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Case study

DNA collected with Oragene®•DNA facilitates multiple skin biology studies at the Institute of Medical Biology (IMB), Singapore

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Study overview

The Institute of Medical Biology (IMB), Singapore is involved in a range of research studies including stem cell development and differentiation, skin biology, cancer and genetic diseases. The institute believes that new knowledge will lead to novel therapeutic strategies for improved quality of life by studying how molecular changes lead to increasing cell specialization and complexity in the context of human tissues and diseases. The institute is involved in a large number of overlapping genetic studies into atopic dermatitis (AD) in children and acne vulgaris (AV). These studies are designed to examine the spectrum of fillagrin-null (*FLG*) mutations in Singaporean Chinese individuals and investigate the association of *FLG* mutations with atopic dermatitis and its clinical markers while comparing the *FLG* landscape between Asia and Europe.

Main challenges

There were two main challenges for these studies. The first was related to the age range of the target population for the AD study (2–21 years old). The researchers wanted to make participation in the study as easy as possible for young people and their parents/guardians. The second challenge was that while the DNA samples would be collected in a clinic, the clinic was not in the same location as the lab so samples would have to withstand storage in the clinic until transport to the lab could be arranged.

Collection methods considered

Oragene[®]•DNA was the primary method considered for the AD study. As the studies were extended to include adults and acne vulgaris patients, the team decided to offer both saliva collection using the Oragene•DNA collection kit as well as blood sample collection.





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"We chose Oragene-DNA primarily because we were working with a cohort of patients aged 2–21 years old and we felt that it was ethically better to be able to offer saliva collection to the parents and guardians of these young people."

John Common, PhD Institute of Medical Biology, Singapore

Why Oragene•DNA

The IMB researchers believed that offering a non-invasive, saliva-based collection device would be the best option for the cohort of patients participating in the AD study (aged 2–21 years old). Oragene•DNA provides a completely non-invasive option for collecting DNA while meeting the requirements for both high quality and high quantity DNA. Oragene•DNA can be stored at ambient temperature for years, offering the study team the flexibility they needed with regard to storage and extraction.

Results

Over a two year period, the research team successfully collected 500 Oragene•DNA samples for the AD study. The acne vulgaris study is ongoing but to date, 500 samples have been collected with 25% of the participants choosing to provide blood samples and 75% preferring to provide saliva with Oragene•DNA. With these samples, the research team published the following key findings:

- Null mutations in *FLG* cause IV and are a major risk factor for AD.
- The study characterized *FLG* in Singaporean Chinese with IV and AD and found a wide spectrum of 22 *FLG* mutations, of which 14 are novel.
- The current study corroborated the association of *FLG* mutations with AD, palmar hyperlinearity and keratosis pilaris in Chinese and highlights the skin genetic differences between Europe and Asia¹.
- FLG null mutations are not a protective factor for acne vulgaris².

2 J.E.A. Common, S. J. Brown et al., *Journal of Investigative Dermatology*, Filaggrin null mutations are not a protective factor for acne vulgaris.

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¹ H. Chen, J.E.A. Common, et al., *British Journal of Dermatology*, Wide spectrum of filaggrin-null mutations in atopic dermatitis highlights differences between Singaporean Chinese and European Populations.