An overview of the bardet-biedl syndrome

Health & Medicine, Disease

An 11-year-old boy was admitted to a hospital in Turkey due to complaints about “ loss of vision, speech deficit, learning difficulty, poor balance, and ataxic gait”. Results of the physical examination showed that the boy manifested the following special features: facial dysmorphism, visual problems that included nystagmus or rapid, involuntary movement of the eyes from side to side, poor coordination, being overweight, an extra digit on the left foot, mild mental retardation, among other distinctive characteristics. What the authors had presented was the usual case of a developmental disorder called Bardet-Biedl Syndrome (BBS). The WorldHealthOrganization (WHO) developed the International Classification of Functioning, Disability, and Health (ICF), which is a news classification system that focuses on the components of health. The ICF also emphasizes the relationship between the health condition and contextual factors, as illustrated in Figure 1 using the ICF model.

This document aims to explain the Bardet-Biedl Syndrome in terms of the patients’ body structure & functions, activities, participation, and their limitations. General Description Bardet-Biedl syndrome is an autosomal recessive condition or an inheritable disease involving non-dominant chromosomes. It was first described by George Bardet and Alfred Biedl in 1920. This syndrome has a prevalence rate of 1/160, 000. The cardinal features of BBS as seen in numerous case reports include retinal dystrophy, obesity, cognitive deficit, hypogonadism, polydactyly, mental retardation, and renal dysfunction. Arguments regarding the accuracy of the cardinal features emerged and consequently prompted Beales et al. to conduct a

study that aimed to clarify and determine the major criteria for proper diagnosis of the syndrome.

According to the results, individuals diagnosed with BBS must have the following cardinal features:

1. Visual disorders. The individual suffers from rod-cone dystrophy, also known as atypical retinitis pigmentosa, which is characterized by “ night blindness”, loss of peripheral or tunnel vision, loss of central vision inchildhood, and adolescence, and “ severe visual impairment by adulthood”.
2. Limb defects. A widespread condition among individuals with BBS is postaxial polydactyly or having extra accessory digits in the hands or feet. Some are born with only one extra finger or toe, but in other cases, the condition is “ present in all four limbs”. There were also cases that extra digits were only found in both hands or in both feet. In thecase studymentioned above, the 11-year-old boy had an extra digit on the left foot.
3. Height and weight. In the survey conducted by Beales et al. (1999) among children who developed BBS with an average age of 9 years old, the average height of males was 1. 73 m, which was significantly lower than the average height of males in the general population which was 1. 76 m; the average height of females, on the other hand, measured 1. 62m—a number that was not significantly different from the average height of females in the general population that measured 1. 63 m. When it comes to weight, Bardet-Biedl syndrome is widely linked to being obese or overweight. Obesity was considered by a

number of researchers as one of the accepted major criteria of BBS. Uzun et al. (2007) even addressed obesity in his case study as “ one of the most common features of BBS”.

4. Education. Individuals with BBS exhibit learning difficulties which “ can range from mild cognitive disability to mental retardation”. However, Green et al. (as cited in Hrynchak, 2000) questioned this feature and found that “ when appropriate verbal and performance IQ tests were given, only a minority of patients were found to have a cognitive deficit”.
5. Renal tract abnormalities. The kidney is one of the affected body organs when a person is developed to have BBS. Most patients were found to have structural abnormalities. Kidneyfailurehas been one of the leading causes of death among individuals with the syndrome. The symptoms mentioned above are considered the major criteria; however, the descriptions of BBS are not limited to these items.

Hypogonadism was considered by some researchers as one of the key features; it is described as a structural deficiency characterized by Beales et al. (1999) as having small penises buried in adipose tissue. These concrete descriptions of the major criteria or symptoms for proper diagnosis of the Bardet-Biedl syndrome are one thing; behavioral patterns in patients with the syndrome are another. Beales et al. (1999) were able to include brief descriptions of BBS patients. These “ difficult” behaviors include “ emotional immaturity, frequent volatile outbursts, and poor reasoning”. They also claim boldly that “ all BBS children preferred fix routine. Barnet et al. (2002) extensively studied the behavior of individuals with BBS and found, apart

from the aforementioned description by Beales et al. (1999), that children with BBS felt “ withdrawn”, “ anxious”, or even “ depressed”. One of the main arguments of the article by Barnet et al. (2002) was the existence of a “ behavior phenotype” or a behavior gene. One revelation that fuelled his speculations was the emergence of closely similar behavior patterns of individuals born with BBS.

That is simply one way to explain behavior. On the other hand, we can find links between affect, behavior, and cognition to explain why they act the way they do. Consider feelings of withdrawal, anxiety, anddepression. These are all negative feelings indirectly pointed towards the self. A person with BBS would want to withdraw from the normal population because of possible shame or fear of encountering ridicule due to other people’s inconsiderate and usually unkind remarks. Anxiety might be felt due to a developed fear of being ridiculed. Depression, which is most probably the worst thing a child can feel, might have developed due to low self-esteem and self-worth. They would probably make comparisons between people like themselves who have impairments and people who are declared by society to be normal. One of the most notable behavior patterns observed was traces of obsessive-compulsive behavior; as with individuals who are diagnosed with autism, which is another developmental disorder linked to the cognitive deficit, those with BBS are most comfortable with a “ fixed routine”. Activities & Limitations in Activities It is an observable fact that the features of Bardet-Biedl syndrome entail a lot of limitations in learning and applying knowledge. First, once the visual problems emerge, they interfere with visual learning, which is an important facet of education since most instructional media are

visual. Hrynchak (2000), however, claimed: “ early identification of the visual disorder and appropriate educational intervention can be of great benefit in people with this condition”. In the study of Beales et al. (1999), half of the patients received education in a “ special school”; some even further pursued their education and earned university degrees. Visual problems also hamper the day-to-day activities of older patients. Hrynchak (2000) presented the case of a 20-year-old white man with the condition who worked as a janitor. He complained about “ decreasing visual acuity”, specifically, “ missing debris while sweeping”. “ He also had reduced ability to see the fine print, difficulty seeing the credits on television, mobility problems (especially judging depth), and difficulty adapting to changing illumination conditions”.

Hrynchak (2000) also mentioned that majority of individuals become “ legally blind” before the age of 30. Obesity also entails a lot of health risks including heart disease. This affects the amount of emotional and physicalstressthe individual is capable of doing. Ataxia, which is characterized by poor coordination, was also found in patients with BBS. This affects how people with BBS move properly. It is probable that obesity is prevalent in some cases because of ataxia, which hinders activities that need proper coordination such as sports and other physical exercises. There is also the matter of speech deficit. Beales et al. (1999) pointed out that there is a problem in language use and vocabulary is limited due to learning difficulties. Participation & Limitations in Participation The case presented by Hrynchak (2000) shows the ability of individuals with BBS to function effectively in society as a working individual. However, Beales et al. (1999)

described that “ adult patients are often disinhibited and appear to recognize social cues”. This means that they often find it difficult to relate and mingle with other people. Perhaps this is a consequence of feelings of depression and anxiety in childhood brought about by possible ridicule and rejection among peers. Fatherhood is a major societal role that males will not get to play. Due to hypogonadism, males are infertile and are unable to procreate. According to Hrynchak (2000), “ there have been no reports of a man with Bardet-Biedl syndrome having fathered children. ” Treatment & Outcome It is an unfortunate matter that this genetic disorder has no known holistic treatment according to The Foundation Fighting Blindness (2000).

To perhaps decrease the complications of the condition, one must address each symptom separately. For visual problems, an eye specialist should be consulted to see how the vision could be improved or what adjustments should be made by the patient or by the people around him. Hrynchak (2000) suggested the use of “ field expansion devices” or a guide dog. Poor vision might hamper an individual’s ability to learn, hence, cognitive disabilities. Learning disabilities, on the other hand, can be overcome with early intervention and had not been a hindrance to those who completed university degrees. For renal abnormalities or kidney problems, one can approach a nephrologist or “ a physician that specializes in kidney diseases”. Kidney diseases are usually not detected until the patient undergoes radiological testing or x-ray after being diagnosed with BBS. Take note that the leading cause of death among BBS patients is renal or kidney failure.

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