

Investigating a Frisian Genetic Mutation

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INVESTIGATING A FRISIAN GENETIC MUTATION

Two Dordt University professors are trying to raise community awareness about a genetic mutation in the phospholamban (PLN) gene, which can cause heart arrhythmias and possible cardiac death in people of Frisian descent.

Genetic studies have shown the mutation first appeared somewhere in the province of Friesland, Netherlands, around 700 years ago.

"Since there are many Dutch descendants at Dordt, we believe there are potential carriers of the gene in our constituency. It would be good for them to know that they carry the mutation and take steps to treat potential complications," says Professor of Nursing Dr. Deb Bomgaars.



JAMIN VER VELDE ('99)

Professor of Nursing Deb Bomgaars became interested in the PLN gene, which originated in the Netherlands, after a family member tested positive for it.

Symptoms of the disease include reduced stamina, shortness of breath, arrhythmias, heart rate surges, and full cardiac arrest. Some individuals show no symptoms, but that doesn't make the condition any less serious. The mutation is dominant, meaning that it will be passed on to half of a person's children on average—and their cases might not be asymptomatic.

Bomgaars first took interest in the genetic mutation when she found out that a close family member had tested positive for it. She remembers that, while in a meeting with Professor of Biology Dr. Tony Jelsma, he requested prayer for a family member hospitalized with a racing heart and mentioned that heart disease is prevalent in his family. She later pulled Jelsma aside and mentioned that he might want to investigate PLN.

Jelsma and Bomgaars, along with Professors of Nursing Melanie Wynja and Deb Kleinwolterink, are working with Dr. Dean Jansen, a physician and administrator for the PLN Heart Foundation—and a PLN carrier himself—to help raise awareness for this dangerous mutation.

Research into PLN provides a real-life application to the content that he and Bomgaars teach, says Jelsma—particularly in his human physiology and genetics classes.

"Despite knowing the molecular mechanism of this condition, the fact that it presents in a variety of ways and in different degrees of severity shows that there is still much we don't know about our physiology," says Jelsma. "Our bodies are complex, indeed! Moreover, we see how we can use our knowledge to save lives, whether it's by developing treatments or cures, or whether it's simply by raising awareness."

Bomgaars says the research will impact her nursing students and beyond.

"This research helps me to collaborate with not only other departments on campus, but with the Dordt community," she says. "By researching PLN, I can be of service to God's creation. It is a way for me to help others be aware of potential heart problems that can be treated."

SARAH MOSS ('10)

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Gene research is not new to Dr. Tony Jelsma, who is now helping raise awareness about one gene.