Original article

Retinal Screening of Patients Suffering from Bardet – Biedl Syndrome Using Electroretinography

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Abstract

Background: Bardet-Biedl Syndrome (BBS) is a complex genetic disorder that affects various systems in the body, including the visual system. The aim of this study is to investigate the adverse effects of BBS on the retina using Electroretinography (ERG).
Material and Methods: In this case-control study, 10 BBS patients (6 males and 4 females) aged between 12 and 20 years were selected as the case group. Additionally, 10 age- and sex-matched healthy individuals with normal retinal function were included as the control group. ERG recordings were performed to measure the amplitude (μV) and latency (msec) of the ERG b-wave in both groups.
Results: The mean age of the case group was 15.4 ± 3.06 years, and it was 15 ± 2.78 years in the control group. The mean visual acuity in the case group was 0.49 ± 0.14 (LogMAR), significantly different from the control group with a mean visual acuity of 0 ± 0 (LogMAR). The difference in visual acuity was statistically significant between the two groups. Furthermore, the case group showed significantly lower amplitude and higher latency of the ERG b-wave peak compared to the control group ($p < 0.001$).
Conclusions: Bardet-Biedl Syndrome has adverse effects on the visual system, particularly the retina, resulting in a decrease in the amplitude and an increase in the latency of the ERG b-wave. These findings indicate impaired retinal function in BBS patients, highlighting the importance of early detection and management of retinal abnormalities in individuals affected by this rare genetic disorder.

Keywords: Bardet-Biedl Syndrome; Electroretinography (ERG), Visual System; Retina; Adverse; Effects, Impaired Retinal Function.

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Introduction

Bardet-Biedl Syndrome (BBS) is a complex genetic disorder with a wide range of clinical features, affecting various systems in the body. Among the affected systems, the visual system can exhibit abnormalities, leading to vision problems and retinal degeneration. To assess the visual system in BBS patients, visual system electrophysiology is employed, involving tests such as Electrooculogram (EOG), Visual Evoked Potential (VEP), and Electroretinogram (ERG).

Sanaie et al. (2014) conducted a study to investigate the effects of ocular toxoplasmosis on the visual system, specifically the retina, in patients using EOG. They observed significant differences between the Arden Index (AI) of EOG in case and control groups, indicating pathological changes in the retina, particularly the retinal pigment epithelium[1]. Similarly, Sanaz Abdolalizadeh and colleagues (2022) evaluated the retinal status of patients treated with antiseizure medications using ERG. Their findings revealed that these medications affected the retina, leading to a reduction in the amplitude of ERG b-wave peak[2]. Furthermore, Shushtarian (2020) investigated patients with albinism using flash and pattern reversal checkerboard VEP, revealing visual pathway disturbances through delays in VEP latency, especially the P100 peak [3].

Numerous studies [4-37] have explored the visual system and its relationship to various factors such as occupational vibration, hydroxychloroquine retinal toxicity, and the effects of antiepileptic drugs on the retina [4-37] . Ojani et al. (2021) specifically investigated the visual pathway abnormalities in BBS patients using VEP, highlighting significant findings[38]. However, the precise mechanisms underlying the visual complications associated with BBS remain an area of ongoing research.

Given the existing literature and the importance of understanding the visual implications of BBS, a research work was planned to explore potential pathological changes in BBS patients using ERG techniques. The study included ten BBS patients (6 males and 4 females) with reduced visual acuity, a common characteristic of the disease, in the case group. The results obtained from the two groups were compared to identify potential differences in retinal function between BBS patients and healthy individuals. This research aims to contribute to the understanding of visual system involvement in Bardet-Biedl Syndrome, aiding in early detection and management of visual impairments in affected individuals. However, due to the limited sample size, further studies with larger cohorts are needed to validate and expand on these findings. The combined data from multiple studies may provide more comprehensive insights into the retinal abnormalities and functional changes in BBS patients, leading to improved management and potential therapeutic interventions for preserving vision in affected individuals.

Material and Methods

In this case-control study, we selected ten patients with Bardet-Biedl Syndrome (BBS) as the case group, comprising six males and four females, totaling 20 eyes. The patients in the case group were within the age range of 12 to 20 years. For the purpose of assessing retinal function, flash-type Electroretinogram (ERG) recordings were performed using the Mangoni device. We measured both the amplitude (μV) and latency (msec) of the ERG b-wave to evaluate the retinal response to light stimulation.

To conduct the ERG recordings, three electrodes were utilized to connect the Mangoni device to the patients. The active electrode was placed

Table 1: Demographic findings in case and control groups

Variable	Number of participants	Groups (Mean ± SD)		P value
	Case		Control	
Age	20	15.4 ± 3.06	15 ± 2.78	0.796 **
sex	20	-	-	0.653 *
VA LogMar	20	0.49 ± 0.14	0 ± 0	0.000 **

*Based on chi-square

**Based on Mann-Whitney Test

on the sclera, the reference electrode on the ear pinna, and the ground electrode on the forehead. This configuration ensured accurate and reliable measurements of retinal activity during the ERG procedure.

As a comparison group, we included ten healthy subjects who were of similar age and sex to the patients in the case group. These individuals had normal retinal function, and they served as a control group for the study.

The aim of this research work was to explore potential pathological changes in the retinal function of BBS patients using ERG techniques. By comparing the ERG findings between the case and control groups, we sought to gain insights into the retinal abnormalities associated with Bardet-Biedl Syndrome and potentially contribute to improved management and intervention strategies for vision preservation in affected individuals.

Results

The results of the study are presented in Table 1 and Table 2. In Table 1, the demographic findings show that there is no statistically significant difference between the case and control groups in terms of age (P value = 0.796) and sex (P value = 0.653). However, there is a highly significant difference in the case of best-corrected visual acuity (BCVA) between the two groups (P value < 0.001). This indicates that the visual acuity of the BBS patients (case group) is significantly different from that of the healthy subjects (control group), as expected due to the retinal abnormalities associated with BBS.

Table 2 presents the measurements of the mean amplitude and latency of the ERG b-wave in both the case and control groups. The results indicate significant differences in both amplitude and latency between the case and control groups (P value = 0). The reduced amplitude and increased latency of the ERG b-wave in the BBS patients (case group) compared to the healthy subjects (control group) suggest impaired retinal function in BBS, which is consistent with previous research findings. These ERG abnormalities further validate the visual complications associated with Bardet-Biedl Syndrome and highlight the importance of early detection and management of retinal degeneration in affected individuals.

Overall, the study findings contribute valuable insights into the retinal manifestations of BBS and provide a basis for potential interventions and therapies aimed at preserving vision and improving the quality of life for individuals living with this rare genetic disorder.

Table 2: Measurements of mean ± S.D in case of amplitude and latency of ERG, b wave of case and control groups

Variable	Number of participants	groups (Mean ± SD)		P value *
		Control	Case	
Latency (msec)	11	43.3 ± 2.9	51.4 ± 2.9	0
Amplitude (μv)	11	111.9 ± 8.24	32.05 ± 7.18	0

* Based on Mann-Whitney U Test

Discussions

Bardet-Biedl Syndrome is a complex genetic disorder that can affect various systems in the body, including the retina of the visual system. In this research work, we investigated the retinal function in BBS patients using Electroretinography (ERG) and found significant changes in the amplitude and latency of the ERG b-wave. These findings are indicative of retinal dystrophy in the visual system [38, 39].

Our results are consistent with previous studies conducted by other researchers in related areas. Meng X and colleagues (2021) conducted research in China, where they selected 12 BBS patients from 10 Chinese families and tested them using full-field flash ERG. The results revealed severely damaged cone-rod cells in the retina, further supporting the presence of retinal dystrophy in BBS patients [39].

Similarly, Tabl MA (2020) studied three female patients with BBS who complained of progressive visual loss. The flash ERG performed on these patients showed a severe form of rod-cone dystrophy, a type of pigmentary retinal dystrophy. This finding aligns with our observation of retinal dystrophy in BBS patients[40].

Conclusion

In conclusion, our research work provides evidence that patients with Bardet-Biedl Syndrome suffer from retinal dystrophy, which is evident through the changes observed in the ERG b-wave. The reduction in amplitude and prolongation of latency of the ERG b-wave indicate impaired retinal function in BBS patients. These findings are consistent with other studies in the field and highlight the importance of regular retinal screening in BBS patients to detect retinal abnormalities early on. Early diagnosis and intervention may lead to improved management and potentially better outcomes for preserving vision in individuals affected by this rare genetic disorder. Further research is needed to explore therapeutic interventions aimed at managing retinal dystrophy in BBS patients and improving their visual outcomes.

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Footnotes and Financial Disclosures

Conflict of interest:

The authors have no conflict of interest with the subject matter of the present manuscript.