# DR. KANG'S RESEARCH

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**Aim 1: Biorepository Creation for Xq27–Xq28**

- establish a biorepository for confirmed Xq27–Xq28 deletions individuals.
- build a centralized, long-term resource supporting future genomic and clinical studies for deletion.

**Aim 2: Long-Read Genome Sequencing**

- conduct long-read genome sequencing and advanced analysis using Oxford Nanopore Technologies.
- Provide high-resolution genetic and epigenetic data on Xq27–Xq28 deletions.

**X-chromosome deletions can lead to a range of complex disorders. These conditions are difficult to study because symptoms can vary, even among people with similar genetic deletions. This is the case for Xq27-Xq28 deletion disorders. In females, this variability increases due to the X-inactivation process, and even more so because some genes can "escape" this process. These disorders present unique challenges for researchers in this way. By gathering high-quality and diverse samples, scientists can link specific genetic deletions to particular traits or symptoms with greater accuracy, while also assessing the significance of epigenetic data like with X-inactivation. This will strengthen the foundation for future breakthroughs in X-chromosome deletion disorders.**

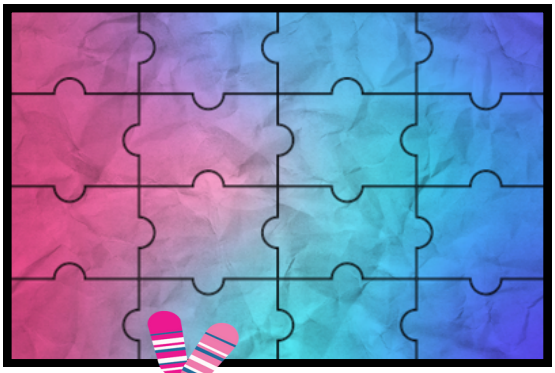
**Traditional DNA tests capture small, simplified fragments of genetic information. Nanopore sequencing captures much larger fragments of DNA and reveals epigenetic marks.**

Traditional

- short, fragmented pieces of DNA
- must estimate how fragments connect

Nanopore

- larger, continuous pieces of DNA
- can characterize stretches of DNA that were previously difficult to analyze



TRAORDINARY JOY!