Institute of Genetic Ocular in partnership with Retina São Paulo

Psychosocial Aspects of Genetic Ocular Diseases

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ABSTRACT

Genetic ocular diseases are a heterogeneous group of hereditary diseases that cause substantial vision loss at various age ranges. In addition, there may be syndromic cases that affect other systems such as the skeletal and vestibular systems. The literature indicates that individuals affected by genetic ocular diseases have low quality of life and high rates of depression. They tend to have high healthcare costs, and their caregivers often leave the workforce. Therefore, the objective of this study is to investigate the psychosocial aspects of individuals with genetic ocular diseases, their family members, and caregivers.

Keywords: psychosocial aspects; genetic ocular diseases; psychotherapy; emotional health; family relationships;

Introduction

Ocular genetics is a specialized field within ophthalmology that focuses on the study and treatment of ocular diseases of genetic origin, known as genetic eye diseases (GED), which can be transmitted through different hereditary patterns, such as autosomal recessive, autosomal dominant, X-linked, or mitochondrial (BERGER; KLOECKENER-GRUISSEM; NEIDHARDT, 2010; HAIM, 2002).

Among the most common GEDs are Hereditary Retinal Dystrophies (IRD), a heterogeneous group of conditions that cause degenerative and functional changes in the retina (BERGER; KLOECKENER-GRUISSEM; NEIDHARDT, 2010). These include Stargardt disease (STGD), retinitis pigmentosa (RP), Usher syndrome (US), and Leber congenital amaurosis (LCA), affecting approximately 1 in 4,000 individuals.

STGD, for example, is a macular dystrophy that typically begins in adolescence or early adulthood, leading to gradual central vision loss. RP, initially characterized by night blindness, progressively limits the individual's visual field (BESSANT; ALI; BHATTACHARYA, 2001; HAIM, 2002). The syndromic form of RP, associated with hearing loss, is called US. LCA, in turn, is considered the most severe form of IRD, affecting both central and peripheral vision from the first months of life.

So far, there is no cure for GED, except for gene therapy developed to treat LCA caused by a mutation in the RPE65 gene (NASH et al., 2015; SAHELI; MARAZOVA; AUDO, 2015; RUSSELL et al., 2017). In addition to IRDs, other GEDs include Marfan syndrome, which affects multiple body systems, and Knobloch syndrome, characterized by high myopia, retinal detachment, and brain abnormalities (SALLUM; CHEN; PEREZ, 2002; LI et al., 2021).

Patients with GED experience low quality of life and also have high rates of anxiety and depression. This is due to the lack of social understanding of their condition and the challenges in adapting to vision loss. Vision loss can be considered a grieving process, as the loss of sight represents the death of a sense. This grief process, combined with a decrease in quality of life, makes it increasingly difficult for GED patients to experience psychological well-being (HAMBLION; MOORE; RAHI, 2011; NASCIMENTO et al., 2015; DEAN et al., 2017; ANIL; GARIP, 2018; TAIPALE et al., 2019; GALVIN et al., 2020; KUMAR, 2023).

Family members, parents, and caregivers also suffer psychosocial impacts related to GED. They also experience high levels of anxiety and depression and low quality of life. Additionally, they struggle to maintain financial health and are often overwhelmed by the care required for patients with GED.

The psychosocial impacts of GED vary depending on an individual's life history and emotional resources, requiring different forms of support such as psychotherapy, psychiatry, and rehabilitation (NASCIMENTO et al., 2015; DEAN et al., 2017; TAIPALE et al., 2019; KUMAR, 2023).

Psychotherapy can help patients in accepting visual impairment and developing coping strategies for their condition. There are adaptive coping strategies, which aim to solve challenges and regulate emotions, and maladaptive coping strategies, which avoid or escape obstacles without regulating the emotions arising from them (GARIP; KAMAL, 2019).

One of the current psychotherapeutic approaches is Acceptance and Commitment Therapy (ACT). ACT is considered a third-wave cognitive-behavioral therapy rooted in functional contextualism and relational frame theory (HAYES, 2004; RUIZ, 2012). It has six core principles that guide its interventions:

1. **Psychological Acceptance**: A conscious process of accepting unpleasant events that are inevitable, meaning that there is no denial of the situation, but rather the realization that it is necessary to experience difficult situations, thoughts, and feelings to accept and learn to deal with them.
2. **Cognitive Defusion**: The process of distancing oneself from cognitive fusion. Cognitive fusion is a state in which a person is fused with their thoughts and feelings, believing them to be facts of reality.
3. **Contact with the Present Moment**: Thinking clearly about things that are happening in the present moment, identifying thoughts and interpretations as subjective psychological events.
4. **Self-as-Context**: The process of understanding that a person changes with every new experience. There is a rigidity in seeing the individual as something static, but individuals constantly evolve.
5. **Committed Actions**: The act of behaving consciously, committing to one's values regardless of external reality.
6. **Values**: Qualities of actions that individuals believe are appropriate in any situation in life, such as honesty, justice, and charity.

   ACT has proven effectiveness in individual therapy for various psychiatric disorders and chronic illnesses (MANGIONE et al., 2001; CASTRO et al., 2006). Additionally, it has shown effectiveness in group therapy, where interventions typically focus on a theme related to the group's suffering, and these interventions usually involve a specific number of sessions, ranging from 8 to 20 (SIMÃO et al., 2008; SANTOS et al., 2011).

The literature indicates that ACT in groups is effective in improving emotional health for groups suffering from anxiety and depression, domestic violence, chronic illness in children, and substance abuse (SIMÃO et al., 2008; SANTOS et al., 2011).

No data was found in the literature regarding ACT psychotherapy for people with visual impairment. However, other psychosocial interventions have already been conducted. A systematic review and meta-analysis evaluated interventions to improve emotional health, anxiety, depression, and loneliness in adults with visual impairment. The results indicated that interventions significantly reduced depressive symptoms, but did not improve anxiety, psychological stress, or loneliness (VAN DER AA et al., 2016).

There were also no studies investigating psychotherapeutic interventions for parents of children with LCA, but one study conducted a two-day Acceptance and Commitment Therapy (ACT) workshop with parents of children with ASD. This training, based on the six pillars of ACT, led to improvements in the parents' emotional health and reduced experiential avoidance (HAYES; STROSAHL; WILSON, 1999; BLACKLEDGE; HAYES, 2006).

Justification

As mentioned above, GED directly impacts the quality of life of those affected (HAHM et al., 2008; HAMBLION et al., 2011; DEAN et al., 2017; NASCIMENTO et al., 2015; TAIPALE et al., 2019; GALVIN et al., 2020; KUMAR, 2023). Their physical health is compromised by alterations with no cure or treatment. Their emotional health is drastically affected by anxiety and depression. Their social relationships may be reduced or altered, as these individuals may not be understood in terms of their limitations. Furthermore, society's structure is not adequately accessible to meet the needs of people with GED (BUCKMAN, 1984; LATHAM et al., 2015).

Given the impacts listed above, it is evident that investigating the psychosocial aspects of this population is crucial. Understanding how socially unadapted they might be and the extent of their psychological distress will lead to the creation of public policies aimed at improving the quality of life for this group.

Living without quality of life and psychological well-being can accelerate physical health deterioration for individuals with GED, further emphasizing the need for a comprehensive evaluation of these individuals (SALLUM et al., 2002; CHAUMET-RIFFAUD et al., 2017; LI et al., 2021). This should be approached from medical, psychological, and social perspectives. The best care for individuals with GED is holistic care.

Lastly, family members and caregivers of individuals with GED also suffer various psychosocial impacts due to their direct connection with these patients. This population is not widely studied, and new research shedding light on their reality could lead to ways of mitigating the impacts on their psychological well-being.

Hypothesis

The hypothesis of this study is that individuals with genetic eye diseases (GED) and their families experience high levels of anxiety and depression, as well as low quality of life. It is believed that during medical consultations, these patients report difficulties in dealing with the disease, its genetic aspects, and the adaptation to a life with visual impairment.

Psychotherapy groups are known to produce significant results in improving the quality of life of patients with anxiety, depression, or chronic pain. Based on interventions tailored for the GED population, it can be assumed that psychotherapy will improve the quality of life and emotional health of these patients and their families.

Objective

General:

To assess the psychosocial impacts of GED on patients, family members, and caregivers.

Specific:

- Investigate anxiety and depression in patients with GED (study 1)
- Review medical records of patients with GED (study 2)
- Evaluate the effect of a psychotherapy group for adults with GED on quality of life and emotional health (study 3)
- Evaluate the effect of a group psychotherapy intervention for parents of children/adolescents with LCA on indicators of emotional health, parental stress, acceptance, cognitive defusion, and mindfulness (study 4)
- Evaluate the effect of a psychotherapy group for family members of individuals with GED on quality of life and emotional health (study 5)

Method

Prospective cross-sectional and longitudinal study and medical record review.

Sample

The sample for the assessment of anxiety and depression will be obtained through convenience sampling, in contact with patients from the Instituto de Genética Ocular and Retina Brasil, with the aim of including patients with multiple conditions for a better understanding of the impact of each disease on anxiety and depression (study 1). The medical records to be reviewed will be those that involved joint care between psychologist and counselor (study 2). For the psychotherapy groups, the sample will be drawn from the interest of patients and family members in participating (study 3, 4, 5). Additionally, forty parents of children and adolescents with LCA will be included. Twenty will participate in psychotherapy groups, and twenty will be in the control group.

Inclusion Criteria

- Patients over 18 years old diagnosed with any disease leading to visual impairment
- Parents and family members over 18 years old of individuals with GED

Exclusion Criteria

- Children and adolescents with visual impairment
- Patients without a conclusive diagnosis of GED

Instruments

Hospital Anxiety and Depression Scale (HADS)

The HADS questionnaire was validated and translated into Portuguese (ZIGMOND; SNAITH, 1983; ZIGMOND; SNAITH, 1986; CASTRO et al., 2006). It consists of two subscales: the HADS Anxiety Scale (HADS-A) and the HADS Depression Scale (HADS-D). Each of the 7 anxiety questions and 7 depression questions is scored on a four-point scale (0-3), with final scores ranging from 0 to 21 for each subscale. The presence of anxiety or depression symptoms is classified as absent for scores below 7, possible between 8 and 10, and probable for scores above 11.

National Eye Institute – Vision Functioning Questionnaire 25 (NEI-VFQ 25)

The NEI-VFQ-25 is an instrument validated for the Brazilian population that evaluates vision-related quality of life. It contains 25 items covering the following domains: general health status (1 item); overall vision (1 item); eye pain (2 items); difficulty with near vision activities (3 items); difficulty with distance vision activities (3 items); limitations in social functioning due to vision (2 items); mental health problems due to vision (4 items); functional limitations due to vision (2 items); dependence on others due to vision (3 items); driving difficulties (2 items); difficulty with color vision (1 item); and difficulty with peripheral vision (1 item). Each subscale score is converted to a score between 0 to 100, with higher scores indicating better quality of life. (MANGIONE et al., 2001; SIMÃO et al., 2008)

Self-Reporting Questionnaire (SRQ-20)

The SRQ-20 was validated for the Brazilian population and contains 20 items assessing the presence of mental disorders. The questions are scored dichotomously, and a score of 7 points or higher indicates the presence of a mental disorder. The SRQ-20 serves as a screening tool but does not identify the specific mental disorder experienced by the patient. (SANTOS et al., 2011)

Freiburg Mindfulness Inventory (FMI)

The FMI assesses mindfulness through 14 questions. It was validated for the Brazilian population and is self-reported. Each question is answered on a Likert scale from 1 to 4. Higher scores indicate greater alignment with mindfulness practices. (HIRAYAMA et al., 2014)

Parental Stress Scale (EEPa)

The EEPa was validated and adapted for Brazil and contains 16 items, with eight related to parents' satisfaction with their parenting experience and eight related to parental stressors. Responses are made on a Likert scale, ranging from 0 (strongly disagree) to 4 (strongly agree). The total score is 64 points, and the higher the score, the greater the parental stress experienced by the participants. (BRITO; FARO, 2017)

Ethical Aspects

Data for study 1 will be collected through a questionnaire on the Google Forms platform. In order to complete it, the patient must read, consent, and agree to the Informed Consent Form for Research in a Virtual Environment (TCLE) (Annex 1).

For study 2, patients will be contacted for consent to review their medical records. The records can only be reviewed after the completion of the Informed Consent Form.

For studies 3, 4, and 5, patients wishing to participate in the psychotherapy group process must also read, consent, and agree to the Informed Consent Form in a Virtual Environment. If a participant withdraws from the research, they may continue with the psychotherapy process.

Participants may withdraw their data from the study at any time without providing a reason. The data collected will be used exclusively for the purposes of this research.

Procedures

To assess anxiety and depression in the GED population, a Google Forms questionnaire will be distributed through the social media channels of the Instituto de Genética Ocular and Retina Brasil over a 3-month period. Participants will be invited to join and will accept the consent form attached to the form itself (study 1).

The medical record review will be based on the collection of appointments between the medical advisor and the psychologist. The necessary data will be extracted from the medical records, and each case will be evaluated by the responsible parties for potential inclusion in the sample (study 2).

For studies 3, 4, and 5, participants will be invited via social media at the beginning of each semester. Interested patients will be screened by the responsible psychologists, and a battery of tests will be conducted for the initial evaluation. Afterward, 3 months of weekly psychotherapy groups, each lasting 1.5 hours, will take place. At the end of the group and two months after its conclusion, the same questionnaires will be administered to reassess the patients.

Data Processing and Analysis

For the data of each evaluation, the stages of acquisition, organization, cleaning, processing, and preparation for statistical analysis will be carried out according to the specifics of the method used.

The data will be compiled and analyzed using IBM® SPSS Statistics (version 2.7), and descriptive statistical analyses will be performed to check the means, standard deviations, and the Shapiro-Wilk test to verify normal distribution. If other statistical tests are necessary, they will be conducted.

Primary Outcome

Study 1: Determine the rates of anxiety and depression in the GED population.

Study 2: Understand the main psychosocial issues related to GED in ophthalmology consultations.

Studies 3, 4, and 5: Evaluate the effect of psychotherapy groups on the emotional health and quality of life of family members and parents of people with GED.

Secondary Outcome

Study 1: Investigate the relationship between anxiety and depression and the degree of disability, time of diagnosis, productive life, and rehabilitation.

Study 2: Identify potential interventions for negative emotional aspects perceived in consultations for people with GED.

Studies 3, 4, and 5: Understand the emotional aspects of parents and family members of people with GED and reflect on other possible interventions.

Risks and Benefits

The risks of this research are minimal. Anxiety and depression questionnaires may trigger unpleasant memories for the patients. As for the authorization for medical record review, it is believed that there will be no significant impact on individuals with

GED. Regarding the psychotherapy groups, contact with the patient's emotional suffering may also occur.

For both questionnaire administration and psychotherapy groups, patients will be evaluated and supported by the responsible psychologist, and any additional distress will be addressed individually.

The benefits of this research are numerous. By understanding the rates of anxiety and depression in the GED population, it will be possible to consider public policies aimed at improving these conditions. Understanding the main topics discussed in medical consultations can help create guidelines to serve this population. Psychotherapy groups may significantly improve participants' psychological well-being and quality of life.

Return of Benefits to the Studied Population

All data collected in this study will shed light on the psychosocial condition of people with GED and will assist in planning care for this population in different institutions. Moreover, it will contribute to the development of public policies based on real data about the characteristics of people with GED.

Detailed Financial Budget and Compensation

There will be no costs associated with conducting this research. The principal researchers will not be compensated, and there will be no need for resource allocation.

Timeline

Activities	**Period**
Approval by the Ethics Committee	September to November 2024
Sample Selection	November 2024 to February 2025
Data Collection	February 2025 to February 2027
Analysis, Description, and Discussion	February 2027 to July 2028
Conclusion	July to September 2028

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