Exploring motivators, barriers, and access to genetic testing for a rare eye disease

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BACKGROUND

Aniridia is a congenital eye condition characterized by a complete or partial absence of the iris.

It occurs in **1 in 50,000 to 100,000** births worldwide.

Most aniridia cases originate from a **pathogenic variation in the PAX6 gene**, affecting development of eye structures.

Genetic testing plays a significant role in the diagnosis and treatment of aniridia.

There are **ethical considerations** around the use of genetic information to support individuals with aniridia.

OBJECTIVES

To examine:

1) **Canadian public-facing information** about access to services and reimbursement policies for genetic testing for rare diseases;

2) **Motivators and barriers for genetic testing** for individuals living with aniridia;

3) **Benefits and risks of genetic testing** for rare neurological conditions.

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It is an ethical imperative to improve access to accurate and current information about genetic testing and counselling to better support individuals with aniridia.

METHODS

Conducted a targeted search and analysis of **N=108 Canadian policies and public-facing information** about access and reimbursement for genetic testing for aniridia.

Conducted and analyzed **N=8 semi-structured interviews** with individuals living with aniridia.

RESULTS

Table 1. Summary of Canadian policy and public-facing search on access to genetic testing

Lack of clear information on where to get genetic testing for rare diseases	**No mentions** of genetic testing for rare diseases on federal and provincial government websites
Limited details on reimbursement for genetic testing	**Only 4 of the top 10 insurance companies in Canada** cover diagnostic services

Table 2. Examples of reasons for genetic testing, and perceived barriers and benefits

Reasons for Genetic Testing	• Family planning • Learn about other health conditions associated with pathogenic variation • Learn about treatment options
Perceived Barriers to Genetic Testing	• Lack of information about genetic testing • Cost of testing • Logistical errors during testing
Perceived Benefits of Genetic Testing	• Learn about specific pathogenic variation and treatment options • Help future generations and contribute to research

MOVING FOWARD

- **Access to genetic counselling** should be a priority to support informed decision-making around testing
- Following neuroethics principles, the implications of **opportunities to enroll in research and clinical trials** that offer genetic testing should be discussed to protect participants' well-being and rights
- Future work in this area should capture perspectives from a **diverse sample**

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