

## MEASURING X INACTIVATION SKEW FOR X-LINKED DISEASES

The phenotypic expression of X-linked diseases in females heterozygous for a deleterious variant is highly variable and may be attributed to allelic skew in X-chromosome inactivation. Gocuk et al. (1) introduced a novel approach using nanopore sequencing to accurately quantify skewed X inactivation. They tested this method on two ocular X-linked diseases by sequencing three distinct tissues—blood, saliva, and buccal mucosa—since the retina is not accessible for such analysis. Their findings revealed a strong correlation between X inactivation skew and disease manifestation. The authors state that this method is applicable to all X-linked diseases.

1. <https://genome.cshlp.org/content/early/2024/09/13/gr.279396.124>