



## Case study

### Oragene®/saliva kit enables easy at-home collection while delivering high quality DNA suitable for a large-scale genome-wide association study (GWAS) and Next Generation Sequencing (NGS)



Dr. Shiro Ikegawa

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“Your Oragene/saliva kit is really useful. We are using it in many fields of genome research relating to human diseases – for mutation analysis, exome sequencing and GWAS genotyping – resulting in successful publications.”

*Dr. Shiro Ikegawa, Laboratory of Bone and Joint Diseases, Center for Integrative Medical Sciences, RIKEN, Tokyo, Japan*

#### Introduction

Dr. Shiro Ikegawa is a lead investigator for the Laboratory of Bone and Joint Diseases at the RIKEN Center for Integrative Medical Sciences in Japan. Dr. Ikegawa and his study team are using genome analysis to better understand and diagnosis monogenic and polygenic diseases of the locomotive system, including rare diseases like skeletal dysplasia, and common diseases like osteoarthritis, osteoporosis and disc degeneration. They have discovered 17 disease genes for skeletal dysplasias and 6 susceptibility genes for common diseases throughout multiple ongoing research studies, many of which have benefited from the use of Oragene®/saliva collection kits for non-invasive at-home collection.

#### Study overview

Spinal extradural arachnoid cyst (SEDAC) is a cyst in the spinal canal that protrudes into the epidural space from a defect in the dura mater. Early diagnosis is important to prevent cyst expansion that can compress the spinal cord and cause irreversible neurological defects. Familial SEDAC cases have been reported, suggesting genetic etiological factors. These cases have been associated with lymphedema-distichiasis syndrome (LDS), whose causal gene is FOXC2. To gain further insight into the genetic etiology of SEDAC, Dr. Ikegawa's study team examined FOXC2 mutations in 17 SEDAC subjects, 10 familial and 7 sporadic cases, by exome sequencing, Sanger sequencing and TaqMan® copy number assay.



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## Collection methods considered

At-home collection was required to obtain samples of family members who could not come into a clinic for collection. Furthermore, the study team wanted a non-invasive alternative to blood draws to increase donor compliance and avoid difficulties with stabilizing and transporting blood samples. Buccal swabs, a hair/nail kit, and Oragene/saliva samples were considered to meet their collection requirements.

## Why Oragene/saliva collection kits?

While DNA collection through buccal, hair, and nail samples are non-invasive alternatives to blood draws, they all deliver minimal, often negligible, quantities of DNA that can be highly degraded by bacteria. These collection options can result in unreliable DNA recovery and failures on downstream analysis. The Oragene/saliva self-collection kit offered the study group a non-invasive and patient-friendly alternative that reliably delivers high quantity, high quality DNA that performs as well as blood for GWAS and Next Generation Sequencing. Furthermore, as the Oragene chemistry stabilizes high-molecular weight DNA at room temperature for many years, transporting and storing the samples prior to analysis was not a concern to DNA integrity.

## Results

DNA was easily collected at the donors' homes using Oragene/saliva kits and full compliance was achieved. The patients and family members were quite happy to be freed from the physical and economical burden of coming to hospitals for blood collection. The DNA yield and quality extracted were reported by Dr. Ikegawa as quite good for GWAS genotyping and whole exome sequencing. Through their genomic analysis, Dr. Ikegawa and his research group showed that the FOXC2 mutation is one of the etiological factors of SEDAC, although other gene(s) are likely to be involved. As a result of their work, patients and their family members will be able to use genetic screening to evaluate their risk before developing irreversible neurological defects.

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