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Dentatorubral-pallidoluysian atrophy: a case report and review of literature

Xin Chen[1], Wenwen Xiang[1], Lijun Xu[1], Jiahao Zhao[1], Ye Yu[1], Qing Ke[1,2], Zhipeng Liu[1] and Li Gan[1*]

Abstract

Background Dentatorubral-pallidoluysian atrophy is a rare autosomal dominant neurodegenerative disease. It is a rare disease in the world. Therefore, sharing clinical encounters of this case can deepen global awareness and understanding of the disease.

Case presentation The patient was a 34-year-old male of Han nationality who was unmarried. The patient was admitted owing to weakness of the left lower limb with walking instability for 2 months and aggravation for 1 month. There was no dizziness, headache, numbness of limbs, convulsions, nausea, vomiting, abdominal pain, ataxia, nausea, vomiting, or abdominal pain. No nausea, vomiting, diarrhea, abdominal distension, tinnitus, hearing loss, fever, cough, expectoration. Personal history: worked in Cambodia 5 years ago, worked in Dubai 3 years ago, engaged in computer work, smoking or drinking habits. The patient was unmarried. Family history: the mother had symptoms similar to walking unsteadily (undiagnosed). Positive signs include a wide-base gait with a rotatory nystagmus that jumps upward in both eyes. Bilateral finger-nose instability test was quasi-positive, rapid alternating test was negative, and eye closure tolerance test was positive. Tendon reflexes were active in both upper limbs and hyperreflexia in both lower limbs. Stability of the heel, knee, and tibia. Genetic testing showed that the number of repeats in the dentatorubral-pallidoluysian atrophy ATN1 gene was 18 and 62, and the (CAG)n repeat sequence in the ATN1 gene was abnormal, with a repeat number of 62, and the patient was a pathogenic variant. The patient was diagnosed with dentatorubral-pallidoluysian atrophy. Dentatorubral-pallidoluysian atrophy remains a progressive neurodegenerative disease with no effective treatment. At present, the proband is taking 5 mg of buspirone three times a day, which has been reported to improve the symptoms. The patient was followed up for 6 months after taking buspirone, and there was no significant improvement in the temporary symptoms. At present, there are few cases of dentatorubral-pallidoluysian atrophy, and the characteristics of nystagmus in this disease have not been proposed in the past. This case reported the unusual presentation of nystagmus.

Conclusion Dentatorubral-pallidoluygur atrophy is a rare neurodegenerative disease with autosomal dominant inheritance. To the best of our knowledge, our present case report is the first case report of dentatorubral-pallidoluygur atrophy with specific nystagmus. We describe the special eye shake and its positive signs to increase dentatorubral-pallidoluysian atrophy clinical positive signs.

Keywords DRPLA, Clinical manifestation characteristics, DRPLA gene, Nystagmus characteristics

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Introduction

Dentatorubral-pallidoluysian atrophy (DRPLA) is a rare autosomal dominantly inherited degenerative disorder of the nervous system in which cerebellar ataxia and epilepsy as well as dementia are more common. Spinocerebellar ataxia (SCA) is a group of inherited

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neurodegenerative disorders that are highly heterogeneous clinically and genetically; DRPLA is a type of SCA, and DRPLA is a type of polyglutamine-polyQ disorder. The DRPLA gene ATN1 is located on chromosome 12p13.31, and in the ATN1 gene, the abnormal amplification of the CAG repeat sequence leads to the development of DRPLA. Early genetic presentation occurs in DRPLA, and it has been reported in literature that DRPLA is more prone to paternal transmission [1]. This disease has been reported most frequently in Japan, followed by Portugal, Spain, South Korea, and Venezuela, and is quite rare in China; a literature search revealed more than 30 cases both at home and abroad [2]. The clinical features, imaging, ophthalmoplegia characteristics, and gene mutation characteristics of a patient with adult-onset DRPLA, in which ataxia was the first symptom, are reported and analyzed herein. DRPLA is an extremely rare neurological disease. As clinicians, we cannot miss any opportunity to learn. Therefore, we report a patient with DRPLA to deepen the diagnosis and understanding of this disease, and we are the first to report the characteristics of nystagmus in DRPLA.

Case presentation

The patient was a 34-year-old Han Chinese man who was unmarried. The patient was admitted owing to weakness of the left lower limb with walking instability for 2 months and aggravation for 1 month (first visit: 1 September 2023; no follow-up. The patient presented with left lower extremity weakness without any obvious triggers, occasional choking or coughing while drinking water, unsteady walking with involuntary rightward turning of the head, and aggravated symptoms of unsteady walking in September, with no dizziness, headache, numbness of the limbs, convulsion, nausea, vomiting, abdominal pain, or ataxia. There was no nausea, diarrhea, abdominal distension, tinnitus, hearing loss, fever, cough, or sputum. Personal history: history of work in Cambodia 5 years ago, history of work in Dubai 3 years ago, computer worker, smoking or alcohol habit, and unmarried; family history: mother has similar symptoms of unsteady walking (undiagnosed). There is no particular history of medical and social psychological diagnoses, and no relevant diagnosis and treatment measures have been given before.

Physical examination on admission: temperature: 36.5 ℃, pulse: 68 beats/minute, respiration: 18 breaths/minute, blood pressure: 124/85 mmHg. Cardiopulmonary physical examination revealed no obvious abnormalities in the abdomen. Physical examination of the nervous system showed clear consciousness, wide-base gait, no aphasia, normal comprehension, insight, memory, calculation, and orientation. The double palpebral fissures were large, without drooping eyelids. No visual defects were noted with hand coarse testing. Eye movements were full without gaze palsy, and nystagmus was observed with eyes jumping in rotation. The pupils were equal in size and round, 2.5 mm in diameter, sensitive to direct and indirect light reflex bilaterally, and accommodative reflex was present. Double flank pain touch was symmetric and normal, and the jaw reflex was not elicited. Bilateral frontal lines and nasolabial grooves were symmetric and deep, with no deviation at the tooth angle. Binaural hearing was coarse but normal. Sound was clear, the soft palate elevated bilaterally, the uvula was midline, and the gag reflex was symmetric. Bilateral head turning and shrugging were symmetrical and strong, with no atrophy of the sternocleidomastoid and trapezius muscles. The tongue was midline, with no muscle atrophy or beam fibrillation. Limb joint position sense, motion sense, and tuning fork vibration sense were normal. Grade 5 moderate limb muscle strength with muscle tension was observed. Muscle atrophy and hypertrophy were not seen. Bilateral finger-nose tests were unstable, rapid rotational movement was negative, and Romberg sign was positive. Tendon reflexes were active in both upper limbs and hyperreflexia in both lower limbs. Hoffmann sign, Rosolimo sign, and palmochin reflex were negative bilaterally. Babinski sign was negative bilaterally, and the neck was soft. Kirschner's sign and Buchner's sign were negative.

Routine blood sampling after admission was unremarkable.

Routine cerebrospinal fluid examination, biochemistry, bacterial smear examination, virus II, bacterial culture, and cerebrospinal fluid immunoglobulin G were not abnormal; anti-neuronal cell profile 16 tests were negative (cerebrospinal fluid, serum). Thiobarbituric acid tissue based assay (TBA) test (cerebrospinal fluid): no positive signals were detected; TBA test (serum): weakly positive signals were detected in the hippocampus area, coloring in neuronal cells, and the cerebellar area showed an abnormal signal, coloring within the Purkinje neuronal cells.

The 3-hour video electroencephalogram (EEG) monitoring suggested the following: abnormal EEG, abnormal epileptiform discharges, and diffuse.

Magnetic resonance suggested mild bilateral cerebellar atrophy with multiple abnormal signals in the brainstem and mild atrophy of the cervical spinal cord with multiple abnormal signals in the cerebral white matter (Fig. 1).

Electroconvulsive nystagmography revealed bilateral upward jumping with twisting nystagmus from the upper pole of the eyeball to the left ear (Fig. 2) (of nystagmus electricity figure video visible supplementary material video).

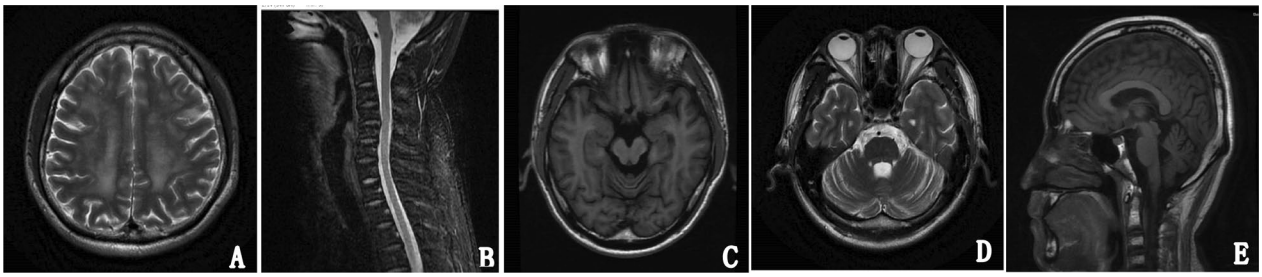

Fig. 1 Magnetic resonance imaging of the head and cervical spine of the proband with dentatorubral-pallidoluysian atrophy (4 September 2023). **A** White matter lesions in bilateral cerebral hemispheres; **B** mild atrophy of the cervical spinal cord; **C** multiple contralateral abnormal signals in the brainstem; **D** abnormal "cross" signals in the dorsal pons; **E** widening of partial sulci in the cerebellar hemispheres and mild cerebellar atrophy

The genetic disease candidate gene panel + dynamic mutation gene test results were as follows: 18 and 62 DRPLA ATN1 gene repeats and abnormal amplification of the (CAG)n repeat sequence in the ATN1 gene with 62 repeats. The patient had a disease-causing mutation (Fig. 3). The patient was diagnosed with DRPLA.

Therapy: idebenone at 30 mg three times a day and mecobalamin at 0.5 mg three times a day were given to the patient from the first day of hospitalization. The patient was hospitalized for a total of 9 days, during which oral medication was administered daily as prescribed. After discharge, the patient continued to take both drugs orally, and the dosage was consistent with that during hospitalization. The patient was followed up for 10 months after discharge. During our telephone follow-up, the patient reported that his walking symptoms had improved.

Discussion

DRPLA is an extremely rare neurological disease. Compared with previous literature, we are unique in that we report the nystagmus features of DRPLA, which have not been reported in previous literature. The patient developed symptoms of ataxia at the age of 34 years with a specific nystagmus: bilateral saccadic nystagmus with the upper pole of the eyeball twisted toward the left ear. In addition, oral idebenone (30 mg three times a day) and mecobalamin (0.5 mg three times a day) for 10 months helped the patient to walk unsteadily. This has not been mentioned in previous literature.

DRPLA is a subtype of spinal cerebellar ataxia (SCA) that is similar to other polyglutamine disorders and is characterized by similar gene dynamics [3]. The causative gene of DRPLA is located in the 12p13.31 region, and the elongation of the polyglutamine chain (PloyQ) is associated with an abnormal amplification of the repetitive sequence of CAG. In PloyQ, there is a specific protein, the atrophin-1 protein, and when this protein accumulates within the neuron, causing cytotoxicity, the neuron then degenerates and dies [4, 5]. DRPLA has been associated with the disruption of protein–protein interactions, in which amplified polyQ bundles play a crucial role, and dysregulation of gene expression [6]. The main clinically characterized symptoms of DRPLA are ataxia and cognitive decline. A summary of Chinese cases revealed that DRPLA disease is extremely rare in the Chinese population [7], and the incidence and age of onset of the disease do not differ significantly by sex, but the clinical manifestations are characterized by different ages of onset of the disease [8]. In China, the typical clinical features of adult-type cases of DRPLA are ataxia, cognitive decline, and involuntary movements, whereas epilepsy and myoclonic seizures are more common in juvenile-type clinical cases. In this case, the patient was 34 years old, an adult, with ataxia and unsteady walking as the first symptom, accompanied by rapid involuntary head rotation to the right, and upward jumping rotational nystagmus in both eyes was observed on examination. On admission, the patient's imaging suggested cerebellar atrophy and multiple abnormal signals in the brainstem; genetic testing confirmed that the number of repeats of the CAG sequence of the ATN1 gene in the preexisting patient was 18/62, which was consistent with the diagnosis of DRPLA.

A review of this case revealed that the clinical presentation characteristics of patients with adult-type DRPLA lacked specificity, and genetic testing was the basis for confirming the diagnosis, suggesting that although DRPLA is relatively rare in China, the detection of the number of CAG repeats in the ATN1 gene should not be ignored in addition to focusing on the characteristics of cerebellar atrophy in patients who are considered for investigating the etiology of ataxia. It is worth noting that previous studies did not mention nystagmus signs in patients with DRPLA, but the examination of the present patient revealed bilateral upward rotational nystagmus, suggesting that binocular upward rotational nystagmus may be a sign characteristic of DRPLA; therefore, the

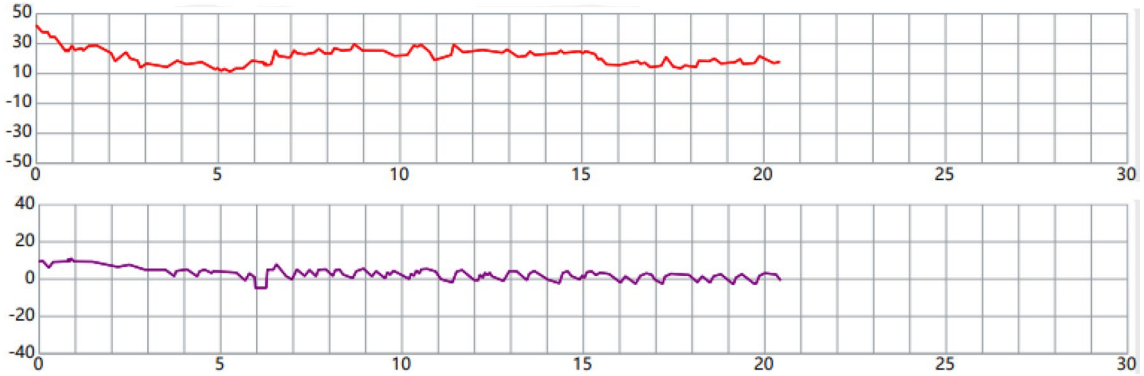

Fig. 2 Abnormal results of electronystagmography in the proband

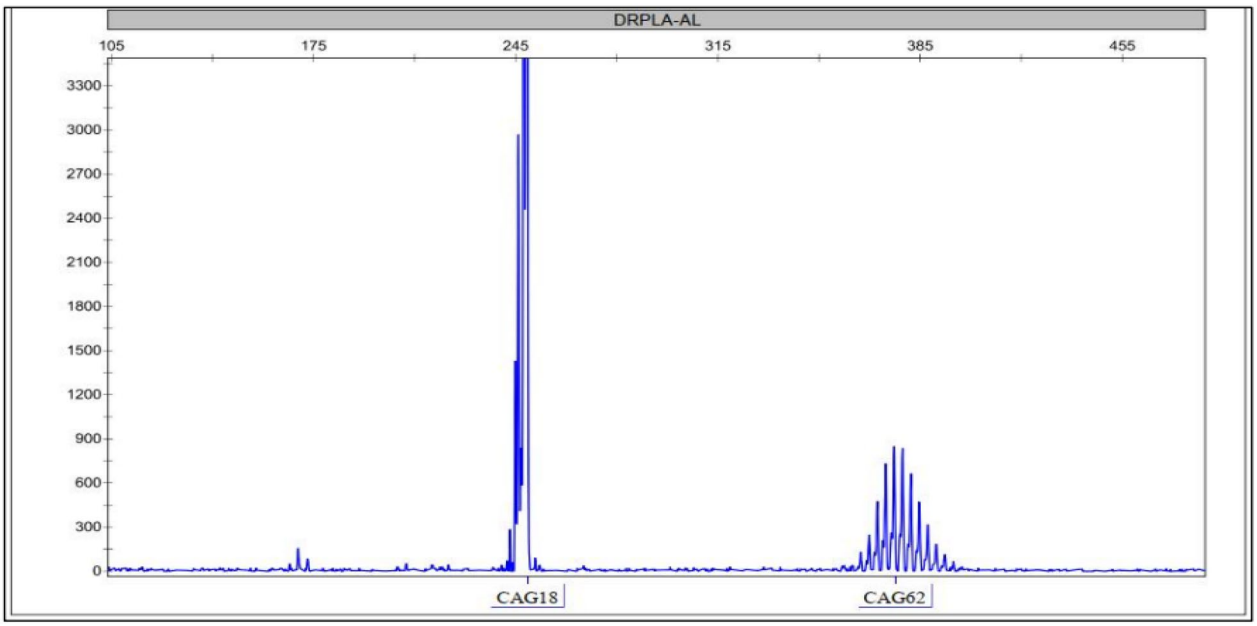

Fig. 3 Genetic disease candidate gene panel + dynamic mutation gene detection results for the proband

nystagmus signs of patients should also be considered when diagnosing patients with DRPLA.

DRPLA is a rare genetic neurodegenerative disease, its clinical features are extremely complex, and some patients' clinical symptoms lack specificity, which makes it easy to miss and misdiagnose. To further understand this disease and improve its diagnosis and treatment, we reviewed the relevant literature both at home and abroad and summarized the basic status of this disease.

(1) Clinical manifestations: DRPLA can occur at all ages, with 31 years as the average age and no significant difference in sex [9]. On the basis of the age of onset and clinical features, DRPLA is categorized into juvenile (< 20 years old), early adult (20–40 years old), and late adult (> 40 years old) subtypes, and the main manifestations of DRPLA in each age group are cerebellar ataxia and dementia [1, 10]. The age of onset is 15–19 years, and juvenile patients with DRPLA usually present with epilepsy, myoclonus, and mental retardation, with epilepsy as the first symptom and rapid progression of the disease. Adult-onset DRPLA has an age of onset of approximately 38–43 years, with cerebellar ataxia, dementia, involuntary movements, and psychiatric abnormalities as common clinical manifestations and sometimes head tremors and vision problems [6]. Ataxia and cognitive decline are usually the

first symptoms and need to be differentiated from other subtypes of SCA, as well as Huntington's disease and spinal medullary myasthenia gravis [11, 12]. Epilepsy is less common in patients with adult-onset DRPLA, but a few cases have been reported [13] (Table 1);

(2) Imaging: cranial magnetic resonance imaging (MRI) is an important test for diagnosing DRPLA, which commonly shows progressive atrophy of the brainstem and cerebellum and extensive cerebral white matter lesions on T2-weighted (T2W) or fluid–attenuated inversion recovery (FLAIR) sequences. Patient age and the number of CAG repeats are two independent factors affecting the severity of brainstem and cerebellar atrophy, and changes in the volume of the brainstem and cerebellum may be important indicators of disease progression. Cerebral white matter lesions, which are significant MRI features of DRPLA, are widely distributed in the cerebrum, brainstem, thalamus, and cerebellum in patients with DRPLA, and among them, cerebellar white matter lesions are a prominent feature of MRI in patients with DRPLA, especially at disease onset. Cerebellar white matter lesions are one of the prominent MRI features of DRPLA, especially in older patients [14], and have been reported in both juvenile and adult patients, with the adult type being the most common. The mechanism by which cerebellar white matter lesions occur in DRPLA has not yet been clarified, and several studies have shown that cerebellar white matter lesions in DRPLA are not related to ischemia-induced hypoperfusion; rather, they may originate from the disease process of DRPLA itself, which involves the accumulation of aberrant proteins resulting in the absence of axons or myelinated fibers. However, it is worth noting that the severity of cerebral white matter lesions did not significantly correlate with the duration of the disease or the number of CAG repeats in patients but was positively correlated with the age of the patients examined, suggesting that cerebral white matter lesions may not only be related to the disease itself but also be affected by other unknown factors, such as failure of the relevant compensatory mechanisms owing to aging, which needs to be further investigated [14–16].

(3) Gene mutation: the ATN1 gene is located on chromosome 12p13.31 and encodes the atroph-1 protein, a transcriptional corepressor widely expressed in the central nervous system. There is an unstable CAG repeat queue in this gene, encoding polyglutamine, and DRPLA is the result of abnormal amplification of the CAG repeat queue. Currently, it is believed that the number of CAG repeats in normal individuals is usually 6–35, and individuals carrying 35–47 CAG repeats show incomplete outgrowth and usually have mild clinical manifestations, while those with more than 48 CAG repeats have a complete outgrowth phenotype. The number of CAG repeats is negatively correlated with age of onset and positively correlated with disease severity. The average number of CAG repeats in juvenile patients with DRPLA is 68 (range: 63–79), that in early adult patients with DRPLA is 64 (range: 63–69), and that in late adult patients DRPLA is 63 (range: 48–67) [4, 9, 17]. DRPLA is similar to other PolyQ disorders in that the phenomenon of early onset of genetic predisposition occurs in PolyQ disorders, with the age of onset advancing from generation to generation and with symptoms appearing earlier and earlier in the same lineage. The age of onset is advanced, and symptoms worsen from generation to generation, which may be related to the erratic amplification of the CAG repeat cohort, which is more pronounced in patrilineal transmission [1, 9, 18], but follow-up of the present prediagnostic patient did not have similar clinical manifestations among the families of the prediagnostic patient (follow-up of the prediagnostic patient's parents, grandparents, and maternal grandparents).

(4) Diagnosis and treatment: the clinical diagnosis of DRPLA mainly relies on clinical manifestations, imaging examinations, family history, and ethnic history. There are no standardized criteria. DRPLA should be considered a possible cause of disease when the patient's relevant history and examinations meet the following criteria:

(1) Clinical manifestations vary according to the age of onset, with patients under 20 years of age mainly presenting with epilepsy, myoclonus, and ataxia, while patients over 20 years of age mainly present with ataxia, athetosis, involuntary movements, cognitive decline, mental behavioral abnormalities, and so on;
(2) Imaging shows atrophy of the brainstem and cerebellum and widely distributed cerebral white matter lesions in the cerebrum, cerebellum, brainstem, thalamus, and and other parts of the brain;

3) There is a family history of the disease, which is consistent with the characteristics of autosomal dominant inheritance, but there are also some DRPLA cases without a family history, and the absence of a family history does not exclude the diagnosis of DRPLA. Clinical patients suspected

Table 1 Summary of literature on the clinical features of DRPLA

Reporting scholar	Onset age	Sex	Clinical features	Genetic characterization	Family
Yao *et al.*	37 years old	Male	The patient presented with vague speech and gradually aggravated symptoms, accompanied by dysarthria, anomic aphasia, cognitive decline, and no nystagmus	The proband's blood DNA test showed that the ATN gene CAG repeat sequence was 65 times, and the other family members refused genetic testing	The proband's father, who was 39 years old, started with walking instability and gradually aggravated, followed by choking and dysphagia, while the other family members were normal
Hao Ying *et al.*	38 years old	Female	The patient began with unsteady cycling, which was gradually aggravated, accompanied by unsteady walking, occasional involuntary limb movements in dreams, dysarthria, and split smooth eye tracking	The proband's blood DNA gene detection CAG repeat 14 and 54 times, the other family members did not undergo gene detection	One of the proband's sisters had epilepsy at the age of 13 years. The sons of the proband's unaffected sister had seizures at 4 and 6 years of age
Review by Tong Suijun *et al.*	Not described	Not described	The most common initial symptoms were ataxia, seizures, and involuntary movements	The CAG repeat number is correlated with the severity of DRPLA	Not described
Fan Rumeng *et al.*	18 years old	Female	The patient presented with head tilt at onset, occasional involuntary jitter, progressive walking instability, vague speech, difficultly standing with eyes closed, and positive reinforcement test, unstable finger-nose test, and no nystagmus	CAG repeat in ATN-1 gene: The proband had 15/59 episodes. The proband's sister had 17/59 episodes. The proband's mother was 19/56 times. The proband's father was 15/17 times. The genetic results of the members of the Yu family were not described	The younger sister of the proband, who had head tilt and intermittent involuntary shaking, was not pregnant. The mother of the proband presented with walking instability. The proband's grandfather started with walking instability. The proband's uncle started with walking instability. The proband's aunt started with walking instability. The second uncle of the proband presented with involuntary shaking of the upper limbs and head, and the age of onset was not described. The proband's cousin started with walking instability
Cai Xiaofang *et al.*	43 years old	Male	The onset of the disease was unconsciousness and limbs began straightening. There were five episodes in 6 years. He was accompanied by involuntary shaking of both hands, memory loss, and inability to move quickly in all directions of both eyes	Not described	The father of the proband died of similar symptoms, and the two younger sisters were healthy
Du Wei *et al.*	48 years old	Female	The patient presented with unsteady gait, involuntary jitter of both upper limbs, blurred speech, and no nystagmus	The CAG repeat number of DRPLA-related genes was 59	The members of the Yu family are not special

Table 1 (continued)

Reporting scholar	Onset age	Sex	Clinical features	Genetic characterization	Family
Jiang Hong *et al*.	45 years old	Female	The onset of the disease was walking instability, gradually aggravated, accompanied by slurrilia, memory and calculation ability decline, and then paroxysmal loss of consciousness and falls, lasting 1–2 seconds each time. Nystagmus was induced by horizontal gaze	The proband's blood DNA test showed that the ATN gene CAG repeat number was 14/54, and the rest of the family members were not described	The proband's aunt was 50 years old and could not walk. The proband's sister was 52 years old and could not walk independently. The elder brother of the proband was 52 years old and could walk independently with an intoxicated gait
Takeshi、Osamu *et al*.	Not described	Not described	The clinical features and onset age of DRPLA are correlated with the degree of CAG repeat expansion	The cDNA of DRPLA gene provides a new idea for the further establishment of animal models	Not described
CAI Pingping *et al*. used PCR amplification to report a family without epilepsy phenotype	Not described	Not described	The proband was a middle-aged male with the onset of ataxia as the main symptom in adulthood. The characteristics of nystagmus were not described	The probands of the two families had CAG repeat expansion	Not described
Wu Yuting *et al*.	14 years old	Male	The patient presented with paroxysmal unconsciousness and limb convulsions, walking instability, head and body deflection to the right, vague speech, irritable personality, and no nystagmus	Blood genetic testing showed that ATN gene CAG repeat number was 15/63	The proband's father and uncle began to have walking instability after the age of 40

of having DRPLA should improve genetic testing, and DRPLA confirmation criteria for genetic testing for abnormal amplification of the ATN1 gene CAG repeat sequence, which is generally greater than 48 [7, 9], should be met. DRPLA is still a progressive neurodegenerative disease, and there is no effective treatment [9]. Moreover, the progression of the disease cannot be stopped, and symptomatic treatment is still needed. This patient received 30 mg of idebenone orally three times a day during hospitalization. Mecobalamin was administered orally at 0.5 mg three times a day. The patient was hospitalized for a total of 9 days and received both drugs daily according to the prescribed dose. After discharge, the patient continued to take both drugs orally and regularly, with doses consistent with those during hospitalization. Patients were followed up for 10 months after discharge. During the follow-up, the patient reported that walking symptoms had improved.

Conclusion

In summary, we report a case of a patient with dentate nucleus red nucleus pallidus globus pallidus atrophicus who began with unsteady walking, with specific nystagmus double upturns accompanied by signs of nystagmus twisting of the upper pole of the eye toward the left ear. Through a literature review, we discussed in detail the clinical presentation and diagnostic criteria of patients with dentate nucleus red nucleus pallidus globus pallidus atrophicus and emphasized the patient's specific nystagmus, cranial magnetic resonance, family history, and genetic testing for definitive diagnosis and early detection to improve quality of life. Additionally, we learned that the age of onset of DRPLA disease in the same family line advances from generation to generation, and symptoms worsen from generation to generation and are more pronounced in paternal transmission. However, following the present case of the preexisting patient, among the family, his mother had similar symptoms of unsteady walking, but owing to the complexity of the family environment of the preexisting patient, it was not possible to perform a genetic test on his mother.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s13256-024-04745-3.

Supplementary material 1.

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Author contributions
XC reviewed the literature and wrote the initial manuscript drafts; WX, LX, JZ, YY, QK, ZL, and LG managed the patient, reviewed the literature, and completed the manuscript. Both authors read and approved the final manuscript.

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Availability of data and materials
The datasets during the current study available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate
Given the patient's extended residence in another province and the inconvenience of walking, telephone contact was maintained with the patient and his father, and the patient agreed to allow us to publish his case.

Consent for publication
Written informed consent was obtained from the patient for publication of this report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests
The authors declare that they have no competing interests.

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