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Case Report

Reading disability due to an ocular motor disorder: A case of an adolescent girl with a previous diagnosis of dyslexia

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Abstract

Dyslexia is a reading disability characterized by difficulties with accurate and/or fluent word recognition, which are thought to stem from a phonological processing impairment. Herein we report the case of a 13-year-old girl who received the diagnosis of dyslexia at age 12 years. We considered this diagnosis to be incorrect because her reading difficulty was caused by a spontaneously repeated eye movement toward the vertical direction; the eyes were likely to show slow, upward drifts followed by quick downward movement at the physical examination, and the amplitude of the downward movement was increased when she changed eye positions to look at the upper direction in the evaluation of the eye tracker. Although we considered there was the possibility that the spontaneously repeated eye movement was classified as the spontaneous downbeat nystagmus, the eye tracker showed the transition of the gaze starting from and returning to was inconsistent with nystagmus, and we concluded that the term of nystagmus like abnormal eye movement was appropriate for the expression of the spontaneously repeated eye movement. There was no apparent abnormality on head magnetic resonance imaging (MRI), and whole exome sequencing showed no known candidate genes to explain the cerebellar dysfunction. An accumulation of similar cases in the future should help elucidate the pathomechanism observed in this case, and we should fully pay attention to evaluate the neurological aspects of the patients before settling on the diagnosis of dyslexia.

Keywords: Dyslexia; Reading disability; Nystagmus like abnormal eye movement; *DNAAF2*

1. Introduction

Dyslexia is defined by the Diagnostic and Statistical Manual of Mental Disorders fifth edition (DSM-5) as a specific learning disability caused by neurological impairment [1]. The core symptom of dyslexia is the inability to read accurately and/or fluently; the cause, such as mental retardation or delayed cognitive development, may not be immediately apparent. We experienced the case of a 13-year-old girl whose reading difficulty was caused by a spontaneously repeated eye movement toward the vertical direction. We present the medical course and emphasize the importance of evaluating the neurological aspects sufficiently before settling on the diagnosis of dyslexia.

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2. Case

A 13-year-old girl presented to our outpatient department due to difficulties with reading. She had no past medical or family history. At age 12 years, her teachers pointed out that she seemed to be having difficulty reading Japanese words. She was apparently guessing the meaning of the sentences based on a rough impression of the meaning of the individual words and numbers she was seeing. Thereafter, she was referred to the outpatient department at another hospital, but her eyes, visual acuity, and eye movements were found to be normal. The neurological findings and head MRI revealed no abnormality (Fig. 1).

The result of WISC-IV suggested a significant dissociation between VCI and the other inspection scores (FSIQ:74, VCI:101, PRI:76, WMI:79, PSI:50). Based on these results, dyslexia was diagnosed, and the patient was referred to our outpatient department for further examination and treatment. In our outpatient department, a spontaneously repeated eye movement toward the vertical direction; the movement that were likely to drift up slowly and bring back to the fixation target by a quickly corrective downward saccade, was always observed at primary position. Although the eyes horizontally showed no limited range of motion with the same degree of nystagmus, at the left or right ends, the spontaneously repeated eye movement was considered to be almost disappeared. As to the vertical direction, the eyes also showed no limited range of motion, but the amplitude of the spontaneously repeated eye movement was increased when she changed eye positions to look at the upper direction. An evaluation using the eye tracker (Tobii) showed her eye positions intensely fell down to the lower direction of the screen after being asked to look at the upper left or right (Fig. 2).

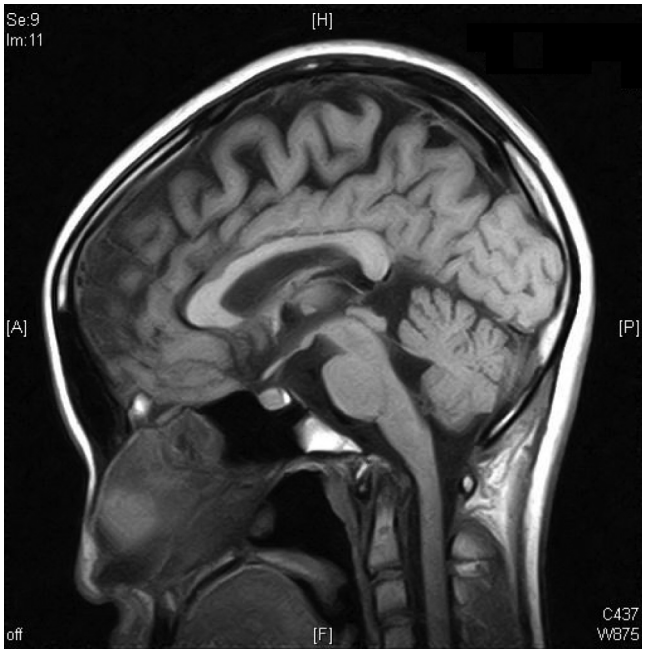

Fig. 1. T1-weighted sagittal image of the head MRI. This image shows no obvious abnormal findings on cerebellum, brain stem, and cerebrum.

She revealed no other symptom of neurological abnormality including ataxia. She confessed that she had concealed this tendency in her eye movements despite the difficulties that this was causing in her daily life. She explained that she was every frightened when getting on an escalator or walking down a flight of stairs from early childhood and had actually suffered a finger bone fracture twice while playing sports due to her disability. Although we performed whole exome sequencing using the trio-based approach (the patient, her mother, and her father) with saliva samples, the results revealed no known gene mutation associated with ocular motor disorders. Although we considered an evaluation by electrooculogram was necessary for further investigation of the eye movement, we could not gain the consent of the ophthalmic evaluation from the parents. Although we were unable to diagnose her condition, we considered it appropriate to provide the patient with support to ameliorate her reading disability and asked her school for help to lighten the difficulties of recognizing characters including allowing her to use an extended copy of test papers, a marker pen, and more time for taking tests. After this help was provided, her learning difficulties decreased, and she succeeded in passing the entrance examination for the top high school of her choice.

3. Discussion

Cases of reading disability due to ocular motor disorder are likely to be misdiagnosed as dyslexia, especially if there are no apparent neurological disorders or delayed cognitive development. The neurological aspects of such as case should be sufficiently assessed before settling on the diagnosis of dyslexia.

Although we were unable to determine whether the patient had a phonological processing impairment in addition to her ocular impairment, we did not consider her symptoms to be indicative of the neurological impairment typical of dyslexia because she was able to write Chinese characters accurately despite her severe reading disability (Fig. 3).

We considered there was the possibility that the spontaneously repeated eye movement was classified as the spontaneous downbeat nystagmus from the findings of the physical examination. In addition, the amplitude of the spontaneous eye movement became apparently more intense when she changed eye position to look in the direction of the slow phase, and we considered the

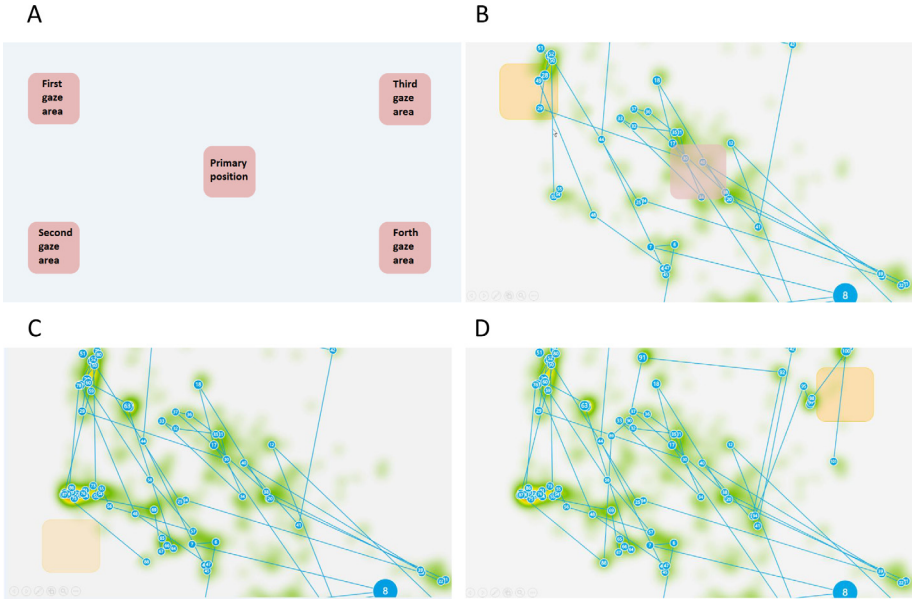

Fig. 2. A shows the contents of tasks evaluated by using the eye tracker (Tobii). The evaluation was carried out by viewing the screen from the front. The distance from the screen to the eyes was about 40 cm. Although the face of the patient was not fixed, the presumed visual angle of the inspection screen was considered about 15 degrees from the center to the edge. First, the patient was instructed to look at the central square of primary position (4 cm × 4 cm in size), and after that, to look at the square of the four corners (4 cm × 4 cm in size) in the order listed every 5 s. The number in the circle represents the order of movement of the patient's gaze. An increase in the area of the circle indicates a longer interval until the subject glances at the corresponding point. B shows the gaze points from the central square of primary position to the upper left square, C shows from the upper left square to the lower left square, and D shows from the lower left square to the upper right square. The figure of gaze points from the upper right square to the lower right square is omitted because the transition of the gaze was similar to the movement observed in C. As the result of the evaluation, the amplitude of the downward movement was increased after being asked to look at the upper left or right square, while the gaze points were relatively kept around the square after being asked to look at the lower left or right.

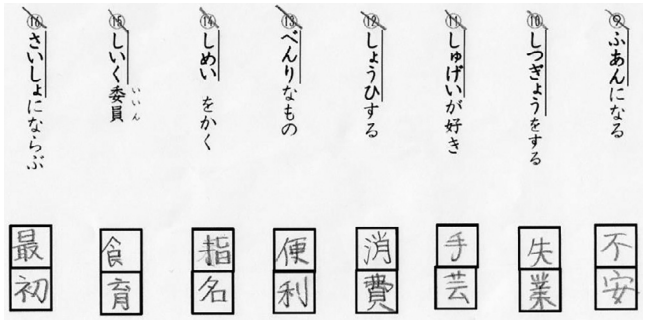

Fig. 3. Chinese characters written by the patient. The patient was able to write the characters accurately and neatly using the correct stroke order.

phenomenon was compatible with the spontaneous downbeat nystagmus based on the Alexander's law.

On the other hand, the results of the eye tracker indicated the findings that were inconsistent with nystagmus. This is because, when the patient was instructed to look at the upper left or right square in Fig. 2B or 2D, the repeated movements of gaze must start from the upper square area or nearby, and move to the lower directions, then return to the upper left or right square or pass there, if the eye movement was classified as nystagmus. The results of the eye tracker revealed that the transition of the gaze starting from and returning to the upper left or right square or nearby was extremely few.

Additionally, although previous studies have presented the downbeat nystagmus is associated with the damage of Purkinje cells contained in flocculus and paraflocculus, the caudal portions of the cerebellar vermis [2], and often clinically associated with the Arnold-Chiari malformation and spinocerebellar degeneration [3], no abnormality was apparent in the range that can be evaluated by the head MRI. Taken together, we concluded that the term of nystagmus like abnormal eye movement was appropriate for the expression of the spontaneously repeated eye movement.

Among the possible candidate genes, the compound heterozygote nonsynonymous single nucleotide variation (SNV), which is considered to be related to primary ciliary dyskinesia, was found in *DNAAF2*: NM_0010083908:c. [2032A > G];[122A > C] [4]. The variant of *DNAAF2* in this study is not registered in The Human Gene Mutation Database. The interaction between DYX1C1 and DNAAF2 is essential for the preassembly of the outer and inner dynein arms, which power the ciliary function [5,6]. Impairment of this function is also considered to be the main cause of Joubert syndrome, which is often associated with abnormalities

of the cerebellar vermis and eye movement [7]. Primary ciliary dyskinesia, associated with the *DNAAF2* mutation, may lead to the dysfunction of the cerebellar vermis, and is a candidate cause of the ocular motor disorder in this case. An accumulation of similar cases should help elucidate the pathomechanism observed in our patient.

Ethical considerations

We obtained written informed consent for this report from the patient's parents and the Institutional Review Boards of the National Center Institute for Child Health and Development (Tokyo, Japan; IRB number: 630) approved the research.

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