

## Study sheds light on genetic differences that cause a childhood eye disease

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Medical researchers at the University of Alberta have unlocked part of the mystery underlying a childhood eye disease. New research shows how children with some types of glaucoma end up with missing or extra pieces of DNA.

The missing or extra bits of DNA are called copy number variations (CNVs). The U of A research team had previously shown how they play a major role in causing some types of pediatric glaucoma – a disease that can lead to blindness. In their current study, published in *Human Molecular Genetics*, the authors describe how the CNVs that cause childhood glaucomas are formed.

Using genetic samples from patients living with pediatric glaucoma, the research team studied the locations where extra or missing pieces of DNA begin and end. Close examination of these break points allowed the team to determine how these copy number variations occur.

"Our findings broaden the mechanisms known to cause copy number variations, which improves our understanding not only of pediatric glaucoma, but also of the growing number of genetic diseases linked to copy number variations, including heart disease and psoriasis. We're really only looking at the tip of the iceberg in terms of how CNVs cause disease." said Dr. Ordan Lehmann, an associate professor with the Faculty of Medicine & Dentistry at the University of Alberta and an ophthalmologist with Alberta Health Services. "These findings will also help us to improve the detection of pediatric glaucoma and, by allowing

earlier diagnosis, will help lead to earlier treatment of this condition."

Source: University of Alberta

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