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Case Report

Maxillofacial Prosthetic Rehabilitation of Extensive Facial Defects in a Patient with Congenital Erythropoietic Porphyria Using a Multicomponent Silicone Prosthesis: A Case Report

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ABSTRACT

Congenital Erythropoietic Porphyria (CEP), also known as Günther disease, is an exceptionally rare autosomal recessive disorder characterized by severe photosensitivity, recurrent blistering, ulceration, scarring, and progressive destruction of sun-exposed tissues. In advanced cases, extensive loss of facial structures can result in severe functional, esthetic, and psychosocial impairment. Maxillofacial prosthetic rehabilitation provides a conservative alternative when surgical reconstruction is limited by extensive tissue loss, compromised local tissues, or unpredictable healing. This case report describes the prosthetic rehabilitation of a 45-year-old male patient diagnosed with Congenital Erythropoietic Porphyria, presenting with extensive facial disfigurement involving the nasal, orbital, auricular, and adjacent facial regions. Following multidisciplinary medical evaluation and clearance, a comprehensive silicone-based maxillofacial rehabilitation was planned. Facial impressions were obtained using a reinforced irreversible hydrocolloid technique, followed by a definitive functional impression using addition silicone during facial movements. The definitive prosthesis was processed using medical-grade room-temperature vulcanizing silicone elastomer with intrinsic coloration and characterization. Retention was achieved using a water-soluble adhesive and spectacles. This case demonstrates the potential of a customized multicomponent silicone maxillofacial prosthesis in the rehabilitation of extensive facial defects associated with Congenital Erythropoietic Porphyria.

Keywords: *Congenital Erythropoietic Porphyria; Günther disease; Maxillofacial prosthesis; Facial prosthesis; Silicone elastomer; Facial rehabilitation; Auricular prosthesis; Ocular prosthesis*

INTRODUCTION:

Congenital Erythropoietic Porphyria (CEP), also known as Günther disease, is an exceptionally rare autosomal recessive disorder of heme biosynthesis caused by a deficiency of uroporphyrinogen III synthase. The resultant accumulation of type I porphyrins within tissues leads to severe photosensitivity, recurrent blistering, ulceration, progressive scarring, and mutilating deformities of sun-exposed tissues. Over time, repeated cycles of tissue destruction and healing may culminate in extensive resorption of facial structures, including the nose, auricles, periorbital tissues, and adjacent facial components, producing profound functional and esthetic impairment.

Beyond the physical manifestations, the severe facial disfigurement associated with CEP can have a substantial psychosocial impact, often resulting in social withdrawal, diminished self-esteem, and a markedly reduced quality of life. Although contemporary medical management may help limit disease progression and prevent secondary complications, restoration of lost facial structures remains a significant rehabilitative challenge. Surgical reconstruction is frequently constrained by extensive tissue loss, compromised tissue quality, recurrent scarring, and the unpredictable nature of the disease process. Consequently, maxillofacial prosthetic rehabilitation serves as a valuable treatment modality, offering a conservative, non-invasive, and esthetically acceptable alternative for restoring facial form and facilitating psychosocial reintegration.

Advances in maxillofacial materials and fabrication techniques have significantly enhanced the predictability and realism of facial prostheses. Medical-grade room-temperature vulcanizing (RTV) silicone elastomers provide excellent biocompatibility, flexibility, durability, and lifelike appearance. This report describes the comprehensive maxillofacial rehabilitation of a 45-year-old male patient with extensive facial disfigurement secondary to Congenital Erythropoietic Porphyria. A customized silicone prosthetic reconstruction was fabricated to restore facial esthetics, improve social confidence, and enhance the patient's overall quality of life.

PATIENT DETAILS:

A 45-year-old male patient presented to the Department of Prosthodontics, Crown and Bridge, with the chief complaint of severe facial disfigurement involving the nasal, orbital, auricular, and adjacent facial regions. The patient had been diagnosed with Congenital Erythropoietic Porphyria (CEP) seven years previously and had subsequently developed progressive resorptive destruction of facial tissues over the preceding five years. The disease process resulted in extensive loss of facial structures, causing significant facial deformity and profound psychosocial impairment, adversely affecting the patient's self-esteem, social interactions, and overall quality of life.

Clinical examination revealed severe mutilation of the midfacial and auricular regions, characterized by extensive tissue loss and distortion of normal facial anatomy. Following comprehensive clinical evaluation and multidisciplinary consultation with the Departments of Dermatology and General Medicine, the patient was deemed medically fit for prosthetic rehabilitation.

Considering the extent of tissue loss, compromised local tissues, and the limitations associated with surgical reconstruction, a comprehensive maxillofacial prosthetic rehabilitation was planned. The treatment objective was restoration of the missing facial structures through the fabrication of a custom-made silicone facial prosthetic system comprising nasal, orbital, auricular, and facial components. The proposed rehabilitation aimed to restore facial esthetics, improve functional outcomes, and facilitate psychosocial reintegration.

CLINICAL PROCEDURE:

Following acquisition of written consent, a comprehensive medical assessment was undertaken and appropriate medical clearance was obtained prior to commencement of prosthetic rehabilitation.

FACIAL IMPRESSION PROCEDURE:

Thorough debridement of crustations was done. The facial skin was lightly coated with petroleum jelly to facilitate atraumatic removal of the impression material. A facial impression framework was fabricated using impression compound (DPI, dental products of India, Mumbai, India) reinforced with modeling wax (DPI, dental products of India, Mumbai, India) and adapted over the facial contours (Fig 1). This framework served as a supporting matrix for the impression material and prevented excessive material flow during the impression procedure.

To ensure maintenance of a patent airway throughout impression making, a modified 3-mL disposable syringe was employed as a mouth-breathing aid. The patient was instructed and trained to breathe through the mouth before the procedure. The nasal defect site was gently packed with petroleum jelly–impregnated gauze to prevent ingress of impression material into the defect, while the contralateral normal eye was protected using adhesive tape.

A thin mix of irreversible hydrocolloid impression material (tropicalgin, zhermack, badia polesine, Italy) was applied over the defect area. Gauze strips were incorporated within the alginate to reinforce the impression, followed by application of impression plaster (Kalabhai Karson pvt. Ltd., Mumbai, India) as a backing material. Additional gauze reinforcement was incorporated to improve strength and dimensional stability. Following complete setting, the impression was carefully retrieved and poured in Type IV dental stone (Kalrock, Kalabhai Karson pvt. ltd., Mumbai, India) to obtain the primary facial cast (Fig 2).

DEFINITIVE FUNCTIONAL IMPRESSION:

In order to avoid dislodgement of prosthesis during functional movements, a functional impression was planned. A custom impression tray was fabricated on the primary cast using auto polymerizing acrylic resin (DPI RR cold cure, dental products of India, Mumbai, India). A definitive functional impression was subsequently made using addition silicone elastomeric

impression material (aquasil, denstsply sirona, charlotte, NC, USA) in putty and light-body consistencies. During impression making, the patient was instructed to perform a series of functional facial movements, including wide smiling and other dynamic expressions, to accurately record the functional contours of the surrounding tissues. This approach facilitated the fabrication of a prosthesis capable of maintaining adaptation and stability during facial movements. The definitive impression was poured in Type IV dental stone (Kalrock, Kalabhai Karson pvt. Ltd., Mumbai, India) to obtain the master cast (Fig 2).

DONOR FACIAL MOULAGE AND WAX PATTERN FABRICATION:
To reproduce a natural contour, a donor facial moulage was obtained from the patient's younger brother, who served as the donor. The donor impression was made using the same impression protocol described previously. Molten modelling wax was poured into the donor impression to obtain a wax replica of the component. The resulting wax form was subsequently modified and sculpted to harmonize with the patient's facial proportions and contours. Pre-defect facial photographs were used as references during wax pattern refinement to achieve optimal esthetic integration.

FABRICATION OF THE ORBITAL COMPONENT:
The orbital component was fabricated by first constructing an ocular prosthesis incorporating a three-dimensional (3D)-printed iris shell. The ocular prosthesis was accurately positioned on the master cast according to established anatomical landmarks. Subsequently, a wax pattern for the orbital prosthesis was carved around the ocular component to restore the lost periorbital tissues.

FABRICATION OF AURICULAR COMPONENT:
Impression of the defective, resorbed auricles were made by the same impression protocol we followed before. Casts were poured. Donor impression was made from patient's daughter, as the auricular component matched the patient. Molten modelling wax was poured into the impression and wax pattern was retrieved. It was then carved and modified to little extent to make it precise for the patient and adapted well on the patient's casts (Fig 3).

WAX TRIAL:
At the wax try-in appointment, the prosthesis was evaluated for adaptation, retention, marginal continuity, facial symmetry, and overall esthetic appearance. An acrylic nasal conformer was fabricated to provide a protective interface between the raw tissues and the silicone prosthesis, minimizing direct contact and thereby reducing moisture retention and the risk of secondary fungal infection (Fig 4). Functional nasal patency and patency of auricular component was established by creating patency within the wax pattern. Adequacy of airflow through the nasal openings was verified using the mouth mirror test. Necessary modifications were performed until satisfactory esthetic and functional outcomes were achieved.

PROCESSING OF THE SILICONE PROSTHESIS:
The finalized wax pattern was invested using plastic utility containers as the flasking assembly. The wax pattern was sealed to the master cast, and the first pour was completed using Type III dental stone. To preserve the morphology of the nostrils during processing, they were blocked with putty-consistency addition silicone. Conical orientation indices were incorporated into the first stone pour to facilitate precise reorientation of the mold components during packing. Following application of a separating medium, the second pour of the investment was completed. After complete setting of the investment material, dewaxing was carried out for approximately 15 minutes. Particular care was taken to ensure complete removal of residual wax from the mold cavity. Auricular wax patterns were flasked separately using three pour technique following the same protocol as mentioned previously followed by dewaxing (Fig 5).

The prosthesis was fabricated using medical-grade room-temperature vulcanizing (RTV) silicone elastomer (Technovent Ltd., United Kingdom). The silicone base and catalyst were mixed in a ratio of 10:1 in accordance with the manufacturer's recommendations. Shade matching was performed under natural daylight conditions (Fig 6). The lightest skin tone identified on the patient's face was selected as the base shade, and intrinsic pigments were incrementally incorporated until an acceptable color match was achieved. An anti-slump agent was subsequently added and thoroughly blended to improve the handling properties of the material.

The intrinsically characterized silicone was packed into the mold, and the flasks were closed under uniform pressure to allow extrusion of excess material. vulcanization was permitted to proceed at room temperature for 24 hours. Following complete vulcanization, the prosthesis was carefully retrieved from the mold and excess flash was trimmed using surgical scissors. The processed prosthesis was examined for defects such as voids, tears, and surface irregularities prior to final characterization and delivery.

PROSTHESIS DELIVERY AND FOLLOW-UP:
The completed silicone prosthesis was evaluated for fit, adaptation, marginal integrity, esthetics, function and patient comfort (Fig 6). The prosthesis demonstrated satisfactory retention and harmonious integration with the surrounding facial tissues. Prosthesis was retained using water soluble adhesive (Technovent, UK). Additional retention was provided using spectacles. Detailed instructions regarding placement, removal, cleaning, storage, and maintenance of the prosthesis were

provided to the patient. Regular follow-up visits were scheduled to assess prosthesis performance, tissue health, and patient satisfaction.

CONCLUSION:

Congenital erythropoietic porphyria is a rare and debilitating disorder that may result in progressive facial mutilation with profound functional, esthetic, and psychosocial consequences. Rehabilitation of extensive craniofacial defects in such patients remains a significant clinical challenge, particularly when surgical reconstruction is contraindicated or unlikely to provide predictable outcomes.

The present case demonstrates the successful rehabilitation of a severely disfigured CEP patient through a comprehensive silicone-based maxillofacial prosthetic approach. The integration of donor facial moulages, functional impression techniques, intrinsic silicone characterization, and three-dimensional printed ocular components facilitated restoration of facial symmetry, esthetic harmony, and prosthesis stability. The definitive prosthetic outcome resulted in substantial improvement in facial appearance, patient confidence, and social acceptance.

This report highlights the pivotal role of maxillofacial prosthodontics in the multidisciplinary management of rare craniofacial deformities and underscores the value of individualized prosthetic rehabilitation in restoring dignity, function, and quality of life in patients affected by advanced congenital erythropoietic porphyria.

"Let's aspire not just to succeed; but to make a change!"

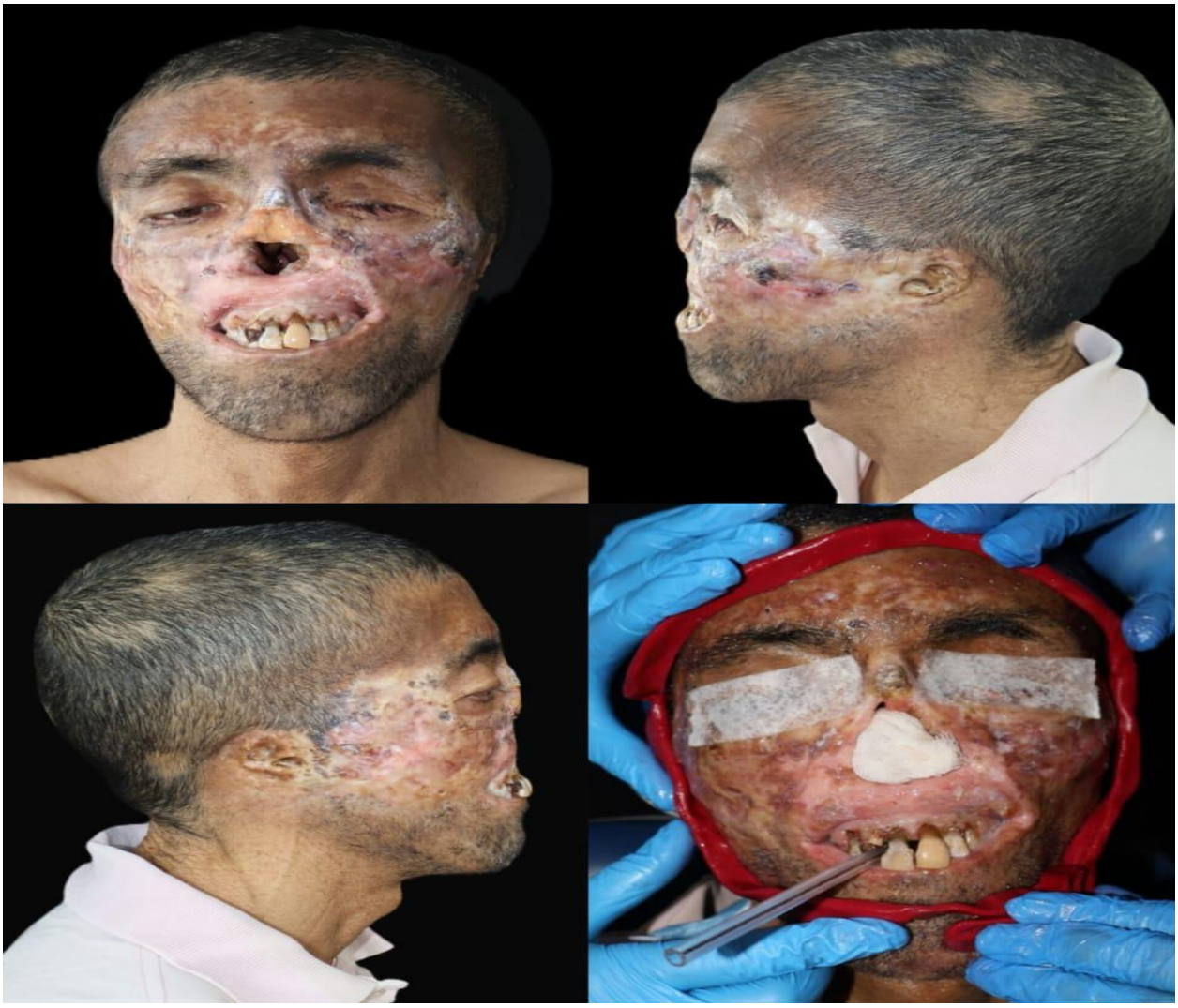

Fig 1: Preoperative photographs, Preparation for impression making.

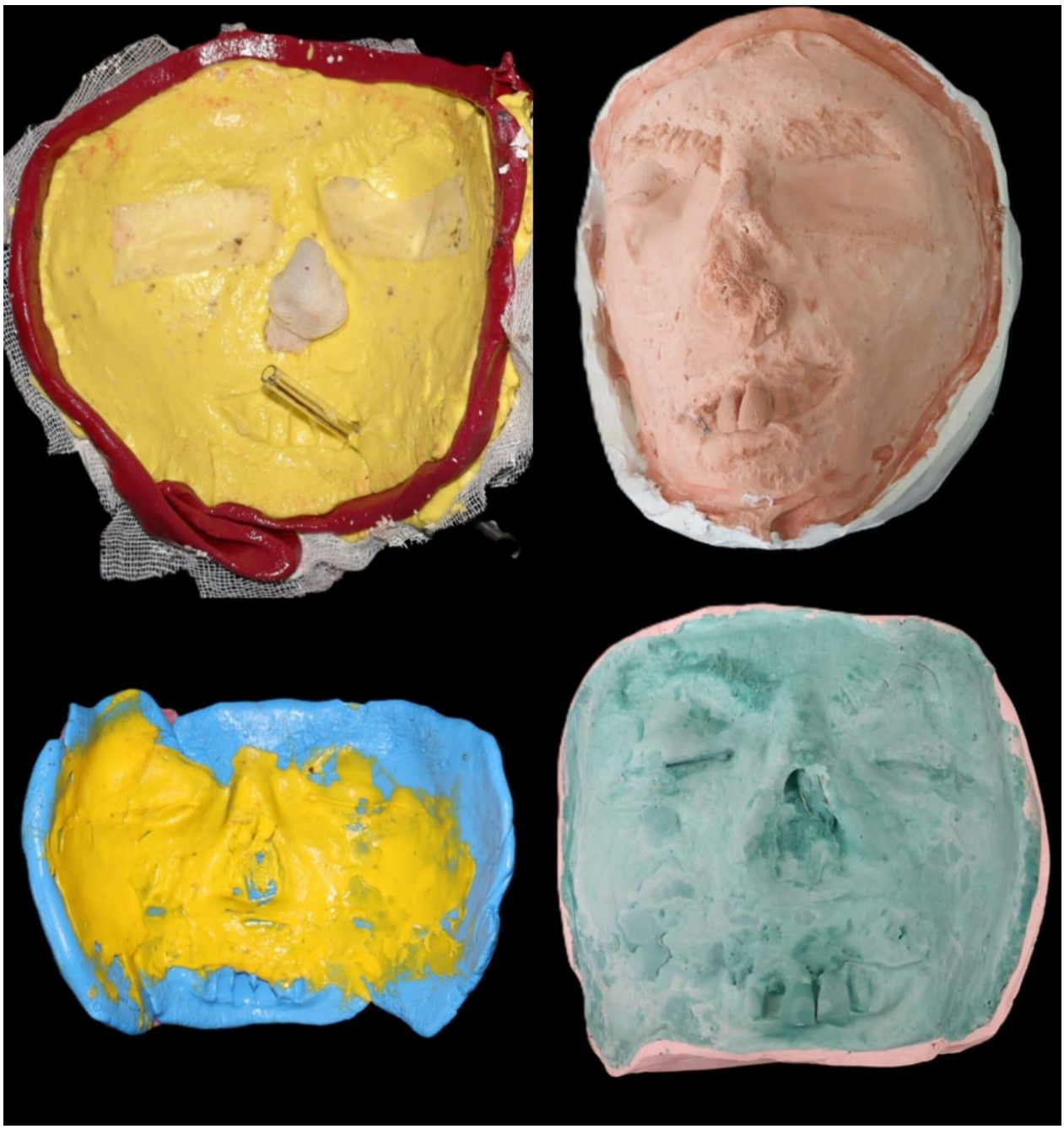

Fig 2: Primary impression, Primary cast, Functional impression, Master cast

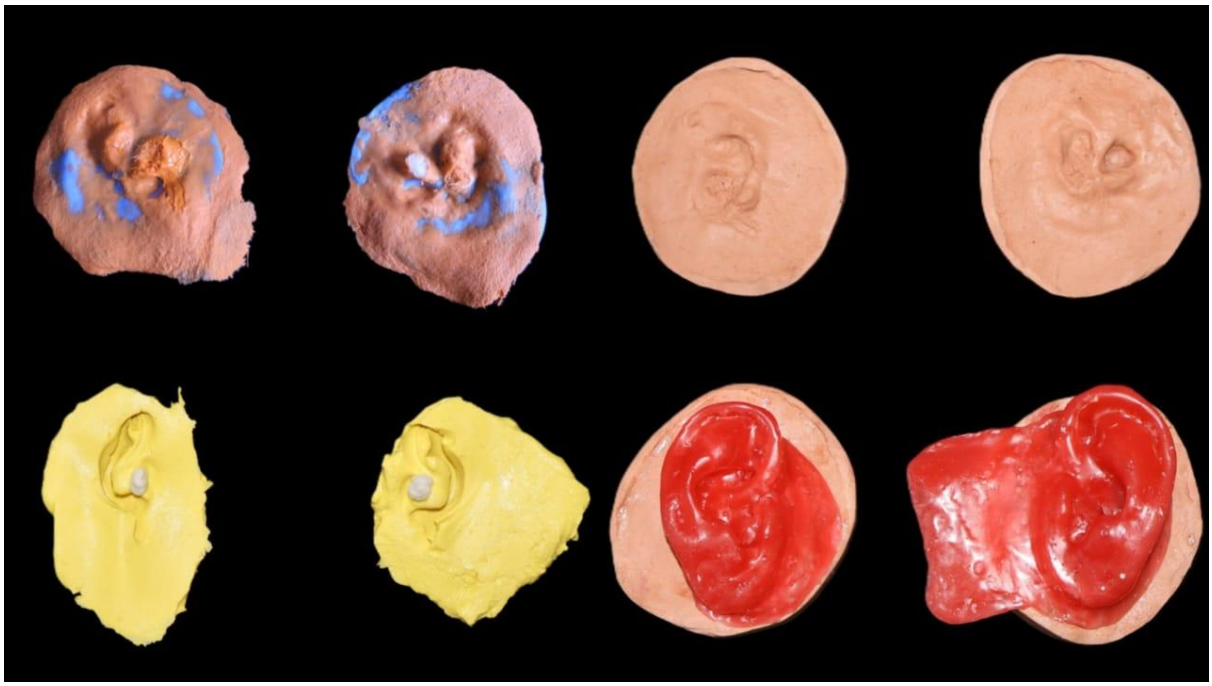

Fig 3: Impressions of auricular component, Casts, Wax pattern for auricular prosthesis

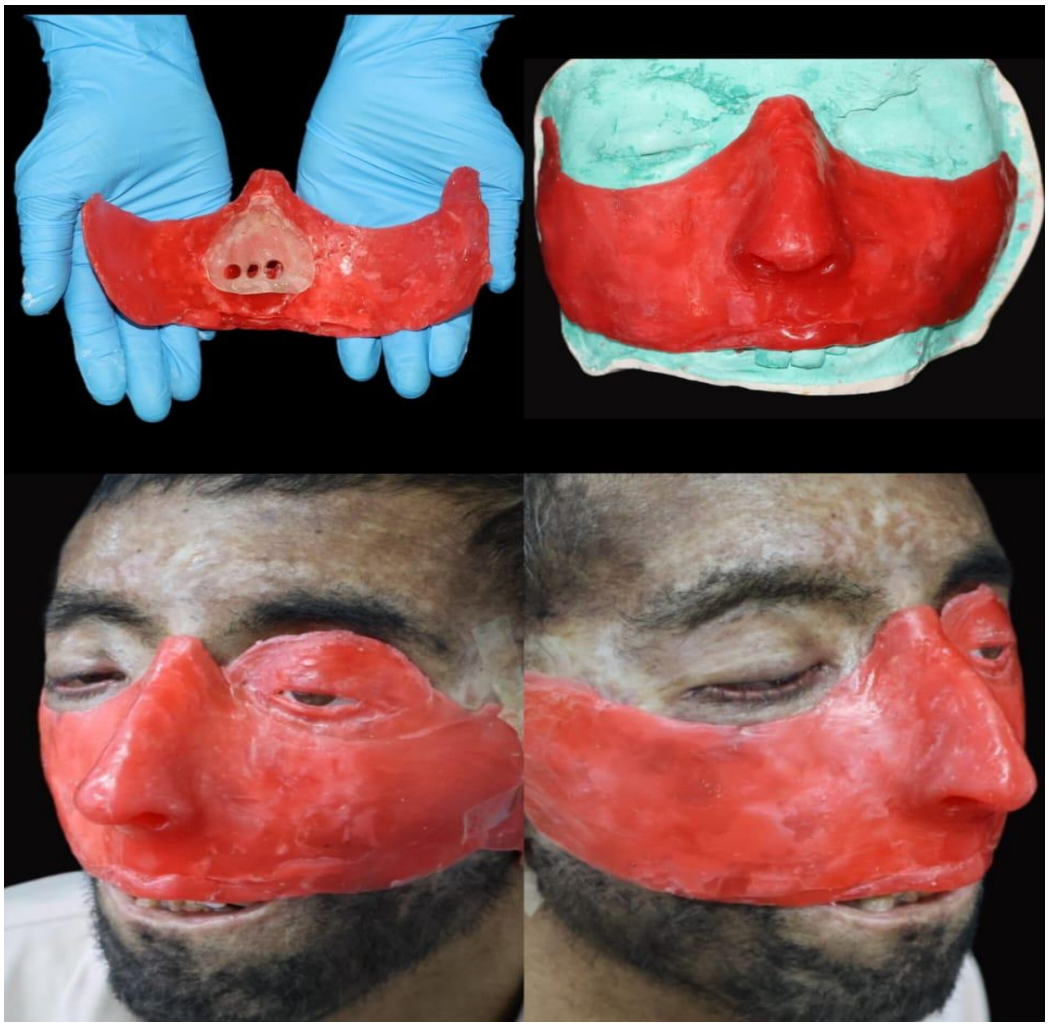

Fig 4: Wax pattern with conformer in place, Try-in of wax pattern

Fig 5: Flasking of prosthesis, Dewaxing of prosthesis

Fig 6: Shade matching, Prosthesis delivery

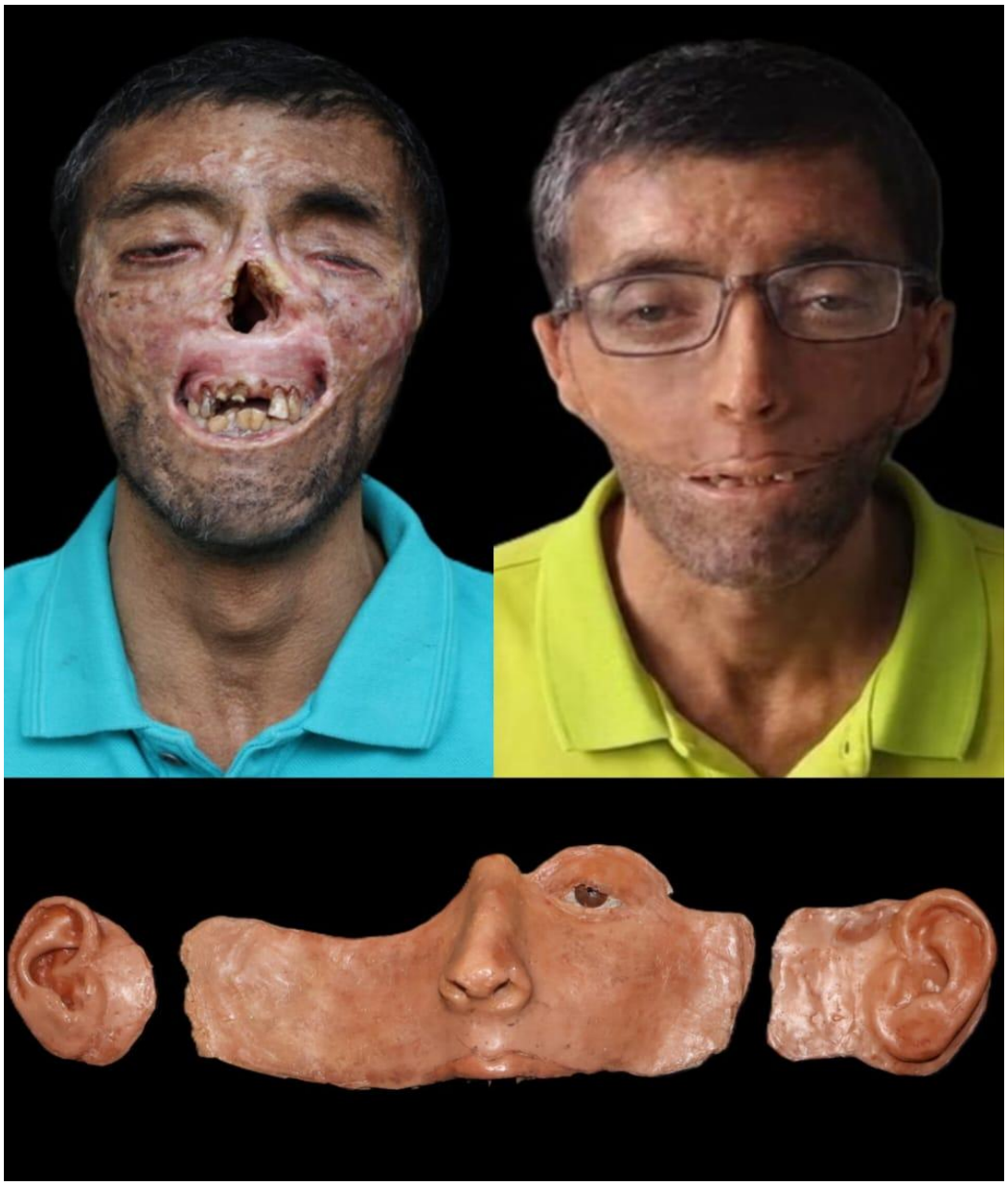

Fig 7: Post treatment outcome, Finished prosthesis

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