



## Case study



"We have found in the end that Oragene/saliva kits are quite cost effective in terms of the amount and quality of DNA, the stability over time and the ease of extraction. It works very well for us."

*Michael Dean, Ph.D.*



"All kids love spitting and almost 100% participate."

### Oragene®/saliva collection kits enable easy and cost-effective DNA sampling for paediatric cancer research

Michael Dean, Ph.D.

Senior Investigator and Head of the Human Genetic Section, Laboratory of Experimental Immunology, Cancer and Inflammation Program, Center for Cancer Research, National Cancer Institute, USA

Member of the American Society of Human Genetics, American Association of Cancer Research, Centre Etude du Polymorphisme Humaine (CEPH), the Human Genome Organization (HUGO) and an adjunct faculty member at Hood College

#### Overview

Dr. Michael Dean and his research team have established significant health disparities in the diagnosis of adult and paediatric cancers among the indigenous Mayan people of Guatemala and Nicaragua. They are actively sampling and sequencing DNA from all cancer diagnoses for multiple genetic studies of this population. They collect cancer tissue, blood and saliva samples and to date their laboratory has sequenced the exome of bladder, cervical, kidney, prostate and adrenal gland tumors.

Dr. Dean collects DNA samples from paediatric cancer patients as young as 2 years old and their family members who may be as old as seventy. To date, he has collected samples from nearly the entire population of the Unidad Nacional De Oncología Pediátrica (UNOP) hospital in Guatemala. Donors come from all areas of Guatemala and many are indigenous Mayan people whose primary language is one of 10 different indigenous dialects.

#### Main challenges

The most critical challenge faced by the research team was obtaining blood samples from children for their DNA collection. Invasive blood draws resulted in a lack of patient cooperation and the need for a professional phlebotomist. Another challenge was very limited access to freezer storage within the Guatemala hospital. These challenges made blood collection and sample storage extremely difficult.

#### Why Oragene/saliva collection kits

Dr. Dean chose Oragene/saliva collection kits because of the ease of collection and long-term stability at room temperature while delivering an abundance of high quality DNA. Buccal swabs were evaluated but concluded to produce lower quality DNA of insufficient yield for the required analysis.



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



## Our team:

### UNOP GUATEMALA

Dr. Federico Antillón, Director

Dr. Margarita Barnoya,  
Paediatric Cancer Specialist

Mauricio Castellanos

Patricia Valverde

Sandra Luna-Fineman

Claudia Garrido

Tania Cerrato

### NCI

Dr. Michael Dean, Head,  
Human Genetic Section, NCI

Julie Sawitzke

Silvia Jimenez

Brittany Kostih

Katy Kimpert

## Results

Nearly 100% of the children are happy to participate in Oragene/saliva collection and no specialized hospital personnel are required to obtain the samples. The majority of saliva samples are collected in the clinic; however, the team has sent Oragene/saliva kits to donors' homes for self-collection when required (i.e. a family member is unable to come to the hospital). Because Oragene/saliva collection kits stabilize DNA at ambient temperature for years, transport and storage is cost-effective and easy.

Once collected, the Oragene/saliva samples are shipped to Dr. Dean's lab at the National Cancer Institute (NCI) where they are processed and analyzed or simply stored within the kits at ambient temperature for future applications.

Dr. Dean and his research team have collected over 5,000 Oragene/saliva samples and have successfully performed Sanger and targeted sequencing as well as SNP array analyses.

Current studies involve genetic ancestry profiling of osteosarcoma in the Mayan populations with the goal to understand why some cancers are more prevalent in Guatemala than in North America and Europe.



"Each saliva sample is divided into aliquots for easy processing. Sample aliquots in bar coded tubes are stable at ambient temperature and ready for transport to our lab at the NCI."

Michael Dean, Ph.D.



"... what's been very important is [the paediatric hospital in Guatemala] doesn't have a lot of storage facilities so we have stored samples there at room temperature now for years that provide good quality and quantity DNA."

## References:

Using Oragene/saliva kits for their DNA collection, they have published various studies on retinoblastoma, mutations in the RB1 gene, TPMT (gene involved in metabolizing chemotherapeutic drugs), and paediatric acute lymphoblastic leukemia.

### Retinoblastoma

Dean et al. *Canc. Letters* 2014

### Acute Lymphocytic Leukemia

Xu et al. *JClinOnc* 2011

Garrido et al. *Mol. Oncology* 2013

Xu et al. *JNCI* 2013

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