reached 6 years and 9 months of age, we used a functional appliance (Klammt's Elastic Open Activator) with posterior bite capping on the left side to improve the transverse jaw-base relationship and to correct the vertical

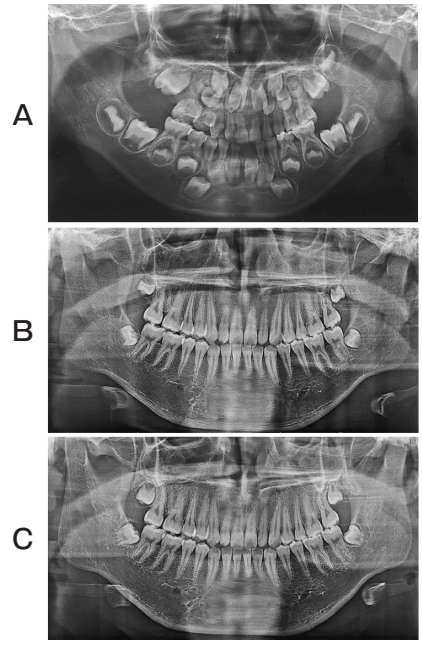

Fig. 3 Panoramic radiographs. A, Pretreatment (5 years, 4 months.); B, Post-treatment (14 years, 9 months.); C, Post-retention (17 years, 6 months.).

dentoalveolar growth for 64 months (Fig. 6C). After the functional appliance treatment, the patient exhibited a

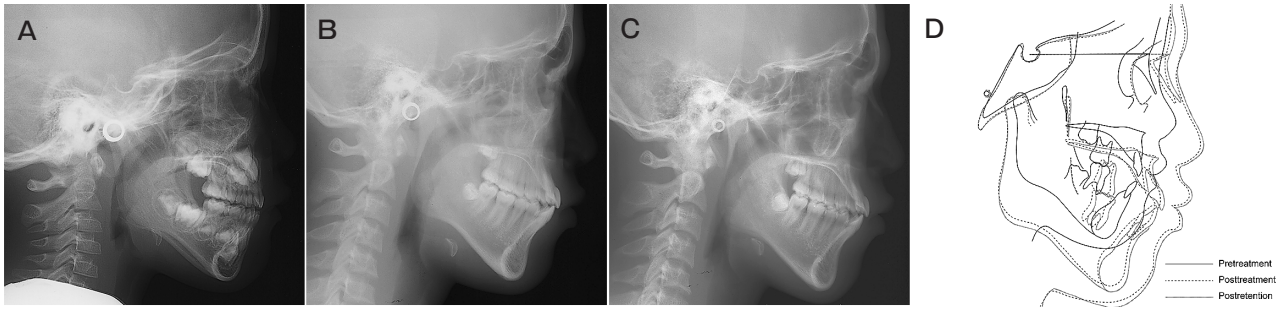

Fig. 4 Cephalometric radiographs. **A**, Pretreatment (5 years, 4 months.); **B**, Post-treatment (14 years, 9 months.); **C**, Post-retention (17 years, 6 months.); **D**, Superimposed cephalometric tracings show the changes from the pretreatment to post-retention stages.

Table 1 The patient's cephalometric measurements at the 3 treatment stages

Variables	Japanese norms (6-year-old male)	S.D.	Pre-treatment	Japanese norms (Adult male)	S.D.	Post-treatment	Post-retention
Angular (°)							
ANB	4.7	1.8	5.4	3.2	2.4	2.2	2.7
SNA	81.6	3.1	84.7	81.5	3.3	78.7	79.1
SNB	76.8	2.7	79.3	78.2	4.0	78.7	79.1
FMA	31.7	4.9	36.0	28.0	6.1	30.6	30.4
U1-FH	100.7	1.7	100.7	112.4	7.6	115.4	115.5
L1-Mp	89.0	6.4	88.0	95.2	6.2	94.2	93.1
Interincisal angle	141.7	5.1	140.5	124.2	8.6	119.9	120.9
Occ. Plane to SN	22.7	3.0	22.4	15.5	4.2	15.8	15.6
Gonial angle	128.4	4.4	131.5	120.9	6.5	121.0	120.9
Linear (mm)							
S-N	63.3	2.3	73.5	72.2	3.3	83.7	85.5
N-Me	107.5	3.8	115.9	135.7	4.0	145.3	149.5
Me/PP	58.8	3.2	65.5	74.6	3.0	82.1	84.6
Go-Me	58.2	2.7	61.7	76.6	4.4	80.3	81.9
Ar-Me	89.3	3.4	97.0	115.6	6.8	122.8	126.9
Ar-Go	39.2	2.8	43.3	53.2	5.7	57.2	60.6
Overjet	1.5	2.1	0.5	3.3	1.0	2.9	3.1
Overbite	1.2	1.9	−1.6	3.3	1.7	0.8	1.1
U1/PP	26.8	1.5	27.1	32.4	3.1	30.3	30.2
U6/PP	15.8	1.9	20.8	26.2	1.9	26.2	26.1
L1/Mp	37.5	1.8	41.8	48.9	2.6	56.5	58.8
L6/Mp	28.5	1.3	35.8	37.5	2.1	44.8	47.3
E-line to Upper lip			2.8			4.9	5.2
E-line to Lower lip			1.6			5.4	5.9

slight open bite due to a tongue-thrusting habit. A tongue crib was placed in the maxillary arch to restrain the tongue in a posterior position for 32 months (Fig. 6D). After the appliances were removed, a modified wraparound-type retainer with a tongue guard was placed in the upper arch.

The orthodontic treatment improved the patient's skeletal discrepancy and occlusal relationships. The post-treatment photographs (when the patient was approx. 15 years old) showed a more symmetrical appearance than that observed before treatment (Fig. 1B). Acceptable occlusion with Class I molar and canine relationships had also been established. The mandibular midline almost coincided with the maxillary midline, and the unilateral crossbite on the right side was improved, except for the right second bicuspid

(Fig. 2B).

A post-treatment panoramic radiograph confirmed good root parallelism, with no apparent root resorption (Fig. 3B). A post-treatment cephalometric analysis showed a 3.2° decrease in the ANB angle. The occlusal plane angle and the FMA angle were decreased by 6.6° and 5.4°, respectively (Fig. 4B). Posteroanterior cephalometric tracing confirmed improvements in the vertical discrepancies on the affected side and the canted occlusal plane (Fig. 5B). Both acceptable occlusion and facial aesthetics were maintained over the 32-month post-retention period (Figs. 1C, 2C, 3C, 4C, and 5C).

Discussion

We have reported the successful orthodontic treatment of a patient with Goldenhar syndrome showing mild facial asymmetry. The therapeutic method proposed for this patient involved myofunctional appliance therapy to achieve satisfactory functional, structural and aesthetics outcomes. This therapy is thought to restore condylar growth and improve the masticatory muscle function in mildly to moderately affected patients during the early stages of growth [21, 22]. A patient's cooperation and the treatment timing are quite important for achieving a successful outcome. Previous studies have shown that a growth modification approach using a functional appliance enables the reso-

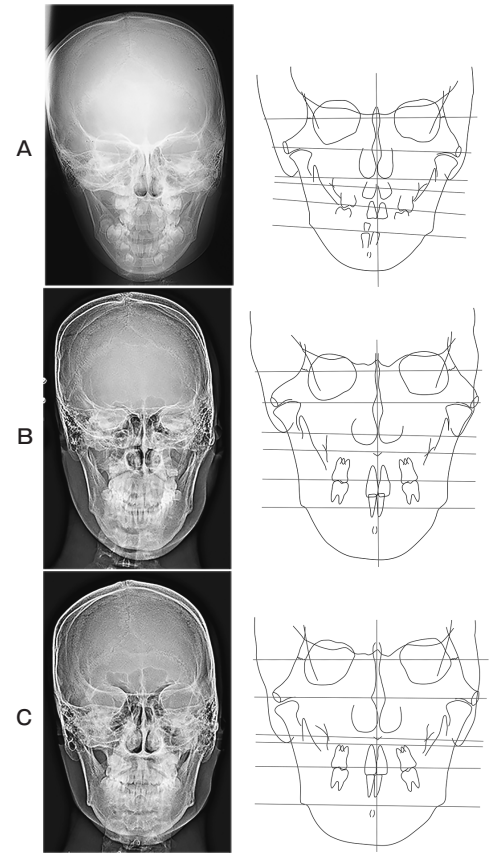

Fig. 5 Posteroanterior cephalometric radiographs and tracings. **A**, Pretreatment (5 years, 4 months.); **B**, Post-treatment (14 years, 9 months.); **C**, Post-retention (17 years, 6 months.).

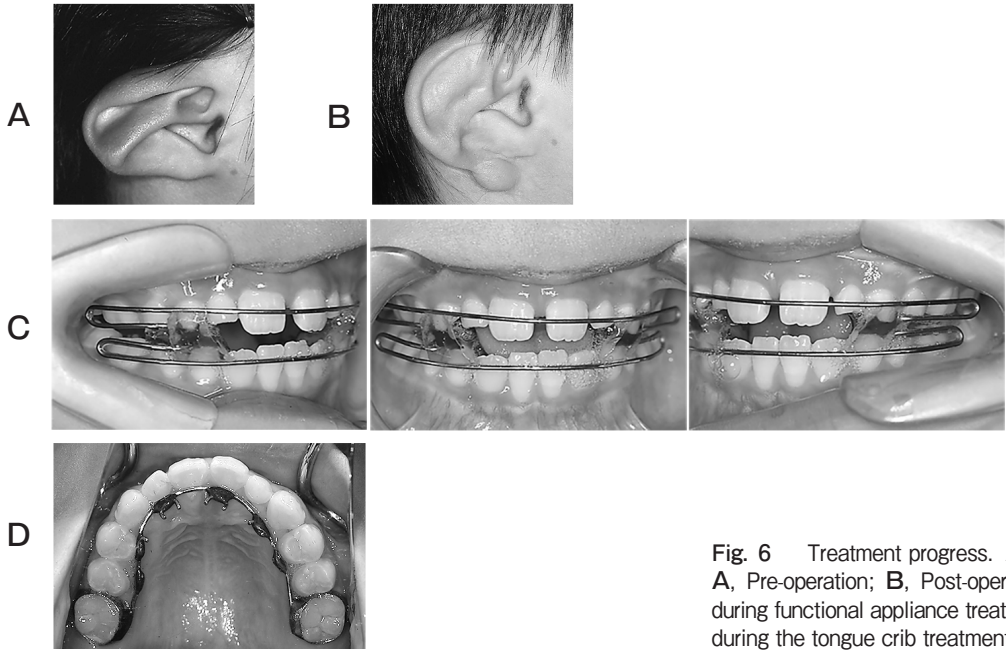

Fig. 6 Treatment progress. **A**, **B**, Auricular reconstruction. **A**, Pre-operation; **B**, Post-operation; **C**, Intraoral photographs during functional appliance treatment; **D**, Intraoral photographs during the tongue crib treatment.

lution of mainly dental asymmetry, thus accepting some degree of skeletal disharmony [18, 23]. In the present case, the dental midline was improved, and the occlusal plane was almost levelled by displacing the jaws and reinforcing dentoalveolar adaptation. These results supported the findings of Sarnäs *et al.*, who achieved the correction of dental malocclusion by dentoalveolar remodeling to some extent [24]. Importantly, the patient and his family experienced markedly positive outcomes with this treatment, although slight facial asymmetry still exists.

Orthognathic surgery with distraction osteogenesis is the treatment of choice for Goldenhar syndrome to correct the transverse skeletal discrepancies, but such treatment modalities depend on the severity of the deformities. Although it was reported that orthognathic surgery is required for severe asymmetry cases [25], long-term reports of mandibular distraction osteogenesis still describe controversial results because of delays in growth and the development of the affected area [26]. In the present case, the patient and his family wanted to avoid orthognathic surgery because of the risk of this approach.

Goldenhar syndrome has a multifactorial etiology that has not yet been fully established. The ingestion of some drugs by the mother during pregnancy (e.g., cocaine, thalidomide, retinoic acid, and tamoxifen) have also been suggested as etiologic factors [8, 27]. Maternal diabetes and infections caused by rubella and influenza during pregnancy may also be related to the development of this syndrome [6, 28]. Ipsilateral hypoplasia of the face and external ear deformities are the characteristic clinical hallmarks of Goldenhar syndrome. Regarding the extraoral clinical findings, our patient showed facial asymmetry and ear anomalies on the right side. Although mandibular hypoplasia is usually seen in patients with Goldenhar syndrome, a straight facial profile with Class I malocclusion was observed in our patient.

Ueki *et al.* claimed that the incidence of morphological changes and internal derangement with TMJ disorder are higher on the deviated side than on the non-deviated side, and a high rate of disk displacement and symptoms of TMJ disorder have also been found on the deviated side of asymmetric patients [29, 30]. An evaluation of the present patient's jaw movement showed no obvious symptoms of temporomandibular joint dysfunction before or after treatment.

In addition to the above facts, a multidisciplinary approach is also important for patients with Goldenhar syndrome. Reconstructive surgery is necessary to correct structural defects such as those of the ears and soft tissues, to minimize aesthetic concerns and improve the patient's quality of life. Because of the complex nature of our patient's general and oral health problems, regular dental care such as scaling and caries prevention (including fluoride application and a professional tooth cleansing program) is needed as a preventive strategy. Our patient's favorable results were quite stable after the 32-month post-retention period, and he and his family were satisfied with the results. However, long-term follow-up and a longitudinal assessment and growth analysis are required to confirm the efficacy of this treatment protocol for patients with Goldenhar syndrome.

In conclusion, we herein reported the successive orthodontic treatment of a male patient with Goldenhar syndrome presenting with mild facial asymmetry, congenital ear deformity, and unilateral hearing loss on the right side. Although conservative treatment has limited applicability in patients with Goldenhar syndrome, our results suggest that the use of functional appliances may greatly contribute to the restoration of typical growth.

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