

# Comprehensive prosthetic rehabilitation of a case of the orofacial digital syndrome

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## Abstract

Maxillofacial defects can result from trauma, treatment of neoplasm, or congenital malformations. Many a time due to the size, location of the defect, or because of the patient's medical condition, surgical reconstruction may not be possible. In these cases, rehabilitation is indicated with the help of a maxillofacial prosthesis. Orofacial digital syndrome (OFDS) is a congenital anomaly that affects the development of the mouth, face, and digits. Such abnormalities or defects compromise form, function, esthetics, and social acceptance and deeply affect the psychological status of an individual. This report describes the comprehensive prosthetic rehabilitation of a case of OFDS with bilateral cleft lip and palate as well as syndactyly and brachydactyly of the foot. The individual was rehabilitated with a definitive obturator and a custom-made foot prosthesis using room-temperature-vulcanizing silicone. The prosthetic rehabilitation significantly improved esthetics, phonetics, and function as well as social and psychological status of the patient.

**Keywords:** Foot prosthesis, maxillofacial prosthetics, obturator, orofacial digital syndrome, prosthodontic rehabilitation

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## INTRODUCTION

The orofacial digital syndrome (OFDS) is a genetic anomaly presenting characteristic developmental disorders of the face, oral cavity, and digits.<sup>[1]</sup> Additional signs may or may not involve the central nervous system (CNS) and visceral organs, such as the kidney. The first case presenting this condition was reported in 1941,<sup>[2]</sup> since then, a number of different OFDS types with overlapping phenotypes have been described.<sup>[3]</sup> The characteristic intraoral features are cleft of the upper lip and palate, absent/multiple labial frenum, oligodontia, malalignment, and other tooth anomalies.<sup>[4]</sup> Mental retardation and other CNS disorders are

seen in 30%–50% of cases.<sup>[5]</sup> Variable digital malformations have been observed that include clinodactyly, syndactyly, brachydactyly, and polydactyly.<sup>[6]</sup> Facial features include asymmetry frontal bossing, hypertelorism, broadened nasal bridge, low set ears, and altered alar base morphology.<sup>[7]</sup> Among the different types, OFDS Type I is the most frequently observed and can be easily recognized by its typical X-linked dominant male-lethal pattern of inheritance in familial cases.<sup>[8]</sup> Most of the other OFDS are transmitted as autosomal recessive syndromes or represent sporadic cases. This article describes the intraoral and extraoral rehabilitation of one such situation of OFD Type I treated with an interdisciplinary approach.

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## CASE REPORT

A 26-year-old girl reported with the complaint of dripping of fluids from the nose, difficulty in chewing food, and unesthetic appearance due to multiple missing teeth [Figure 1]. The past medical history revealed an operated case of bilateral cleft lip and palate. Multiple surgical interventions were carried out in the past for the correction of lip, palate, and nose defects. The past dental history included orthodontic treatment for 5 years and extraction of two maxillary posterior teeth. On general examination, the patient was moderately built and well oriented. On examination of the digits, the index finger on the left hand had clinodactyly and the left foot digits presented with brachydactyly as well as syndactyly of the fourth and fifth digits [Figure 2]. The right foot presented with clinodactyly of all digits [Figure 2]. On extraoral examination, the patient had an asymmetric face in vertical thirds and horizontal fifths with midface deficiency [Figure 2]. Other features noted were frontal bossing, hypertelorism, flattened nasal bridge, altered alar base morphology, scar in the philtrum region, and a concave profile. Intraoral examination revealed multiple missing teeth in the maxillary arch and a defect of the anterior hard palate (10 mm × 12 mm) in dimensions. The mucosal lining of the hard palate was irregular or corrugated. Family history revealed that the parents from a non-consanguineous marriage were healthy, had two girls, and a history of one miscarriage in the second trimester. Cone-beam computed tomography, radiograph foot, karyotyping, and gene sequence analysis were performed. A radiograph revealed multiple missing teeth, impacted maxillary right third molar, and mandibular left third molar. It also showed deficient bone in the maxillary anterior region with oronasal communication. Radiograph of the



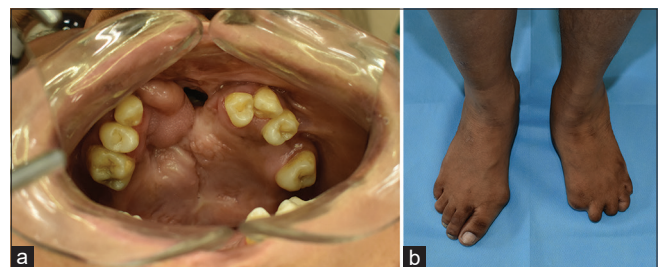
**Figure 1:** Frontal view showing facial asymmetry with midface deficiency, frontal bossing, hypertelorism, broad nasal bridge, altered alar base morphology, and scar tissue at the philtrum region

foot presented complete coalescence of metatarsals in all digits. Karyotyping was normal and gene sequence analysis revealed mutation at short arm of X chromosome at Xp22.23 site. A diagnosis of OFDS (Type – I) was made.<sup>[9]</sup> In this case, since the surgical and orthodontic intervention had already been done, prosthetic rehabilitation was taken up. It was planned in three phases: Phase I – fabrication of interim obturator, Phase II – fabrication of definitive obturator, and Phase III – fabrication of foot prosthesis.

### Treatment procedure

In the Phase I, an interim obturator was fabricated. Primary impressions of the maxillary and mandibular arch were made with irreversible hydrocolloid (Algitec, DPI), and the casts were poured using Type III dental stone (Kalabhai). Jaw relation, teeth arrangement, and try-in were completed. All conventional clinical and laboratory procedures were followed. The finished interim obturator was inserted *in situ* and evaluated for retention, support, stability, esthetics, and phonetics. The patient was called for follow-up for 2 months.

After patient compliance and motivation with Phase I, the next phase included fabrication of a definitive obturator. Primary impressions and casts were made similar to Phase I, and the maxillary cast was evaluated on the surveyor. Designing for the metal framework was done by marking height of contour, selection of components, maxillary complete palate major connector with modified design, occlusal rests on the maxillary posterior teeth, cast circumferential clasp as direct retainer, and cingulum rest on the maxillary left canine providing indirect retention. Teeth preparation was completed as planned and a final impression was made using a two-step polyvinyl impression technique (Affinis, Coltene) and model was retrieved. Block out of undercuts was completed on master cast which was then duplicated using reversible hydrocolloid. A refractory cast was poured using phosphate-bonded investment material (Degugest). Designing of metal framework was completed using wax. Spruing, investing,



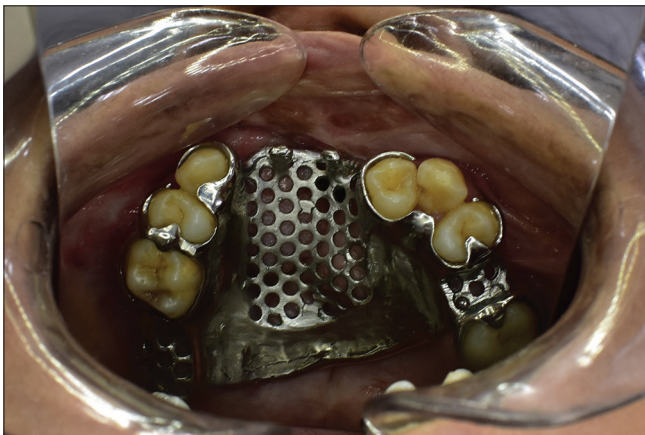
**Figure 2:** (a) Intraoral view showing defect in the anterior region of the maxilla, multiple missing teeth, malaligned teeth, and altered mucosal lining of the hard palate. (b) Defects of digits showing clinodactyly, syndactyly, and brachydactyly



burnout, and casting were completed to fabricate metal framework. The framework was retrieved, finished, and polished. Try-in was done to evaluate for fit, retention, support, and stability [Figure 3]. Jaw relation recording, teeth arrangement, and try-in were completed, and all conventional laboratory procedures were followed to fabricate a definitive obturator. The prosthesis was inserted *in situ* and evaluated for retention, stability, esthetics, and phonetics. The definitive obturator significantly improved form, function, esthetics, and the confidence of the patient [Figure 4].

After rehabilitating the intraoral defect, in the Phase III, the patient was rehabilitated with a foot prosthesis. An impression of the affected foot was made with a modified box technique using an irreversible hydrocolloid (Algitek) and working model was poured using dental stone (Kalabhai). The impression of the contralateral healthy foot was made by fabricating a custom tray with impression compound (DPI), and an impression was made using irreversible hydrocolloid

material (Algitek, DPI). Modeling wax (Ruthinium Dental products) was heated in a wax bath and was poured into the impression to fabricate the wax pattern of the foot. The wax pattern was split into individual digits which were then reassembled on the working model to complete the wax pattern of the planned prosthesis [Figure 5]. Flasking of the wax pattern was done using a multi-pour method with orientation grooves at each pour using Type II and Type III gypsum product (Kalabhai). The assembly was placed in a warm water bath for dewaxing and a four-piece mold was obtained which helped in the fabrication of prosthesis with different shades on the dorsal and plantar surface [Figure 6]. Room-temperature-vulcanizing (RTV) silicone (Copsil T – 30 TN, COP) was used for the fabrication of prosthesis. A nylon stocking was placed on the working model over which the complete prosthesis was fabricated [Figure 7]. The nylon stocking not only eased the use of prosthesis but also the microroughness of the surface provided the retention to prosthetic material. Silicone was mixed with intrinsic colors following the manufacturer's instructions to closely match the shade of the patient [Figure 7]. After packing of silicone on the dorsal surface and ventral surface of the mold, the complete flask assembly was closed and left at



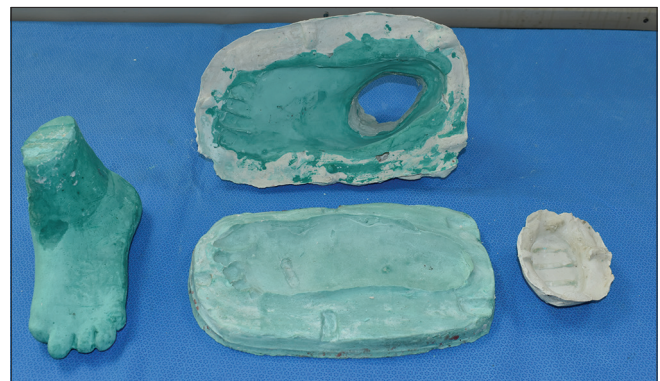
**Figure 3:** Try-in metal framework



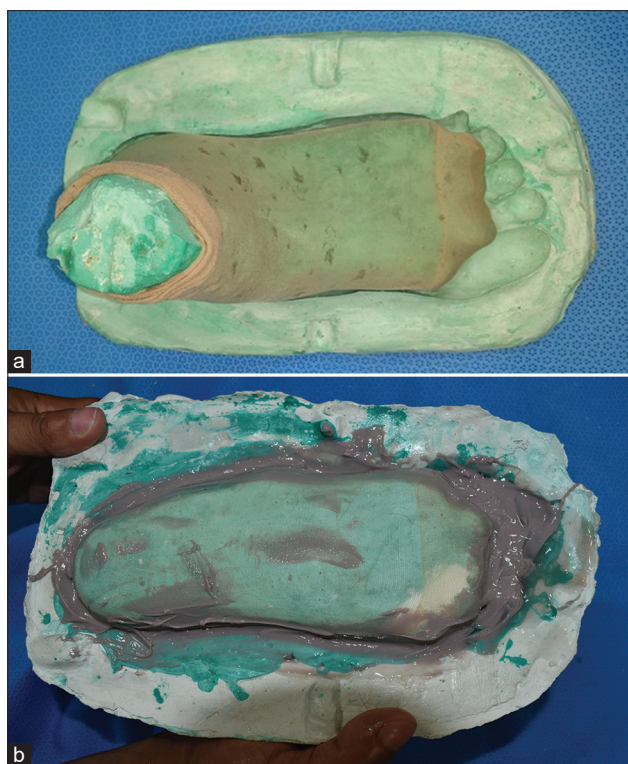
**Figure 4:** Postoperative view after rehabilitation with definitive obturator



**Figure 5:** Wax pattern of the foot prosthesis



**Figure 6:** Four-piece mold for fabrication of foot prosthesis



**Figure 7:** (a) Nylon stocking placed over the working model on which complete prosthesis was fabricated. (b) Packing of silicone after shade matching

room temperature for 24 h to complete curing. Putty index (Flexceed, GC) of nails of the patient's contralateral foot was taken and custom-shaped nails were fabricated using autopolymerizing polymethylmethacrylate (DPI). The prosthesis was retrieved, finished, characterized using extrinsic colors, and custom-made nails were attached using silicone adhesive (G611, Technovent) [Figure 7]. The prosthesis was inserted *in situ* and was evaluated for fit, esthetics, and gait of patient [Figure 8]. A comprehensive prosthetic rehabilitation not only improved form, function, and esthetics but also remarkably improved the confidence, psychological attitude, and social acceptance of the patient.

## DISCUSSION

Multiple variants of OFDS are reported in the literature, but OFDS-I has characteristic prevalence only in females as it is lethal in males; similar observation was found in this case. The intraoral defect was rehabilitated with an interim obturator followed by the definitive obturator. In this case, a modified complete palate maxillary major connector with mesh framework was incorporated in the anterior region to allow for alterations or relining. Various prosthetic materials such as acrylic resins, acrylic copolymers, vinyl polymers, polyurethane elastomers, and silicone elastomers have been reported in the literature.<sup>[10]</sup>



**Figure 8:** Postoperative view after prosthetic rehabilitation with custom-made foot prosthesis

Silicone became popular over other materials as they have a range of good physical properties, low degree of toxicity, easier to manipulate, chemical inertness, and high degree of thermal and oxidative stability.<sup>[11]</sup> Further, they can be stained intrinsically or extrinsically to give them more lifelike natural appearance. In this case, digit defect of the foot was rehabilitated with foot prosthesis fabricated with RTV silicone with customization of form, shape, and shade. The follow-up visit should be 24 h after prosthesis delivery. At that time, the condition of the prosthesis and the health of skin should be assessed. Subsequent visits are planned at 1 week and then every month to evaluate fit, color, function, and retention of prosthesis and also to examine the health of the underlying skin. Maintaining hygiene of the prosthesis is important for the health of soft tissue underneath the prosthesis and for preserving the prosthesis itself in a good condition. Silicone materials are more difficult to clean than resins as these materials are permeable so are more susceptible to microbial colonization. Water and neutral soap, together with gentle brushing using soft nylon bristles, have been recommended. The use of chlorhexidine has been shown as an excellent method of disinfection; in this case, prosthesis was cleaned using 4% chlorhexidine immersion for 1 min everyday followed by rinse with water. Multiple studies highlight that the use of disinfecting agents and rigorous cleaning adversely affects the physical properties of silicone material.<sup>[12]</sup>

## CONCLUSION

OFDS is a genetic disorder affecting the development of the oral cavity, face, and digits. Cleft palate was rehabilitated with a definitive obturator and digits defect was rehabilitated with a custom-made silicone foot prosthesis. The literature available for managing a situation of OFDS is very bleak;



the present clinical report is an attempt to improve the overall quality of life of such patients.

#### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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#### Conflicts of interest

There are no conflicts of interest.

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