Living with Xeroderma Pigmentosum in Guatemala: A Yulmacap Village Experience

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Xeroderma Pigmentosum (XP) is a rare autosomal recessive disorder characterized by DNA repair defects that cause photophobia, sunlight-induced cancers, and neurodegeneration. Many of the genes related to Xeroderma Pigmentosum are part of a DNA-repair process known as nucleotide excision repair (NER), and are named as XPA, XPB, XPC, XPD, XPE, among others.

Yulmacap is a village located in Guatemala containing many patients with XP. This is due to the high grade of consanguinity found in this community. The mutation found in these people was first described by Cleaver J in 2006, and is a stop codon in exon 8 in *XPC* gene (c.940delC, p.Pro323Leufs*).

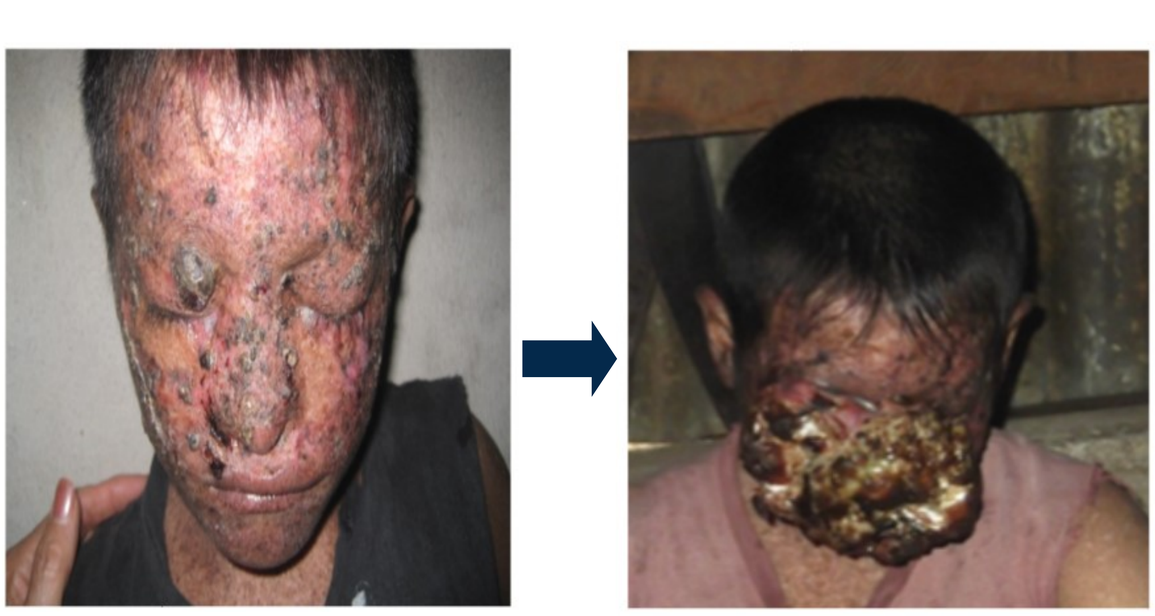

Figure 1 "Evolution of XP in a Guatemalan patient"
Source: INVEGEM.

It has been difficult to explain to these people the effects of sun exposure in the affected children. So the majority of the children have severe non-melanoma skin cancer. Their tumors are advanced, with necrosis and infection and they do not use any sun protection (figure 1).

The main goal for the present study was to improve the quality of life of the affected children and to prevent the transmission of the XP disease to another people.

We performed a molecular test for the mutation detection to stablish the affected children and the XP family carriers. For doing this, we extracted blood samples and DNA extraction to the affected families. And then performed a polymerase chain reaction followed by enzyme restriction for the mutation detection (figure 2).

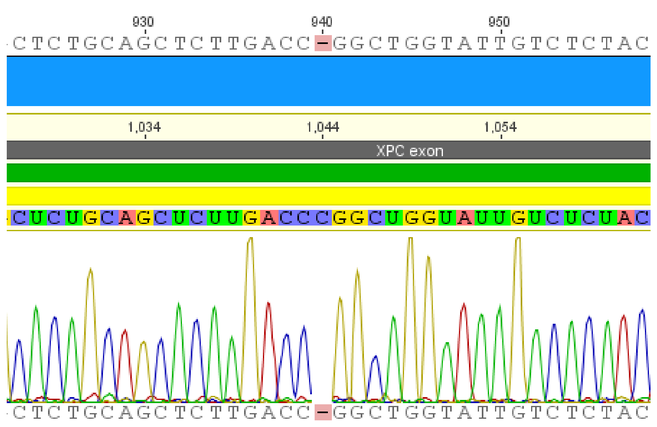

Figure 2 "Electrophogram of mutation in exon 8 *XPC* gene of Guatemalan patients (c.940delC)"
Source: data obtained in the molecular biology laboratory, INVEGEM.

We found 9 children affected with the disease (8 affected families) and 27 people that are carriers of the mutation (figure 3; table 1). This village is very isolated from the majority of Guatemalan population, they only speak a Mayan language and their education level is extremely low.

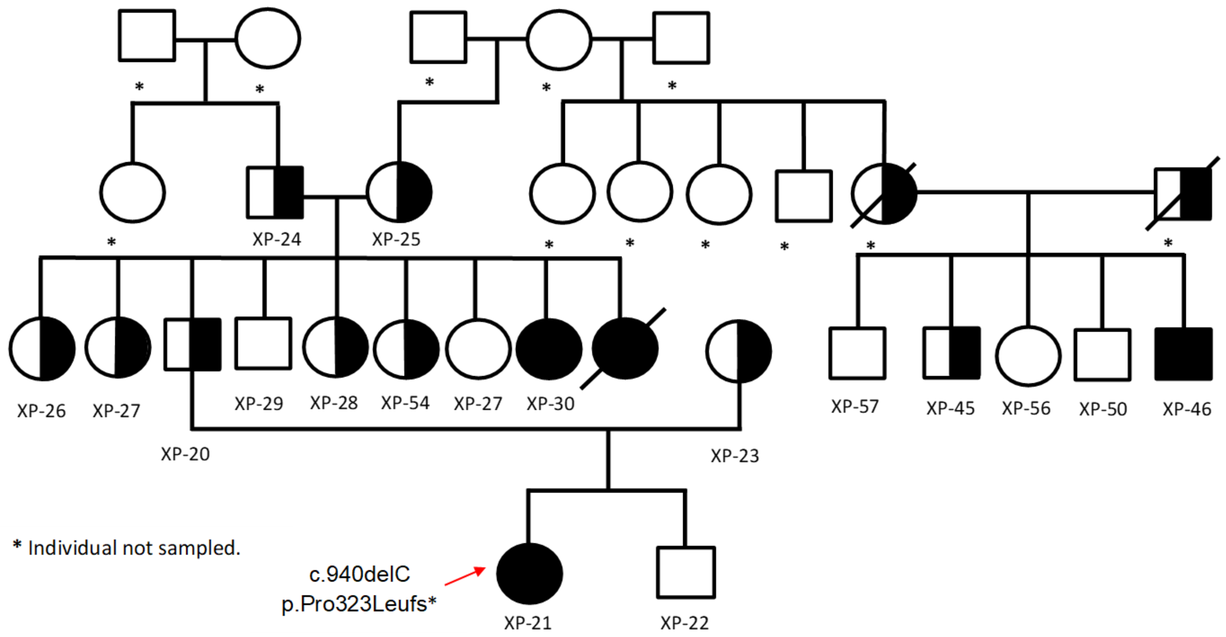

Figure 3 "Pedigree of Guatemalan XP patient"
Source: INVEGEM.

Table 1 "Patients analyzed for determination of mutated genotype of the *XPC* gene"

Mutated genotype, *XPC* gene	No. of individuals
Negative for mutation	25
Carrier	27
Affected	9
Total	61

Source: data obtained in the molecular biology laboratory, INVEGEM.

We developed a special suit that is made from special clothes that prevent the UV light exposure. And we designed the architectural plans for the construction of a day care building, which prevents that UV light penetrates the walls and windows (figure 4). On the other hand, our team gave a genetic counselling to the mutation carriers, explaining the risk of disease transmission.

Figure 4 "Special suit for XP patients"
Source: Castellanos, C., UNIS.

In conclusion, several institutions collaborate to improve the quality of life of children living with XP. The activities that we performed were the genetic characterization of XPC mutation and genetic counselling, the special suits for affected children, architectural planning of the day care center and the performing of skin tumor surgeries.