GACKA, Piotr, DOŁĘGA, Marcin, GOŁDA, Joanna, MĘŻYK, Julia, SNOPKOWSKA, Aleksandra and MUSIALSKA, Dominika. Bobble-Head Doll Syndrome: Understanding a Rare Neurological Enigma. Journal of Education, Health and Sport. 2024;69:55355. eISSN 2391-8306.
https://dx.doi.org/10.12775/JEHS.2024.69.55355
https://apcz.umk.pl/JEHS/article/view/55355

The journal has had 40 points in Minister of Science and Higher Education of Poland parametric evaluation. Annex to the announcement of the Minister of Education and Science of 05.01.2024 No. 32318. Has a Journal's Unique Identifier: 201159. Scientific disciplines assigned: Physical culture sciences (Field of medical and health sciences); Health Sciences (Field of medical and health sciences).
Punkty Ministerialne 40 punktów. Załącznik do komunikatu Ministra Nauki i Szkolnictwa Wyższego z dnia 05.01.2024 Lp. 32318. Posiada Unikatowy Identyfikator Czasopisma: 201159. Przypisane dyscypliny naukowe: Nauki o kulturze fizycznej (Dziedzina nauk medycznych i nauk o zdrowiu); Nauki o zdrowiu (Dziedzina nauk medycznych i nauk o zdrowiu).
The authors declare that there is no conflict of interests regarding the publication of this paper.
Received: 23.08.2024. Revised: 24.09.2024. Accepted: 30.09.2024. Published: 02.10.2024.

Short Article

Bobble-Head Doll Syndrome: Understanding a Rare Neurological Enigma

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Abstract

Bobble-Head Doll Syndrome (BHDS) is a rare neurological disorder characterized by repetitive, involuntary, and stereotypical head movements, resembling the motion of a bobble-head doll, often accompanied by various neurological and endocrinological symptoms. Although primarily observed in pediatric patients, BobbleHead Doll Syndrome can also manifest in adults. The pathophysiology remains unclear, but it is often associated with suprasellar cysts, third ventricle cysts, or aqueductal stenosis. Diagnosis typically involves magnetic resonance imaging to visualize the cystic lesions and assess cerebrospinal fluid flow. Additional diagnostic modalities include computed tomography and electromyography. Treatment primarily involves neurosurgical intervention, such as ventriculocystostomy or ventriculocystocisternostomy, aimed at relieving cerebrospinal fluid obstruction. Despite the challenges in diagnosis and management, early recognition and appropriate treatment offer promising outcomes, including symptom resolution and potential recovery. This review provides a comprehensive overview of Bobble-Head Doll Syndrome, including its clinical presentation, pathogenesis, diagnostic approach, and therapeutic strategies, highlighting the need for further research to elucidate its underlying mechanisms and optimize therapeutic strategies.

Keywords: Bobble-Head Doll Syndrome; BHDS; neurological disorder; neuroimaging; children; cystic lesions.

Abbreviations

BHDS - Bobble-Head Doll Syndrome
MRI - Magnetic Resonance Imaging
CSF - Cerebrospinal Fluid
CT - Computed Tomography
EMG - Electromyography
USG – Ultrasonography

Introduction

The rare neurological condition known as Bobble-Head Doll Syndrome (BHDS) was first identified in 1966 by Brendon et al [1]. The initial patients observed with BHDS were children with a third ventricle cyst and obstructive hydrocephalus. What caught attention was their unique to-and-fro bobbing or nodding of the head and torso, resembling the movements of a doll with weighted heads resting on a coiled spring, hence the name of the disease [1]. BHDS is characterized by repetitive, involuntary, and stereotypical forward and backward head movements or, less commonly, sideways movements [2]. Bobble-Head Doll Syndrome is an extremely rare movement disorder, with only about 70 cases of BHDS described so far, most of which were pediatric patients [3]. Typically, BHDS is a pediatric issue, as symptoms appear in early childhood, between 1 month and 20 years of age [3]. However, characteristic pathological head movements may also occur in older individuals. Olvera-Castro et al. described a case of BHDS in an 80-year-old patient in 2017 [4]. The disease itself is a complex neurological disorder, where early diagnosis and treatment using available neurosurgical techniques offer promising results, even allowing for complete recovery [5, 6, 7].

Symptoms

Various symptoms can occur in the course of Bobble-Head Doll Syndrome. The main symptom present in all patients with the described syndrome is head bobbing forward or backward (yes-yes type), and sometimes from side to side (no-no type), at a frequency of 2-3 Hz. This symptom may occur continuously or with intermittent breaks. Head bobbing typically diminishes during nighttime, is constrained by focus, or can be restrained by the patient's volition. Head bobbing may intensify in stressful situations [8]. An additional symptom is tremors of the torso or limbs [9].

Patients may struggle with engaging in diverse activities, such as walking, often displaying a wide-based gait linked with aberrant movements of the torso and lower limbs, mirroring the frequency of abnormal head movements [10]. Some patients exhibit static and dynamic ataxia, significantly limiting mobility such as walking in a straight line or climbing stairs, and sometimes dysdiadochokinesia is reported [5, 11]. Many patients present with macrocephaly, which may be associated with periodic headaches and vomiting [3]. Insomnia occurs in some patients [12]. Occasionally, eating disorders such as bulimia are described in the course of the disease [12]. Reduced visual acuity is observed, seemingly related to pathologies of the optic nerve and/or visual pathway. The disease may also manifest as attention deficit, developmental delay, and moderate intellectual impairment [13]. Developmental disorders such as premature puberty with significantly elevated testosterone levels, short stature (below the 3rd percentile), or delayed puberty can also occur, along with possible diagnosis of enuresis [14, 15, 16].

Pathogenesis

The pathophysiology and anatomical causes of Bobble-Head Doll Syndrome remain unclear, but suggested mechanisms exist in the available literature [3].

Most cases of BHDS are associated with the presence of suprasellar arachnoid cysts, third ventricle cysts, or aqueductal stenosis [17]. Less common causes include abnormal positioning or malfunctioning of the valve, other cysts obstructing cerebrospinal fluid (CSF) flow through the third ventricle, such as septum pellucidum cyst, cavum velum interpositum cyst, and cystic choroid plexus papilloma, or masses invading the third ventricle, such as craniopharyngioma and chronic hydrocephalus [10].

Suprasellar arachnoid cysts are the most common cause of BHDS [3]. They arise from congenital thickening or non-perforation of the Liliequist membrane, but can also occur later in life after inflammation of the arachnoidea [6]. The non-perforated membrane forms a kind of barrier hindering the flow of CSF, and as a result, it dilates, forming diverticula of the arachnoid membrane [6]. Over time, such a diverticulum may enlarge and its size may compress the third ventricle, impeding CSF flow through the Monro foramen, thereby enlarging the third ventricle [6, 18]. Enlargement of the third ventricle, leading to compression of the hypothalamus, causes endocrinological disorders, including growth hormone deficiency, gonadotropin deficiency, thyrotropin deficiency, vasopressin deficiency, and fluctuations in body temperature, commonly found in individuals with hydrocephalus [19, 20, 21]. The distinctive head movements observed in BHDS enable the shifting of the dome away from the Monro foramen, thereby aiding in the flow of CSF. It should be noted that some suprasellar arachnoid cysts maintain communication with the pontine cistern, which consequently leads to fluctuations in their sizes. It is believed that head movements in such patients lead to periodic emptying of the cysts, alleviating symptoms associated with hydrocephalus [6, 10].

The cyst or another lesion could potentially compress the mediodorsal nucleus of the thalamus within the paraventricular region, consequently activating the diencephalic extrapyramidal pathways, encompassing the rubrotegmentospinal and reticulospinal tracts. These pathways interface with motor neurons that control the neck muscles, suggesting that their stimulation may initiate head movements [10].

Diagnosis

The main diagnostic method for the described condition is brain magnetic resonance imaging (MRI). Despite a similar clinical picture, various pathological changes within the brain are described. The image may show an arachnoid cyst in the suprasellar region of the third ventricle and obstructive hydrocephalus [9]. Sometimes, a thin-walled cyst in the suprasellar and sellar area may be responsible for the disease [10]. In the course of Bobble-Head Doll Syndrome, subsequent enlargement of the fourth ventricle due to an enlarging cyst can also be observed [22]. In some cases, a cyst of the septum pellucidum may be detected [5]. Much less frequently, MRI may describe pedunculated cystic changes in the anterior part of the third ventricle near the Monro foramen with simultaneous enlargement of the choroid plexus of the lateral and third ventricles [23]. A case has also been described in which, in addition to the presence of a suprasellar cyst, there was secondary compression of the vault, corpus callosum, and pons with enlargement of the lateral ventricle and secondary obstructive hydrocephalus [16]. Sometimes, pathological changes near the pineal gland are observed [11]. MR imaging also plays an important role in assessing CSF flow. In MR FLAIR sequence, CSF flow towards the cyst may sometimes be visualized [24].

Another significant diagnostic method is computed tomography (CT) of the head, although currently brain MRI is much more commonly performed due to its less invasive nature and greater accuracy in detecting changes. CT cisternography appears to be a significant examination in which the flow of CSF can be evaluated. Disorders

in CSF flow between the subdural space and the subarachnoid space may be described in the course of the disease [13].

During the prenatal period, one of the important examinations is ultrasonography (USG). In the course of the described disease, hydrocephalus and arachnoid cysts in the posterior fossa may be visualized [22]. While ultrasound is non-invasive and widely available, it represents just one aspect of prenatal diagnostics, and confirming changes observed in it necessitates further imaging examinations, typically brain MRI.

One of the additional diagnostic methods is electromyography (EMG). Through EMG examination of muscles such as the trapezius, head bobbing can be detected, which is the main symptom in the course of Bobble-Head Doll Syndrome [5].

Frequently, diagnosis involves a thorough assessment of the ocular system and visual pathways. In ophthalmic examination in the course of the described condition, bilateral pallor of the optic disc without optic disc edema is often observed [25]. Occasionally, bilateral optic disc edema occurs along with secondary optic nerve atrophy, and the light reflex in patients may be slightly delayed [11]. In visual evoked potentials examination, a decrease in amplitude and conduction along the anterior part of the visual pathway suggests axonopathies. Prolongation of the P100 latency is associated with secondary demyelinating processes [11].

Additionally, attention is drawn to the presence of concurrent endocrine disorders. The specific location of the lesion within the intricate network of the hypothalamic-pituitary area has the potential to trigger a diverse array of endocrinological dysfunctions. These may manifest as deficiencies in essential hormones such as growth hormone, thyrotropin, gonadotropin, and vasopressin [16].

In neurological examination, increased deep tendon reflexes are diagnosed in some patients [13].

Treatment

The main method of treating the cause of Bobble-Head Doll Syndrome is neurosurgical operation. Symptoms usually resolve spontaneously after cyst removal. Historically, the primary treatment modalities included permanent ventriculoperitoneal or cystoperitoneal drainage, as well as open marsupialization [24]. Endoscopic fenestration of the cyst with Ventriculocystocisternostomy appears to be an effective and recommended treatment method for most patients, but in some cases, it may be considered impossible due to the possibility of unfavorable anatomical conditions or the risk of damage to the basal cerebral artery [16, 24]. Neuroendoscopic ventriculocystostomy is also regarded as a treatment method. Throughout the treatment process, wide fenestration of the cyst wall may be conducted, enabling visualization of the pulsation of the basal cerebral artery and its branches with the endoscope. During the procedure, often with the use of a special balloon, the connection between the cyst and the basal prepontine cistern is widened [26]. Occasionally, cystoventriculoperitoneal shunt implantation is executed under neuroendoscopic supervision, particularly if hydrocephalus is closely linked to the cyst, resulting in elevated intracranial pressure [24]. However, utilizing this valve entails a heightened risk of complications, such as infection.

Conclusions

Bobble-Head Doll Syndrome presents a complex clinical challenge due to its rare occurrence and diverse symptomatology. Despite the limited understanding of its pathogenesis, advancements in neuroimaging and surgical techniques have facilitated diagnosis and treatment. Early recognition of BHDS is crucial for

implementing timely interventions, which predominantly involve neurosurgical procedures aimed at relieving cystic lesions and restoring CSF dynamics. However, the variability in clinical manifestations underscores the importance of a multidisciplinary approach involving neurologists, neurosurgeons, ophthalmologists, and endocrinologists for comprehensive management. Further research is warranted to elucidate the underlying mechanisms of BHDS and optimize therapeutic strategies. Through continued collaboration and innovation, the medical community can enhance the care and outcomes of individuals affected by this rare neurological disorder.

Disclosure

Author's Contribution:

Conceptualization, PG and MD; methodology, PG and MD; check, PG; formal analysis, PG, MD, JG and AS; resources, PG, MD, AS, JM and DM; data curation, PG; writing - rough preparation, PG, MD and JM; writing - review and editing, PG, MD, JG, JM, AS and DM; visualization, PG and MD; supervision, PG; project administration, PG; All authors have read and agreed with the published version of the manuscript.

Funding statement: Not applicable.

Acknowledgements: None.

Conflict of Interest Statement: The Authors declare that there are no competing interests.

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