



“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,  
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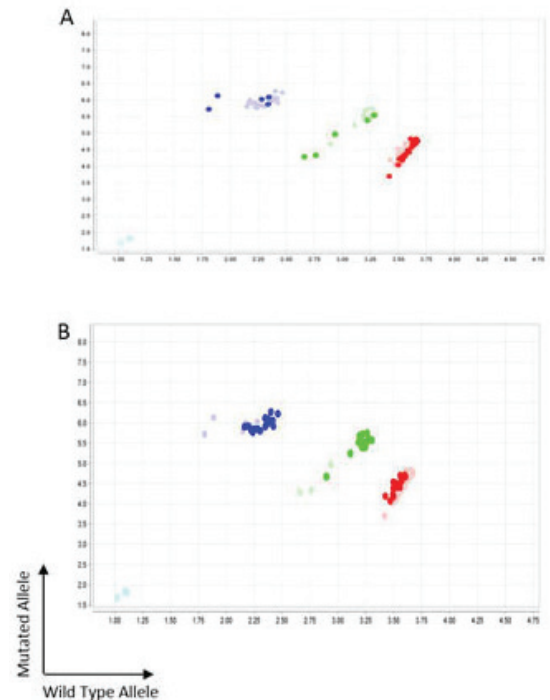


molecular diagnosis. The collection method is an easy 2-step process of providing a 2 mL saliva sample into a tube and capping it to release the stabilization chemistry. Once the sample is provided it is stable at ambient temperatures for years and can withstand extreme high temperatures normally experienced 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.

Oragene®-DNA is not available for sale in the United States.

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