

## Case study

### DNA collected with Oragene®/saliva kits enables remote field collections for rare genetic disease research and diagnosis in Brazil



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#### Study overview

Xeroderma pigmentosum (XP) is a rare autosomal recessive genetic disease of DNA repair in which the ability to repair damage caused by ultraviolet (UV) light is deficient. Multiple basal cell carcinomas and other skin malignancies frequently occur at a young age in those with XP. The Araras' district of Brazil's Goiás state is a small isolated community with high levels of sun exposure, consanguineous marriages and one of the world's highest incidence rates of XP patients, with more than twenty cases in a population of approximately 1,000 residents.

Most XP patients have defects in the nucleotide excision repair mechanism (NER) due to mutations at XPA to XPG genes, but some are mutated at a translesional DNA polymerase, XPV, a variant type. Dr. Carlos Menck and his research team at the University of Sao Paulo collected DNA samples from the Araras and Goiania areas in order to genotype patients and their families to better understand the genetic mutations involved in XP, its inheritance factors and to provide early molecular diagnosis to children to help prevent further skin deterioration.

#### Main challenges

The main concerns regarding the DNA collection for this project were the lack of blood collection facilities in the remote Araras' district of Brazil and transporting the DNA samples to the lab without risk of sample degradation due to extreme high temperatures. The team also wanted an easy-to-use collection method that was non-invasive to maximize donor participation and patient care.

#### Why Oragene/saliva samples?

The Oragene/saliva collection kit is an all-in-one system for the self-collection, stabilization, and transportation of DNA from saliva that has been proven to perform equivalently to DNA extracted from blood for genotyping and



Oragene device prior to collection

Sample ready for transport, storage and processing



Some DNA Genetek products may not be available in all geographic regions, contact your sales representative for details.

“The collection of saliva using the Oragene collection kit was easy and fast. The samples are quite stable in the field and while travelling back to the lab, even in the high temperatures of that region (close to 35°C). Furthermore, our analysis on paired Oragene/saliva and blood samples indicates no difference in DNA yield, quality and genotyping results.”

Professor Dr. Carlos Menck,  
University of Sao Paulo

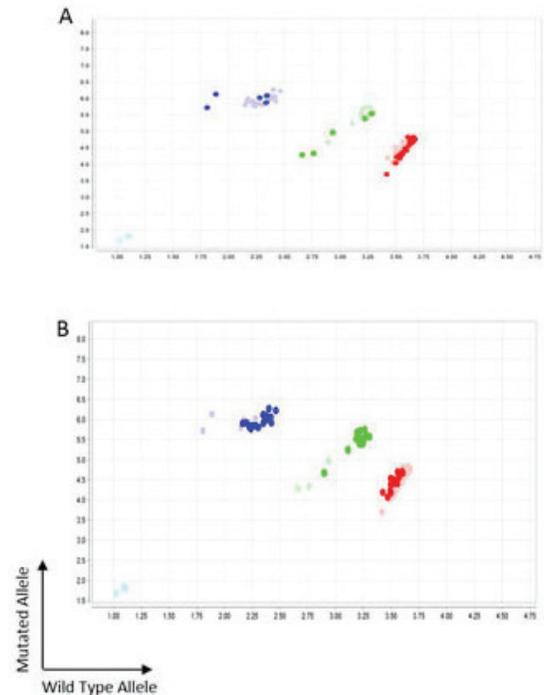


molecular diagnosis. The collection method is an easy 2-step process of providing a 2 mL saliva sample into a tube and capping it to release the stabilization chemistry. Once the sample is provided it is stable at ambient temperatures for years and can withstand extreme high temperatures normally experienced with transporting samples in these regions without any signs of DNA degradation.

## Results

One hundred and thirty-eight (138) saliva samples were collected in the field using the Oragene self-collection kit at Goiania and Araras from XP patients and their families aging from 2–88 years old. Blood samples were also collected from most of the patients, as confirmation controls. Because of the non-invasive and easy procedure of the Oragene/saliva kit, the in-field collection was fast for all donors.

Results shown from independent DNA extractions of paired Oragene/saliva and blood samples indicate similar high yield and sample quality with no difference in genotyping results as analyzed using the TaqMan® SNP genotyping assay as observed in the figure (right).



**Figure 1:** Allelic discrimination plot for Exon 6 mutation highlighting DNA samples extracted from saliva (A) and blood (B).

Two mutations were found at the XPV gene, one of them located at a splice site one nucleotide after exon 6 and the other one, a stop codon located at exon 8. These were screened in the population of that community in order to confirm clinical diagnosis, and identify carriers of the mutations. This screening relieved the concerns of a mother who was afraid her 2 year old son had this disease as he was not a carrier of the mutated alleles.

To learn more about XP and the scientific impact of this human disease, please see: Menck CF, Munford V. DNA repair diseases: What do they tell us about cancer and aging? *Genet Mol Biol.* 2014 Mar;37(1 Suppl):220-33.

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