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.

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

<table>
<thead>
<tr>
<th>Reasons for Genetic Testing</th>
<th>Perceived Barriers to Genetic Testing</th>
<th>Perceived Benefits of Genetic Testing</th>
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</thead>
<tbody>
<tr>
<td>Family planning</td>
<td>Lack of information about genetic testing</td>
<td>Learn specific pathogenic variation and treatment options</td>
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<tr>
<td>Learn about other health conditions associated with pathogenic variation</td>
<td>Cost of testing</td>
<td>Help future generations and contribute to research</td>
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<tr>
<td>Learn about treatment options</td>
<td>Logistical errors during testing</td>
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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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