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Case Report

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Xeroderma Pigmentosum: A Defying Problem

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Abstract

TFour Patients with Xeroderma Pigmentosum are presented, stressing the need for a multidisciplinary approach, that must include genetics, prevention (trying to avoid exposure to irradiation and namely to sunlight) and treatment. The benefits of rotation dermabrasion are presented and finally shown the benefits of radical surgery through the presentation of three of the four Patients with radical surgical treatment, one of them having had a monoblock total facial skin excision (performed for the first time in the world).

Keywords

Xeroderma pigmentosum, Dermabrasion, Radical surgery, Facial skin replacement

Introduction and Objectives

The aim of this presentation is particularly to emphasize the value of a multidisciplinary approach in the treatment of Xeroderma Pigmentosum, which is a rare autosomal recessive disease, that due to its genetic heterogeneity, involving multiple different genes, conditions not only the severity of the disease but also the age at which it starts to show its dangerous skin involvement. These patients suffer from a congenital anomaly of the ectoderm that reacts with hypersensitivity to any form of radiation energy. Due to that, the sun rays or any form of radiation, can lead to the progressive development of skin cancer, frequently of early development (thus affecting mostly children that, eventually, may even not attain adult life). Nevertheless, properly cared for, even the more severe cases may survive until adulthood. One has to call attention to the possible preventive measures to be taken as much as possible, by protecting the skin surface from sunlight exposure (not forgetting about clothes and large hats). It is also important to show the value of early rotation dermabrasion and to emphasize the need for immediate surgical control of all initially small lesions that will appear, thus trying to avoid the need for more extensive Surgery. Obviously, everything is based on regular and frequent clinical examinations.

Material and Methods

We present 4 of our treated children:

Patient number 1

A 11-years-old girl, with a localized upper lip spindle-cell carcinoma, that only required to obtain a cure, a classical cheiloplasty (Figure 1 and Figure 2).

Patient number 2

A 3-years-old girl with a spindle cell carcinoma of the nose and a small basal cell carcinoma of the right malar region, treated by direct surgical excision, complemented by a 2 stage rotation dermabrasion of the face and neck and further small plastic surgery procedures (Figure 3 and Figure 4).

Patient number 3

A 11-year's young boy with a spindle cell carcinoma of the left auricular region and invasion of the temporal bone and thus requiring its partial (although radical) removal, care being taken to preserve the facial nerve. The extensive wound remaining after radical excision of the tumor, was covered by a large rotation temporal flap. That Patient was later subject to a rhinoplasty, using a tubular graft obtained from the inner surface of the arm. Unfortunately, he died some years later because of an inoperable chest wall sarcoma (Figure 5 and Figure 6).

Patient number 4

A 9-years-old boy, son of a fisherman, and living near a beach, showed the typical distribution of the lesions in the face, hands and feet (those later ones not yet malignant). His lesions had started to increase when aged 3, after having

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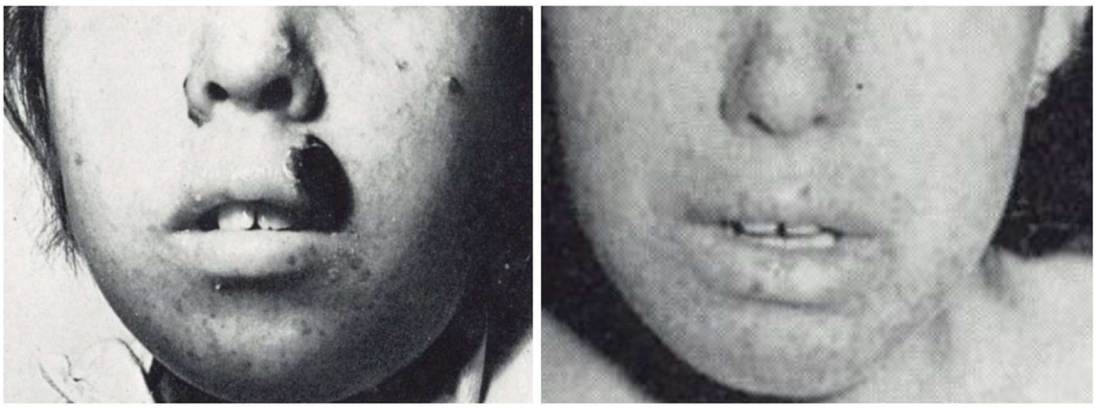

Figure 1: Patient 1- Appearance before and after surgery.

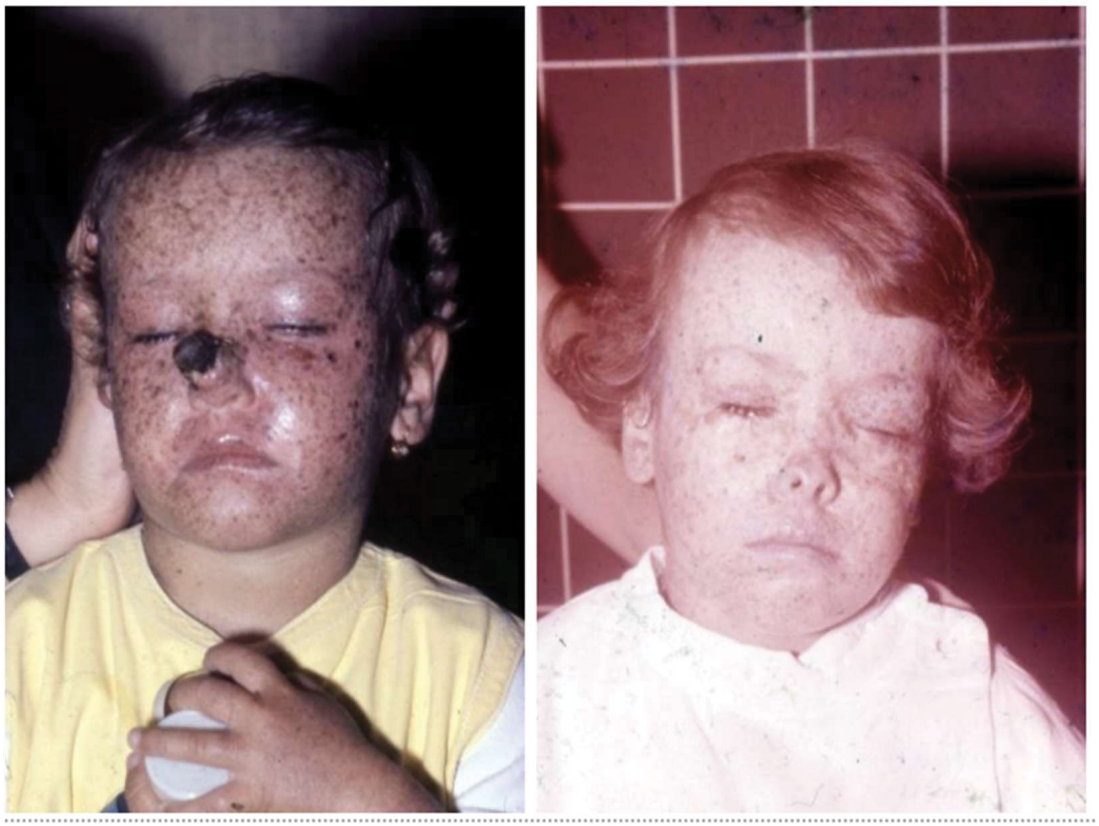

Figure 2: Patient 2- Appearance before and after excision of a spindle-cell carcinoma of the nose.

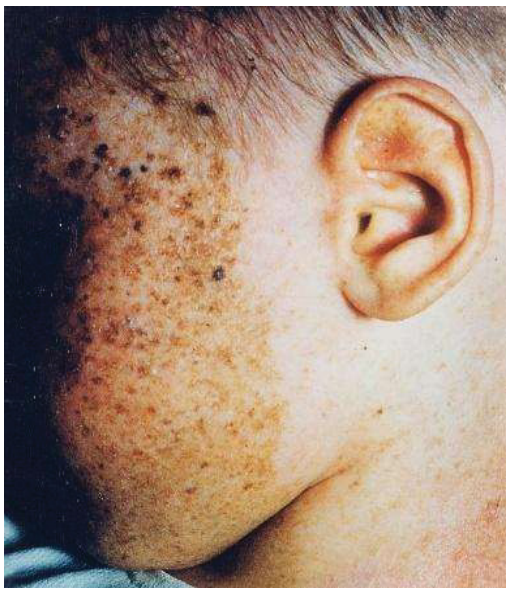

Figure 3: Patient 2- Skin appearance after treatment with rotation dermabrasion

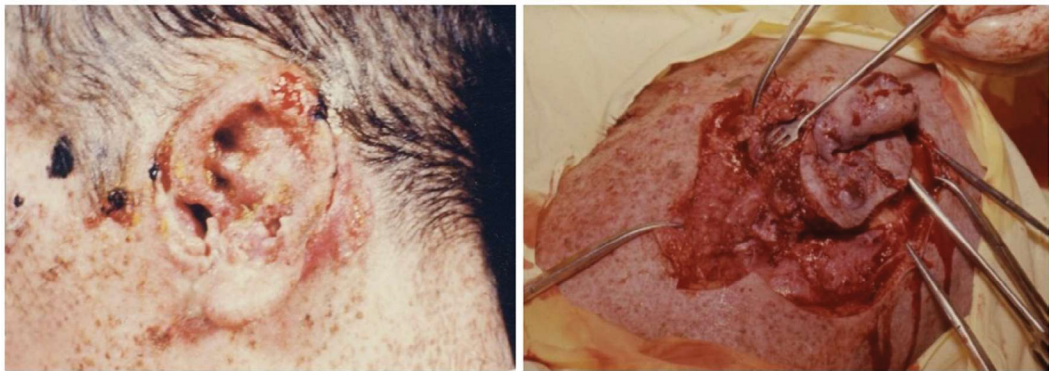

Figure 4: Patient 3- Appearance of the left temporal region before and after tumour excision.

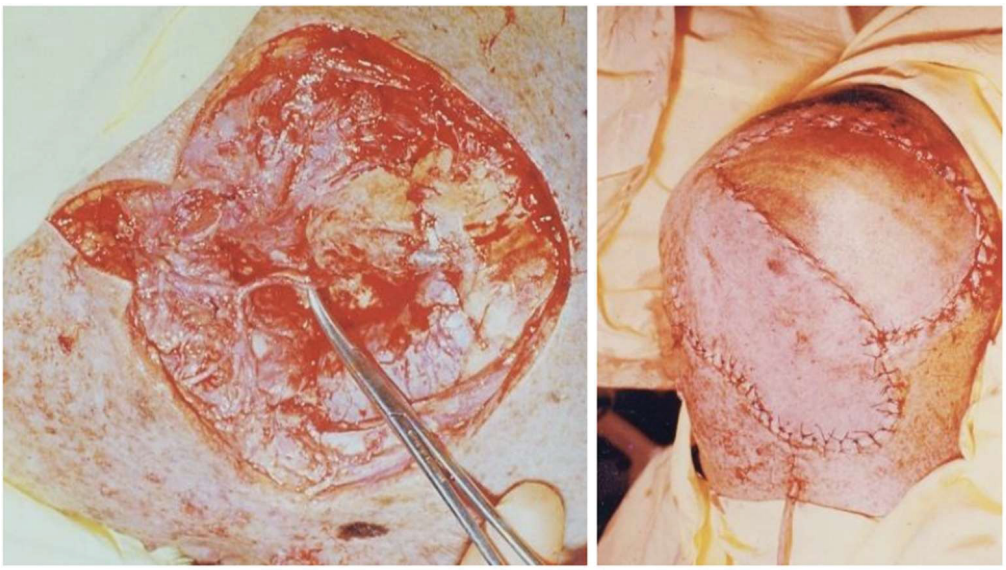

Figure 5: Patient 3- Appearance before and just after total monoblock facial skin removal.

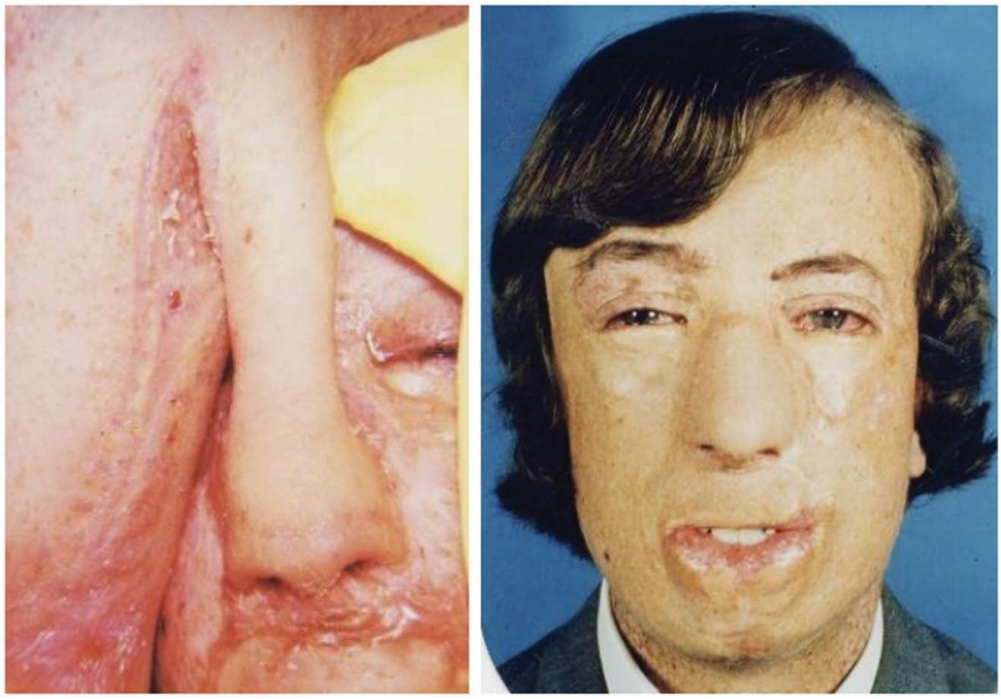

Figure 6: Patient 3 - Showing the tubular Rhinoplasty and final facial appearance.

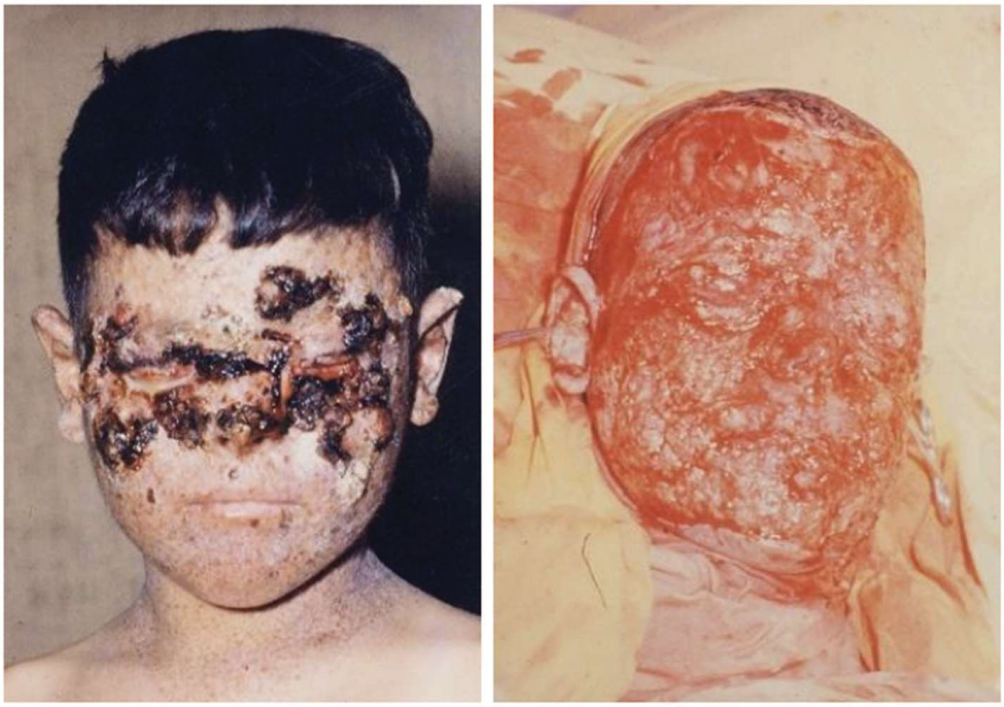

Figure 7: Patient 4 - Appearance before and just after total monoblock facial skin removal.

had irradiation with ultraviolet light, because of rickets. After several small surgical interventions and the use of radiotherapy, he had infected malignant lesions disseminated all over his face as well as palpebral ectropions [1]. It was then that it was decided to perform a surgical intervention never done before, even at World level: the one stage monoblock excision of the total facial skin (Figure 7 and Figure 8).

Surgical Technique: (Patient number 4)

Patient, lying on his back, general anaesthesia was started as endotracheal to change later for a tracheostomy. Radical, monoblock, total facial skin resection was performed including forehead, nose, red lips and lids (of which only could be saved little more than the conjunctiva). That was followed

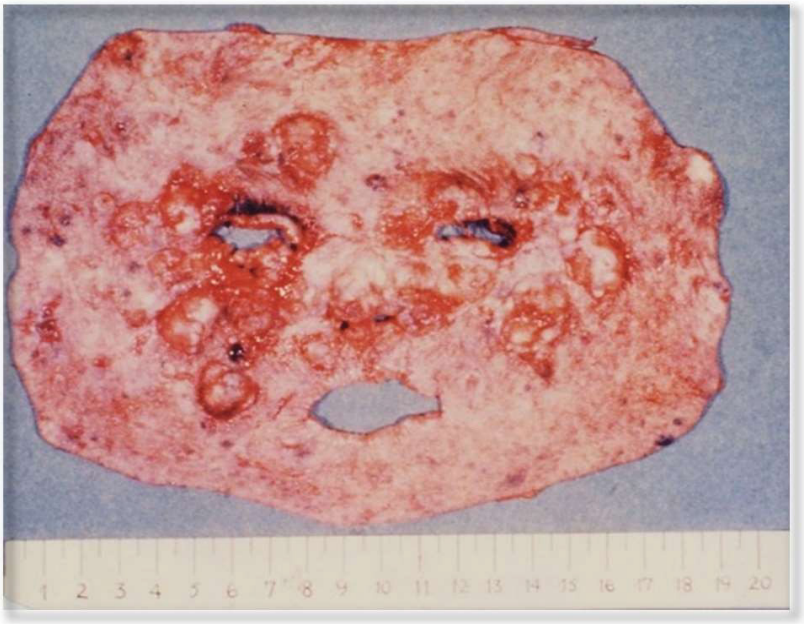

Figure 8: Patient 4 - Operative specimen.

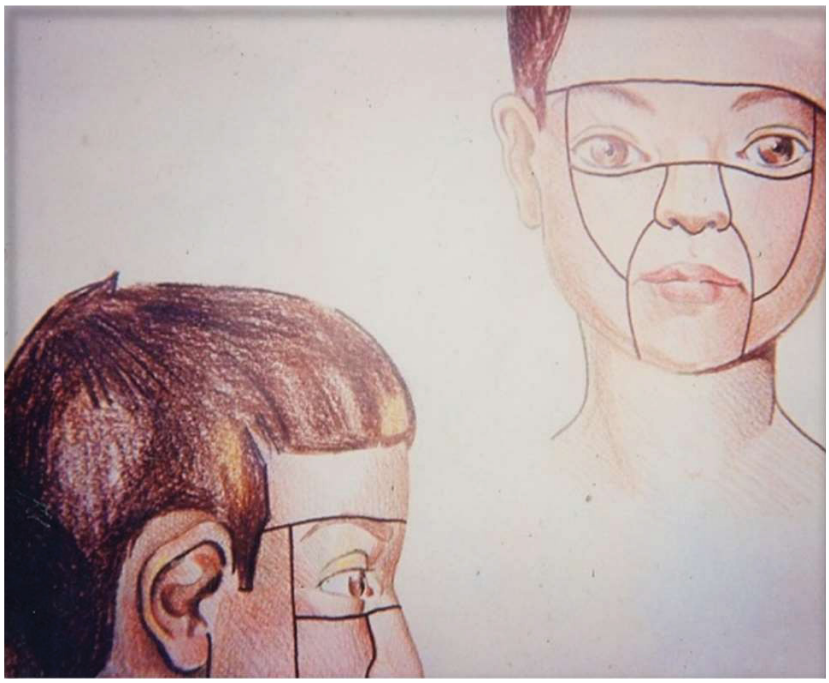

Figure 9: Patient 4 - Ulloa Scheme.

by immediate reconstruction according to the principles of the "Facial Aesthetic Units'' advised by Mario Gonzalez Ulloa, so that the lines of adjoining skin grafts would coincide with the natural skin creases. Taking into account the possibility of retraction of the skin grafts to the lids could result in lagopthalmus and ectropion, a single graft was placed in the palpebral remnants and which was maintained for 2 months (the palpebral cleft being opened then).

Considering the existent skin infection it was decided to use medium thickness grafts, obtained with the electric Dermatome. The skin of the anterior abdominal wall was used for the mouth, so that the hole in the central part of the graft, (corresponding to the umbilicus) would correspond to the lips and open mouth. Several other plastic surgery interventions were needed, including a rhinoplasty with a tubular graft obtained from the inner part of the left arm. The treatment was completed with rotation dermabrasion of the neck.

He was kept with an occlusive dressing for 5 days and fed by a gastric tube for 3 weeks. The intervention lasted 8 hours and 1.500 ml of whole blood was administered (Figure 9).

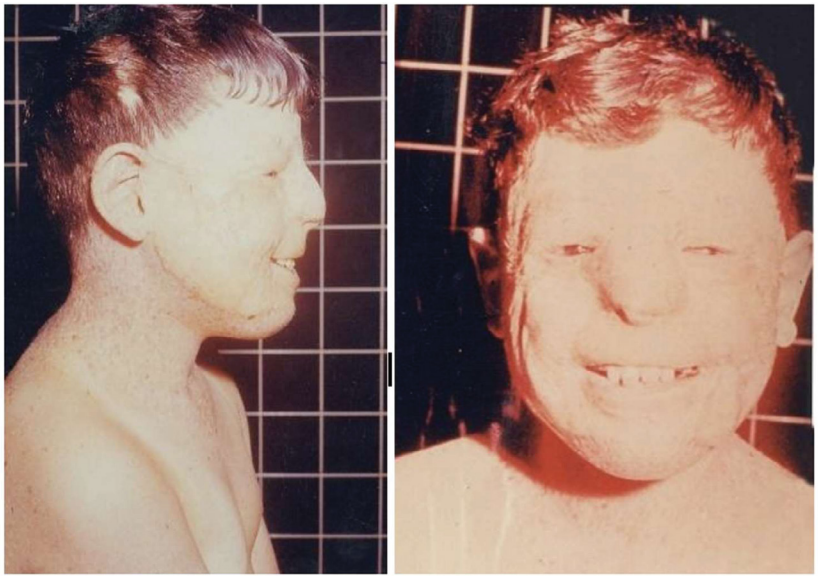

Figure 10: Patient 4 -Facial appearance some months after surgery, front and lateral view.

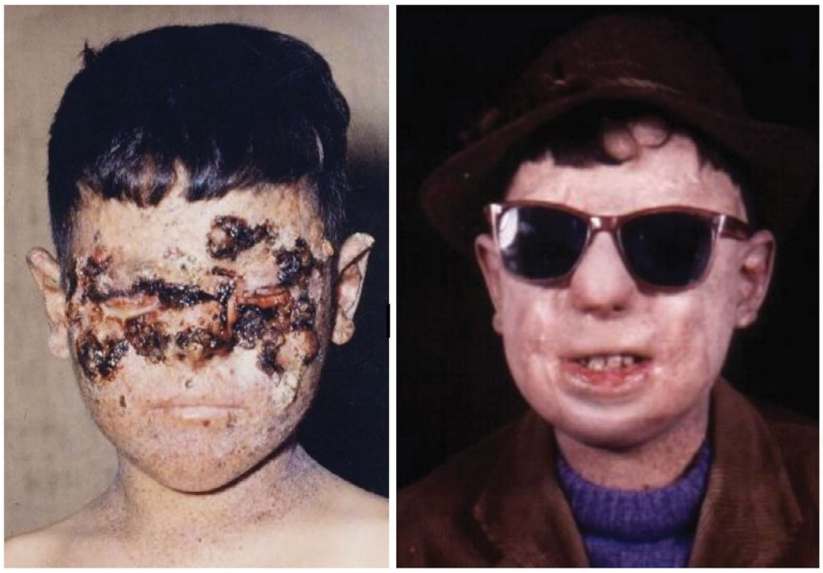

Figure 11: Patient 4 - Facial appearance before and after surgery.

Discussions and Results

Xeroderma pigmentosum is considered to be one of the rare hereditary disorders, studies having shown marked multiple involved genes, genetic heterogeneity conditioning various clinical aspects and intensity of signs. Cell fusion analyses revealed that there are 8 genetic complementation groups in XP. Seven of these groups, XP-A through XP-G are associated with defective nucleotide excision repair (NER), while the remaining group, a variant form of XP (XP-V) is proficient in NER but deficient in a specialized DNA polymerase (Poln), a translation synthesis polymerase that can replicate. DNA templates containing ultraviolet induced damage. Depending on the type of mutation, different pathways could be impaired, resulting in distinct phenotypes. The severity and frequency of the disease varies widely due to its genetic heterogeneity. XP-G is more intense and XP-F usually mild. XP-A and XP-C are relatively common and XPO-E is very rare.

So far, hormonal and radiological studies have been within normal limits. Nevertheless, EEG studies always reveal alterations although with no fixed pattern. Psychiatric evaluation has always shown a low intellectual level that seemed to be particularly significant at older ages.

The treatment of Xeroderma Pigmentosum must be fundamentally prophylactic, avoiding exposure to any type of irradiation, namely sunlight (using adequate clothes, sun glasses and protective creams). Dermoabrasion may eventually be useful in selected cases, but will have eventually to be repeated several times.

Surgery is indicated in all localized single lesions, being associated with the plastic surgery procedures usually required, as well as the excision by electro coagulation of all the small-suspected lesions that may appear in the disseminated forms of the Disease. The Patients must be seen frequently. Fortunately metastases seem to occur rarely, perhaps because the vascular blood supply to the skin seems to be scarce in these Patients and there is an excess of fibrous tissue as a consequence of the disease itself (Figure 10).

Basically, on Patient number 4, one couldn't avoid having severe doubts about what to do and the common opinion of all contacted and the one that could be taken from the literature, was that it was an hopeless situation. It would not be correct cancer surgery (if at all possible?) to try to perform the isolated removal of all the multiple malignant lesions, not only because of their multiplicity but also because of their close relationship. So that led us to believe that the only possible solution would be the one stage monoblock excision of all the malignant and infected facial lesions, although that had never been performed in the world and at that time facial skin transplants (still today with extremely rare indications) were not known [2-4].

One can say that the aesthetic result is far from perfect. Nevertheless, function is normal and it was markedly rewarding to see the change in behaviour and happiness of the Patient, who changed from a fearing and suffering child to an active and happy Human being (Figure 11).

The cooperation of a psychologist is certainly essential, concerning not only the Patient itself (many times with impaired mental health), but also considering the devastating impact on the family because, for the disease to exist, both Parents would have to be XP carriers [5-8].

The skin lesions are very frequently associated with other lesions, namely eye and neurological problems. Photophobia is common, as well as dry eyes, with redness and even chronic inflammation and keratitis (even leading to blindness). Neurological degeneration is not infrequent, with eventual hearing loss, progressive cognitive impairment, poor muscle control and coordination, etc [9,10].

There are also several genetically related disorders, like Cockayne Syndrome, Trichothiodistrophy, UV Sensitive Syndrome and Cerebro-Oculo-Facial-Skeletal Syndrome.

Vitamin D supplementation and its blood levels must be carefully evaluated, considering that the Patients must avoid sunlight exposure [11,12].

Conclusions

In the case of our most challenging Patient, we believe that the monoblock total excision of the facial skin followed by immediate skin grafting was the right approach, although it had never been performed in the World (and so far has not been done again). That 4th Patient (abandoned by the Parents at the door of the Cancer Institute!) survived many years, working as an employee in the lifts of the Institute (thus avoiding exposure to the sun rays) and died more than 20 years later, from a brain tumour [13].

Xeroderma Pigmentosum makes it mandatory frequent medical examinations and the action of a multidisciplinary team, of which prevention as well as surgery are essential elements. So we would suggest:

Examinations 3 monthly by the Patient's Doctor (either a Paediatric Oncologist/General Practitioner) and as often as required by a Dermatologist and a Plastic Surgeon (both with experience in Oncology), 6 monthly (and as often as required) by an Ophthalmologist, Yearly (and also as often as required) by a Neurologist, and, finally (last but not least….), by a Psychologist, as initially suggested [14,15].

Disclaimer

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Consent

Was given orally at the time of surgery (to be used on science work and on any other work I would wish).

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