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A family case of hereditary olivopontocerebellar atrophy: features of diagnosis and course of the disease

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Abstract: *OPCA is a heterogeneous group of degenerative ataxias, the common feature of which is the occurrence of changes in the lower olives of the medulla oblongata, nuclei and transverse fibers of the pons, and cerebellar cortex. OPCA is not explicitly considered in general epidemiological surveys on spinocerebellar syndromes. It was described that in Cantabria (Spain) the prevalence ratios of autosomal-dominant cerebellar ataxia (ADCA) and idiopathic late-onset cerebellar ataxia (ILOCA) were 1.2 and 2.2 cases per 100,000, respectively. Some 60% of patients included in these groups had a «cerebellar-plus» syndrome and their computed tomographic (CT) or magnetic resonance imaging (MRI) scans revealed cerebellar and brainstem atrophy, allowing a presumptive diagnosis of OPCA. According to these estimations, the prevalence ratio of OPCA is about 2 per 100,000 (Berciano, 1991). The clinical picture is characterized by significant inter- and intra-familial polymorphism. Symptoms of the disease start to appear, usually at the age of 30-40, in the form of a disorder of coordination and unsteadiness when walking quickly (later, with the progression of the disease, a typical ataxic gait starts to develop). At the same time, intentional tremor and dyscoordination of hands appear, and in some cases – an asynergy of facial muscles. Speech disorders manifest themselves quite early and have a severe cerebellar-dysarthric character. An important place in the diagnosis of OPCA belongs to neuroimaging methods – CT, and MRI (the presence of an atrophic process and the absence of focal changes in the brain parenchyma). One of the factors that confirms the diagnosis is the presence of a family history and the relentlessly progressive nature of the disease.*

Keywords: Ataxia, Brainstem, Genetic Anticipation, Cerebellum, Olivopontocerebellar Atrophy.

Introduction

Under the term of olivopontocerebellar atrophy different nosological pictures are grouped, all characterized by showing clinical signs of deficiency of the structures of the pons and of the cerebellum (Giuliani G et al., 1992). Progressive ataxias are a group of many uncommon yet often very disabling diseases, which can be inherited or acquired. The average prevalence of recessive hereditary ataxias is 3.3/105 and of dominant hereditary ataxias is $2.7/10^5$ (Mascalchi, 2022). Spinocerebellar ataxias are inherited or sporadic diseases characterized by progressive dysfunction and loss of neuronal systems in the spinal cord, brainstem, and cerebellum (Mascalchi, 2022). Some scientific works mentioned that olivopontocerebellar atrophy was widely accepted as part of the neuropathological spectrum of multiple system atrophy (MSA) (Wenning et al., 1996). Approximately one-fourth of sporadic olivopontocerebellar atro-

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phy patients will evolve to multiple system atrophy within 5 years, and this transition carries a poor prognosis for survival. Older age at onset of ataxia and earlier presentation in a neurologic specialty clinic predicted transition to MSA (Gilman et al., 2000). Olivopontocerebellar atrophy is a rare neurodegenerative syndrome associated with 2 distinct disorders: multiple system atrophy and spinocerebellar ataxia (Harfourch et al., 2020).

Dejerine and Thomas introduced the term olivopontocerebellar atrophy (OPCA) to designate the pathological framework in a sporadic case with idiopathic late-onset progressive cerebellar ataxia in 1900. Nine years before, however, Menzel had reported a family with a complex clinical picture characterized by progressive ataxia, spasmodic dysphonia, rigidity in the lower limbs, dysphagia, and dystonic posture of the neck. The onset of symptoms was at about 30 years of age. There were four affected members over two generations. An autopsy revealed olivopontocerebellar lesions, together with degeneration of posterior and Clarke's columns, pyramidal and spinocerebellar tracts, and substantia nigra. Menzel found "very flattened and reduced subthalamic nuclei," but unfortunately, he did not give any microscopic description of these structures; demonstration of luysian atrophy would have been of great interest in view of the dystonic postures of the patient. The early reports of Dejerine and Thomas and, later, Loew's thesis, developed under the tutelage of Dejerine himself, considered OPCA to be atypical when there was a hereditary factor (as is the case for the aforementioned family reported by Menzel), lesions extending beyond the olivopontocerebellar framework, or a clinical presentation and reviewed by Berciano (Doctoral thesis, University of the Basque Country, Spain, 1978). However, the concept of atypical OPCA fell into disuse with the recognition of familial OPCA (FOPCA) and the many lesions that frequently accompany olivopontine degeneration (Berciano, 1982).

Olivopontocerebellar atrophy is a pathological label implying not only olivopontocerebellar changes but also cases with more widespread lesions involving the CNS (Berciano et al., 2006).

Harding proposed the clinicogenetic classification which was soon universally accepted and has been modified later (Table 1) (Berciano, 1982).

Table 1. Harding's Clinicogenetic Classification of the Hereditary Ataxias and Paraplegias. Adapted, with permission, from Harding, A. E. (1983). Classification of the hereditary ataxias and paraplegias

I. Congenital disorders of unknown etiology II. Ataxic disorders with known metabolic of other cause III. Ataxic disorders of unknown etiology	A. Early onset cerebellar ataxia (EOCA) (onset usually before 20 years)	I. Friedreich's ataxia II. Early onset cerebellar ataxia with retained tendon reflexes III. With hypogonadism ± deafness and/or dementia IV. With myoclonus (Ramsay Hunt syndrome, Baltic myoclonus) V. With pigmentary retinal degeneration ± mental retardation and/or deafness VI. With optic atrophy ± mental retardation VII. With cataracts and mental retardation (Marinesco–Sjögren syndrome) VIII. With childhood-onset deafness and mental retardation IV. With congenital deafness X. With extrapyramidal features XI. X-linked recessive spinocerebellar ataxia
	B. Late-onset cerebellar ataxia (onset usually after 20 years)	I. Autosomal-dominant cerebellar ataxia (ADCA) with optic atrophy/ophthalmoplegia/dementia/extrapyramidal features/amyotrophy (probably includes Azorean ataxia) (ADCA type I) II. ADCA with pigmentary retinal degeneration ± ophthalmoplegia and/or extrapyramidal features (ADCA type II) III. "Pure" ADCA of later onset (over 50 years) (ADCA type III) IV. Periodic autosomal-dominant ataxia IV. Periodic autosomal-dominant ataxia

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Table 1. (*continued*)

		V. "Idiopathic" late-onset cerebellar ataxia (ILOCA) (with either "pure" or cerebellar-plus syndrome)
IV. Hereditary spastic paraplegia (HSP)	A. "Pure" HSP B. Complicated forms of HSP	

Aim

To confirm the increasing of intrafamilial polymorphism of the clinical picture of the hereditary form of olivopontocerebellar atrophy in each subsequent generation along with a decrease in the age of onset of the disease.

Materials

A patient E., who was born in 1991 has been examined in the neurological department of the State Institution «Head Medical Center of the Ministry of Internal Affairs of Ukraine». Since 2013 she has been under the dispensary supervision of a neurologist for olivopontocerebellar atrophy and undergoes preventive treatment courses every 6-12 months. Over the last 10 years, the disease has been progressing, and for the last five years, the patient requires care because of disability.

Family history: Symptoms of the disease were first detected in the family history of the patient's great-grandfather when he was 45 years old. The girl's grandfather had the first symptoms of the disease at the age of 41, and her mother at the age of 36 (Fig.1).

The great-grandfather and grandfather had coordination disorders in the form of dynamic (unsteadiness when walking – "drunken gait") and static form of cerebellar ataxia. The patient's mother, in addition to coordination disorders in the form of dynamic and static form of cerebellar ataxia (unsteadiness when walking – "drunken gait"), also had muscle atrophy, and most importantly – bulbar syndrome with significant swallowing disorders, which led to her death.

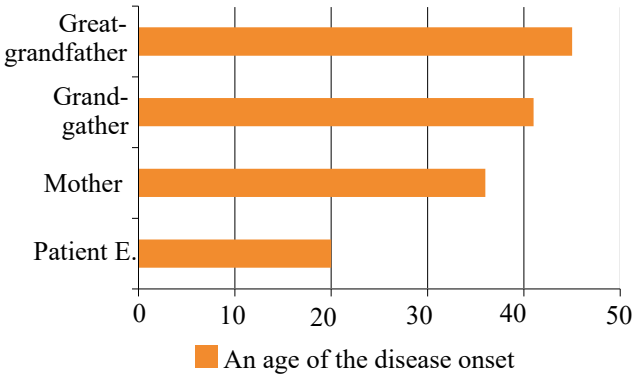

Figure 1. Ages of the disease onset in different members of one family

Patient E.'s medical history: The onset of the illness was at the age of 20 with severe gait deterioration. At the moment, she can walk only under additional help and support (crutches, which help less and less) for a small distance – a few meters. She has severe cognitive impairments, cerebellar ataxia, a significant decrease in the volume of movements in upper and lower limbs, and bulbar syndrome: pronounced dysphonia (change in the voice timbre), dysphagia (frequent regurgitation of liquid food), progressive dysarthria (speech disorder with affected articulation). Also, she suffers from urinary disorders.

The above-mentioned symptoms, which were observed in 4 representatives of the same family are evidence of not only the polymorphism of neurological manifestations but also its increase in each subsequent generation. The increasing severity of the disease and the appearance of its manifestations for the first time at a younger age with each subsequent generation are manifestations of anticipation that are observed in OPCA.

Neurological status of patient E.: the consciousness is clear, progressing in cognitive decline. Decreased convergence with insufficiency of the internal rectus muscle of the eyes more on the right, asymmetry of the eye slits (more severe on the right), asymmetry of the face due to the right corner of the mouth, dysphagia, dysphonia, dysarthria, slight deviation of the tongue to the left, pronounced amyotrophy and fasciculations of the tongue and scanned speech are observed. Positive grasping reflex on the right side. Deep tendon and periosteal reflexes in the hands D=S, and in the legs S>D. Abdominal reflexes are present. Pathological Strümpell and Babinsky reflexes are indicated

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on the right side. Positive Marinescu-Radovichi symptom is present on the left side. The volume of active movements of the limbs is reduced. Muscle strength in hands – 4 points, and in legs – 3 points. Atrophy of small interphalangeal muscles of the hands. Muscle tone is reduced. The patient performs coordination tests with a severe intentional tremor and misses. Adiadochokinesis is observed. Romberg's test shows severe imbalance – swaying. Hypoesthesia in the left limb (forearm). The gait is shaky ("drunk"). Makes several steps with additional help. Disorder of urination by the central type.

The patient underwent the following diagnostic procedures in the State Institution «Head Medical Center of the Ministry of Internal Affairs of Ukraine»:

1) Genealogical study: collected genealogical history;

2) MRI of the brain. Conclusion: MRI signs of degenerative changes in both cerebellar hemispheres (cortical furrows of both cerebellar hemispheres are unevenly expanded);

3) Examination by an ophthalmologist: moderate myopia in both eyes. Peripheral retinal dystrophy of both eyes. Dispensary supervision by an ophthalmologist is recommended;

4) Echocardiography: aortic valve leaflets are compacted. Cavities are not enlarged. The kinesis of the walls is satisfactory, ejection fraction – 69%.

After the neurological examination and getting results of additional diagnostic procedures, a diagnosis was formulated: Hereditary olivopontocerebellar atrophy with severe stato-locomotor disorders, bulbar syndrome, tetraparesis, which prevails in the lower limbs, dysfunction of the pelvic organs by the central type. Highly progressive course of the disease.

Treatment that was used for preventive purposes every 6 months: L-lysine aescinat 10.0 №10, Phenibut 250 mg – 2 twice per day, group B vitamins 2.0 i/m №5 every other day, Nucleo C.M.P. 1.0 i/m #3, then 1 year – 2 years per day for 9 days, physical therapy.

Results

In the example of a family case of hereditary OPCA, the polymorphism of manifestations of the disease within 4 generations of one family with manifestations of anticipation was demonstrated.

The clinical presentation of OPCA usually begins with cerebellar ataxia, especially involving gait. When dementia or extrapyramidal rigidity is among the first manifestations, they remain dominant symptoms throughout the disease. In OPCA associated with SCA7, the visual defect is usually an early symptom, which may precede gait ataxia. Spasticity or psychomotor retardation is the most common presenting semeiology of OPCA associated with congenital ataxia (Berciano, 2007).

Neuropsychological analyses revealed executive dysfunction as a common sign in SCA1. Additionally, mild deficits of verbal memory were found in SCA1, SCA2, and SCA3.94 In SCA2, dementia has been found in four of 17 patients (24%) according to Mini-Mental State Examination. Dementia is a common and prominent symptom in SCA17. Cognitive deficits of variable degrees are also reported in patients with SCA12, SCA13, SCA19, and SCA21, as well as in patients with mutations in FGF14. A study of psychopathological abnormalities in patients with various types of cerebellar degeneration found a high prevalence of depressive episodes (67%) and personality change (26%) as well as dementia (10%) and mild cognitive impairment (10%) (Schöls, et al., 2004).

Our patient has manifestations of ataxia, as members of all previous generations of her family with OPCA, but there are also manifestations of cognitive decline, which were observed in both her grandfather and mother but to a lesser degree.

Magnetic resonance imaging is a safe technique and constitutes a fundamental tool for both differential diagnosis of the causes of acute and subacute ataxia and the characterization of patients with progressive chronic ataxia. Main MRI characteristics of OPCA include atrophy of the cerebellum, brainstem, and cervical spinal cord and characteristic diffuse signal changes of the pons, middle cerebellar peduncle, and cerebellum in proton density and T2-weighted images (Mascalchi, 2008). Atrophy of the brainstem, more pronounced in the inferior portion of the basis pontis, of the vermis, of the middle cerebellar peduncles, and of the cerebellar hemispheres characterizes olivopontocerebellar atrophy. On the one hand, the typically flattened shape of the basis pontis in sagittal images and the “pointed” shape in the coronal images of the middle cerebellar peduncles, are observed in

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OPCA. On the other hand, the "dragonfly" or "butterfly" appearance of the cerebellum on coronal MR images that are peculiar to pontocerebellar hypoplasia, with the flattened cerebellar hemispheres representing "the wings", is associated with a less pronounced (dragonfly) or proportional (butterfly) vermis size decrease (Mascalchi, 2022). In our patients, we observed changes characterizing OPCA on brain MRI.

In some patients, pyramidal signs (enhanced stretch reflexes, extensor-plantar responses, or both; rarely was spastic paraplegia or pseudobulbar dysarthria) could be present and tendon reflexes can become hypoactive or hyperactive. Pyramidal signs may be explained by a disease-specific pattern of neuronal loss in the spinal ventral horn involving small neurons of the dorsomedial zone (Terao et al., 1994). Pathological foot reflexes of the extensors group were positive in our patient.

Atrophic changes in the intercarpal spaces of the bones of both hands were observed in our patient. Some reports inform that around a quarter of SCA2 patients show fasciculations or amyotrophy or both (Infante et al., 2005). Loss of large myelinated fibers and secondary demyelination to axonal loss have been found on sural nerve biopsies. These features, together with high proportion white column demyelination and anterior gray horn atrophy, suggest that peripheral neuropathy in OPCA is due to degeneration of posterior root ganglion and anterior horn cells in the spinal cord, that is, a type of spinal sensory and motor neuronopathy (Berciano, 2007).

Bladder dysfunction first can be observed as urinary urgency and afterward combined with incontinence. Double incontinence rarely occurs in patients, but urinary retention is uncommon. Erectile dysfunction was not recorded in patients with OPCA SCA (Berciano, 2007). Our patient had periodic urinary incontinence.

Dysphagia is a relatively common manifestation of OPCA and OPCA associated with SCA and swallowing disorders are characteristic of intermediate and advanced disease. Dysphagia has been related to dysfunction of the superior esophageal sphincter. Like the striated muscles of the urethral and the posterior cricoarytenoid muscle, the cricopharyngeus muscle is tonically and rhythmically active as a result of spontaneous discharge of reticular interneurons adjacent to the nucleus ambiguus. Conceivably the tonic firing of these motor neurons (Onuf's nucleus and nucleus ambiguus), which differs from other skeletal muscles, accounts for their joint tendency to degenerate in OPCA (Berciano, 2007). Our patient currently has severe bulbar syndrome with dysphagia and mainly dysphonia.

The main sleep disorders related to hereditary ataxias include rapid eye movement (REM) sleep behavior disorder, insomnia, excessive daytime sleepiness, obstructive and central sleep apnea, periodic leg movement in sleep, and restless legs syndrome (Huebra et al., 2019). Sleep disorders, including nocturnal stridor, characteristic of OPCA associated with Multiple system atrophy (MSA), have exceptionally been reported in familial OPCA (FOPCA). Restless legs syndrome and impaired sleep appear to be relatively common manifestations of SCA3 (Berciano, 2007). We did not observe sleep disorders in our patient, nor did her close relatives complain of dyssomnia.

Patient E. complains of a severe worsening in gait, coordination disorders, a decrease in the volume of movements in the upper extremities and atrophy in hands, a change in the timbre of the voice, disorder of swallowing, and periodic urinary incontinence. In addition to coordination disorders, the patient's mother also had muscle atrophy and bulbar syndrome. The great-grandfather and grandfather had unsteady gait and impaired coordination. After analyzing the neurological deficit in family members, it becomes clear that in each subsequent generation, symptoms of the disease first appear at a younger age. In subsequent generations, clinical symptoms appear due to the increasing severity of the disease. The same symptoms of polymorphism with increasing intensity of manifestations of all neurological deficits, as well as an earlier age of its occurrence in each subsequent generation, prove the presence of anticipation in patients with OPCA (Fig. 2, 3).

Diagnosis: The diagnosis of OPCA is based on the clinical picture that is characterized by progressive cerebellar-plus syndrome. The nature, sporadic or familial, and age at the onset of the disease allow the planning of genetic molecular studies to detect dynamic or point mutations associated with dominant ataxias and EOCA. Electrophysiological

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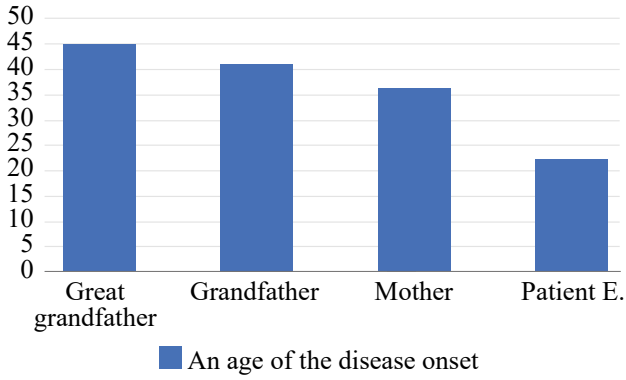

Figure 2. Ages of the disease onset in different members of one family

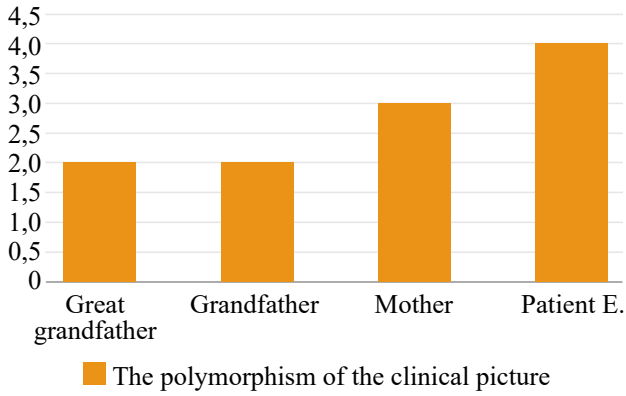

Figure 3. The polymorphism of the clinical picture of the disease in different members of one family

studies, including nerve conduction studies, multimodal evoked potentials, eye movement recordings, and central motor conduction time investigation determination using magnetic stimulation, are useful to assess the participation of the corresponding neural system in OPCA. Neuroimaging techniques are the gold standard proof for delineating brainstem and cerebellar atrophy, which is the hallmark of OPCA (Berciano, 2007).

To diagnose the disease in our patient we used: brain MRI, genealogical, ophthalmoscopic, as well as echocardiography studies. Based on the classifications mentioned above in this article, we concluded that our patient has OPCA familial (Menzel) type, autosomal-dominant cerebellar ataxia (ADCA) type I (classified by Harding's Clinicogenetic Classification of the Hereditary Ataxias and Paraplegias).

Treatment: Symptomatic treatment is only possible and depends on the clinical manifestations of the disease. Levodopa may provide some benefit for extrapyramidal rigidity or bradykinesia, treatment may be initiated with carbidopa/levodopa 25/100, 0.5–1.0 tablet twice a day, and increased every few days to efficacy or toxicity. Dopamine agonists in maximally tolerable doses may also be used if there is no response to levodopa. Urinary disturbances due to detrusor hyperreflexia are treated with a peripherally acting anticholinergic agent (oxybutynin (5–10 mg at bedtime) or propantheline). In any case, treatment of either urinary or sexual disturbances should be carried out in close collaboration with experts in neurology and urology. In fact, none of the trials has produced results convincing enough to justify the routine use of any drug in OPCA. Psychosocial, physical, occupational, and speech therapies may be helpful to reduce the patient's disability and to maintain independent functioning longer. Gait training and assistive devices to prevent falling will avoid further debilitation of the patient (Berciano, 2007).

Conclusions

Thus, we confirmed not only the demonstrative polymorphism of symptoms in the hereditary form of OPCA but also anticipation, which is one of the characteristic features of the disease.

We concluded that our patient has OPCA familial (Menzel) type, autosomal-dominant cerebellar ataxia (ADCA) type I (classified by Harding's Clinicogenetic Classification of the Hereditary Ataxias and Paraplegias).

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This study received no external funding.

Conflict of interest

The authors declare no conflict of interest.

Consent to publication

All authors have read the text of the manuscript and given their consent for its publication. Informed consent was obtained from the patient.

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Сімейний випадок спадкової оливопонтоцеребелярної атрофії: особливості діагностики та перебігу захворювання

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Анотація: *оливопонтоцеребелярна атрофія (ОПЦА) — це гетерогенна група дегенеративних атаксій, спільною ознакою яких є виникнення змін у нижніх оливах довгастого мозку, ядрах і поперечних волокнах моста та корі мозочка. ОПЦА не враховується в загальних епідеміологічних дослідженнях спиномозочкових синдромів. Було описано, що в Кантабрії (Іспанія) співвідношення поширеності аутосомно-домінантної мозочкової атаксії та ідіопатичної мозочкової атаксії*

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з пізнім початком становило 1,2 і 2,2 випадку на 100 000 відповідно. Близько 60% пацієнтів, включених у ці групи, мали синдром «мозочок-плюс», і комп'ютерна томографія (КТ) або магнітно-резонансна томографія (МРТ) виявила атрофію мозочка та стовбура мозку у цих пацієнтів, що дозволило припустити діагноз ОПЦА. Згідно з цими оцінками, коефіцієнт поширеності ОПЦА становить приблизно 2 на 100 000 (Berciano, 1991). Клінічна картина характеризується значним між- та внутрішньосімейним поліморфізмом. Симптоми захворювання починають проявлятися, як правило, у віці 30-40 років у вигляді порушення координації та хиткості при швидкій ходьбі (пізніше, з прогресуванням захворювання, починає розвиватися типова атактична хода). При цьому з'являються інтенційний тремор і дискоординація рук, а в окремих випадках - асинергія м'язів обличчя. Порушення мови проявляються досить рано і мають виражений мозочково-дизартричний характер. Важливе місце в діагностиці ОПЦА займають методи нейровізуалізації — КТ, МРТ головного мозку (наявність атрофічного процесу та відсутність вогнищевих змін паренхіми головного мозку). Одним з факторів, що підтверджує діагноз, є наявність сімейного анамнезу та невпинно прогресуючий характер захворювання. Таким чином, ми підтвердили не тільки демонстративний поліморфізм симптомів при спадковій формі ОПЦА, але й антиципацію, яка є однією з характерних ознак захворювання. Ми прийшли до висновку, що у нашої пацієнтки є сімейний тип ОПЦА (Мензеля), аутосомно-домінантна мозочкова атаксія (АДМА) типу I (класифікована за допомогою Клініко-генетичної класифікації спадкових атаксії та параплегії Гардінга).

Ключовіслова: атаксія, стовбур мозку, генетична антиципація, мозочок, оливопонтоцеребелярна атрофія.