

Improvement in the Quality of Life of a Patient of Ectodermal Dysplasia with Reconstructive Surgeries

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Abstract

Ectodermal dysplasias are a complex group of heterogenous, heritable disorders entailing two or more developmental abnormalities in ectodermal structures, such as hair, teeth, nails, and/or sweat glands. The most common subtype of these disorders is X-linked hypohidrotic that significantly impairs the quality of life of its sufferers. A 15-year-old boy, who sought the treatment for protuberant lips, saddle nose, dental anomalies, fine sparse hair, decreased sweating, intolerance to heat and photosensitivity, experienced dramatic improvement in his quality of life and confidence with aesthetic correction comprising autologous fat grafting, rhinoplasty, lip reduction, microblading and comprehensive prosthodontic and orthodontic treatments undertaken in collaboration with dental and plastic surgery departments and expert psychological counseling.

Keywords: Ectodermal dysplasia, fat grafting, lip reduction, microblading, rhinoplasty

INTRODUCTION

Ectodermal dysplasias are a complex group of heterogenous, heritable disorders entailing developmental abnormalities of structures such as hair, teeth, nails, and/or sweat glands of ectodermal origin.^[1] The most common subtype of these disorders is X-linked hypohidrotic that reveals sparse eyebrows and dental anomalies (100%), scalp and body hair (62–89%), hypohidrosis (85%) and onychodysplasia (39%).^[2] Although an early diagnosis of ectodermal dysplasia helps to avoid life-threatening complications, such as hypothermia, loss of temperature regulation and infections, a pervasive concern for the physical appearance and the consequent psychosocial impact severely impairs the quality of life (QoL) of its patients.^[3–5] Anxiety and depression have been demonstrated to impair QoL in a majority of cases.^[6] Hence, a well-planned multidisciplinary (surgical, dermatological, dental and psychiatric) approach to improve physical appearance and thereby QoL is imperative for their optimal societal rehabilitation.^[7] We report gratifying outcome with this approach in an adolescent male.

CASE REPORT

A 15-year-old boy, a known case of hypohidrotic ectodermal dysplasia, presented to us in early 2015 with protuberant lips, saddle nose, dental anomalies, fine sparse hair, decreased sweating, intolerance to heat, and photosensitivity [Figure 1]. He was markedly anxious due to his physical appearance with a Dermatology Life Quality Index (DLQI) score of 24. His sibling, a younger sister, also suffered from similar milder disease for which she was unwilling for surgical treatment. The other family members were unaffected.

He had undergone prosthodontic treatment two years back; orthodontic workup for the alignment of teeth was pending. However, unsatisfied with the aesthetic improvement, he desired further enhancement. Thus, a comprehensive management plan was drawn in collaboration with the departments of reconstructive surgery and dentistry.

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The autologous fat transplantation was carried out by using the Coleman technique, and the raw fat was harvested using Tulip® (Tulip Medical Products, San Diego, California, USA) cannula.^[8] The fat was harvested from the abdomen after infiltration with tumescent local anesthesia (0.9% sodium chloride, epinephrine, and local anesthetic). Then, the fat was centrifuged and processed to separate 5mL of purified fat that was injected with an injection cannula. Approximately 1.5mL of purified fat was injected in each nasolabial fold and 1mL in each temporal hollow.

Six months later, after extensive radiological, hematological, and biochemical workup, a reconstructive surgeon performed an external rhinoplasty to correct the saddle nose by suturing the graft harvested from his costal cartilage between the septa on both sides and by attaching it to the anterior nasal spine [Figure 2]. Nine months later, the same surgeon performed a lip reduction surgery under local anesthesia. An “M”-shaped marking at the upper vermillion border was made, extended laterally, making a similar one on the lower lip with allowance for 50% greater volume, thereafter a triangular wedge of tissue was excised using a beveled incision and sutured [Figure 3]. Concomitantly, topical treatment, for skin hydration, improvement of sparse scalp hair and psychological counseling continued.^[9]

In June 2018, the patient expressed a desire for hair transplantation of the scalp and eyebrows. The same was deemed inappropriate due to his hair being too fragile.

Instead, after charting a symmetrical eyebrow shape with stencils, microblading was performed to enhance the appearance and density to the eyebrows provided using a manual blade for deposition of chocolate brown pigment in the upper dermis [Figure 4].

At the end of August 2018, his DLQI score decreased to 13.

DISCUSSION

Ectodermal dysplasias are a group of rare (with incidence rate of 1 in 100,000), nonprogressive, and diffuse



Figure 2: Appearance after autologous fat grafting and external rhinoplasty



Figure 1: Initial presentation with fine sparse hair on scalp and eyebrows, saddle nose, and protuberant lips



Figure 3: Appearance after lip reduction



Figure 4: Appearance after completion of cosmetic correction including microblading

genodermatoses with more than 192 distinct subtypes. The most common subtype of these disorders is X-linked recessive hypohidrotic (anhidrotic), which is characterized by anhidrosis, hypodontia, and hypotrichosis.^[14,15] The typical facial appearance consisting of frontal bossing, thick lips, a broad depressed nasal bridge, and deformed ears frequently leads to dissatisfaction, social anxiety, and thereby an impaired QoL. Although no large-scale studies quantifying the psychosocial impact of this disorder exist, isolated case reports of children and adolescents made targets of criticism, mockery and exclusion by their peer group, and the resultant social isolation have been documented.^[10-12]

Studies have also revealed that aesthetic correction, surgical or nonsurgical, usually leads to an improved QoL along with an overall improvement in their psychological well-being.^[13]

The extensive rehabilitation needed by these patients needs a multidisciplinary approach, spanning many months to years. Dental anomalies such as hypodontia and hypoplastic maxilla, usually the initial concern, also reduce the chance of uptake of implants.^[16] Prosthodontic treatment was therefore carried out sequentially with fitting of implants and crowns followed by orthodontic alignment. These along with concomitant cosmetic corrections led to an acceptable aesthetic improvement at the end of five years.

Lipofilling, having undergone significant reforms in technique, now gives predictable and sustainable correction of facial asymmetry. It was initially performed to augment

nasolabial and temporal hollows in a minimally invasive way and at affordable cost as the cost of fillers was beyond the patient's capacity.

Saddle nose, one of the most challenging deformities to manage, has a positive outcome following corrective surgery in up to 78% of patients.^[17] Correction of hypertrophic lips was performed while maintaining their relative proportion using an M-shaped incision instead of a simple one in which a horizontal wedge of tissue is removed.^[18] This was made more challenging due to the inherent asymmetry of the lips, eyebrows, and the nose. This asymmetry also led to less than optimal outcome than that is observed generally. However, the inability to achieve optimal outcome should not deter dermatologists and surgeons who should strive over years to achieve some degree of facial improvement and resultant improvement in the patients' lives.

Declaration of patient consent

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

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Conflicts of interest

There are no conflicts of interest.

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