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Late diagnosis of cerebral palsy in a 16-year-old girl – a case report

Późne rozpoznanie mózgowego porażenia dziecięcego u 16-letniej dziewczynki – opis przypadku

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Abstract

Cerebral palsy is the most common cause of motor disability in children. Cerebral palsy is a static encephalopathy with a variable clinical picture and multifactorial aetiology. Disorders arise from disturbances in the early development of the brain in the foetal, perinatal or postnatal period. The disease affects around 17 million people worldwide; its incidence is estimated to be 1.5–3 per 1,000 live births. A slight male predominance is observed. The disease has a multifactorial aetiology, with prematurity being the most important risk factor. There are four types of cerebral palsy: spastic (the most common – (70%), dystonic (10%), mixed (15%) and ataxic (5%). In addition to motor disability of varying severity, the majority of patients present with other accompanying deficits, such as mental retardation, epilepsy, dysphagia, impaired hearing and vision. The diagnosis of cerebral palsy should be based on detailed medical history, including pregnancy and childbirth as well as a regular assessment of the child's development from the first months of life. In the case of clinical doubts, the diagnosis is extended to include magnetic resonance imaging, electroencephalography, metabolic and genetic tests. Children with cerebral palsy require a comprehensive, multidisciplinary care, including physical therapy and rehabilitation. Bobath and Vojta concepts are the most common rehabilitation approaches. Early diagnosis and regular rehabilitation are crucial to ensure adequate quality of life for a child with cerebral palsy. The paper presents a case of a 16-year-old girl with a long history of pain in the lower limbs, spine and temporomandibular joints. The symptoms were accompanied by chest pain as well as numbness and weakness of the left upper limb.

Keywords: cerebral palsy, aetiology, disability, prematurity, spasticity

Streszczenie

W krajach wysokorozwiniętych mózgowe porażenie dziecięce stanowi najczęstszą przyczynę niepełnosprawności ruchowej występującej w dzieciństwie. Jest to stałe, choć zmieniające się w czasie zaburzenie ruchu i postawy, wynikające z trwałego i niepostępującego uszkodzenia mózgu w stadium jego niezakończonego rozwoju. Choroba dotyczy około 17 milionów ludzi na całym świecie, a zapadalność wynosi 1,5–3 na 1000 żywych urodzeń. Obserwuje się niewielką przewagę zachorowań wśród chłopców. Etiologia choroby jest zróżnicowana, jednak najważniejszy czynnik ryzyka stanowi wcześniactwo. Wyróżnia się cztery postaci mózgowego porażenia dziecięcego: spastyczną (70%), dystoniczną (10%), ataktyczną (5%) i mieszaną (15%). Towarzyszą mu również inne zaburzenia, w tym upośledzenie umysłowe, padaczka oraz zaburzenia wzroku, słuchu i połykania. Podstawą diagnozy są szczegółowy wywiad lekarski, uwzględniający przebieg ciąży i porodu, oraz regularna ocena rozwoju dziecka od pierwszych miesięcy życia. W przypadku wątpliwości klinicznych diagnostykę uzupełnia się o badanie rezonansu magnetycznego głowy, elektroencefalografię, testy metaboliczne i genetyczne. Dzieci z mózgowym porażeniem dziecięcym wymagają kompleksowej, wielodyscyplinarnej opieki. Najczęściej są rehabilitowane według metody Bobath lub Vojty. Wczesna i systematyczna rehabilitacja jest niezbędna do tego, by zmniejszyć trwałe następstwa choroby i poprawić jakość życia pacjenta. W pracy przedstawiono opis przypadku 16-letniej dziewczynki z obciążonym wywiadem okołoporodowym, konsultowanej przez licznych specjalistów z powodu długotrwałego bólu kończyn dolnych, kręgosłupa i stawów skroniowo-żuchwowych. Objawom towarzyszyły ból w klatce piersiowej oraz drętwienie i osłabienie lewej kończyny górnej.

Słowa kluczowe: mózgowe porażenie dziecięce, etiologia, niepełnosprawność, wcześniactwo, porażenie spastyczne

Cerebral palsy (CP) is the most common cause of motor disability in children in highly developed countries[1]. CP is a static encephalopathy exhibiting an evolving clinical picture over time, which is caused by permanent, non-progressive damage to the developing brain. The disease was first described in 1843 by William Little, a British orthopaedic surgeon, who noticed a relationship between palsy, limb deformation, spasticity and prematurity in a child[2].

It is estimated that CP affects around 17 million people worldwide; the incidence is 1.5–3 per 1,000 live births[3–7] and is gradually growing due to improved perinatal care and reduced neonatal mortality[8,9]. A minor male predominance is observed (1.4:1)[10].

The aetiology of CP is multifactorial (Tab. 1)[11–15]. Prematurity is the most important risk factor. CP is diagnosed in 14.6% of children born before 28 weeks gestation and 0.11% of children born at term. Administration of antenatal corticosteroids to women at risk of preterm birth is the only factor that has been shown to reduce the risk of CP[16]. The role of genetic factors in the development of CP is also emphasised[17].

CP primarily manifests with motor impairment, with about 42% of the affected children unable to walk independently[5]. Four types of cerebral palsy have been distinguished: spastic (70%), dyskinetic (dystonic) (10%), ataxic (5%), and mixed (15%). Spastic cerebral palsy is characterised by central motor neuron damage and manifests in increased muscle tone and, consequently, postural disorders and pain. In the dystonic form, muscle tone disorders and involuntary movements, such as dystonia, athetosis and chorea, dominate. Ataxic CP is characterised by cerebellar damage leading to ataxia, balance and postural disturbances, nystagmus and hypotonia[11,16]. Mixed cerebral palsy is diagnosed in patients with a combination of the above mentioned symptoms – usually co-occurrence of spastic and dystonic symptoms is observed[16].

The prenatal period (70–80%)	• Intrauterine growth retardation • Twin pregnancy • Gestational diabetes • Exposure to teratogens • Cocaine use during pregnancy • Intrauterine infection • Placental abnormalities • Rh disease
The perinatal period (about 6%)	• Premature birth • Intracranial haemorrhage • Low birth weight • Perinatal injuries • Perinatal infections • Perinatal hypoxia • Convulsions • Episodes of hypoglycaemia • Hiperbilirubinemia
The postnatal period (11–21%)	• Central nervous system infections • Injuries • Coagulopathy • Periventricular leukomalacia

Tab. 1. CP risk factors[11–15]

Other symptoms, including intellectual disability, are also observed in patients with CP. About 35–62% of patients with CP develop epilepsy, which usually manifests in the first year of life, and is more common in children with intellectual disability[18–20]. Furthermore, 6.9% of children are co-diagnosed with autism spectrum disorders[20]. Impaired vision (refraction disorders, squint, retinopathy, cataract) is reported in 20–50% of patients[21]. Hearing impairment and consequently speech disorders co-occur in 12% of children with CP, usually in the dystonic presentation. Patients may also develop dysphagia leading to malnutrition.

CP is a clinical diagnosis. It is usually diagnosed by the age of 2 years. The diagnosis is based on detailed medical history, including pregnancy and birth. Most children with CP receive a normal Apgar score after birth[16]. Medical history should be supplemented with neurological evaluation and regular monitoring of child's development from the first months of life. Unilateral domination observed in the first year of life, i.e. right or left hand preference, should raise paediatrician's suspicions[1].

In the case of clinical doubts, the diagnosis is extended to include head magnetic resonance imaging (MRI)[1], electroencephalography as well as metabolic and genetic testing[22]. Brain imaging abnormalities may be undetectable before the age of 2 years due to incomplete myelination and grey matter development. No MRI abnormalities are found in about 10% of children with CP[1].

Children with CP require comprehensive, multidisciplinary medical care. The treatment is based on early rehabilitation aimed at preventing muscular atrophy and postural defects. Bobath and Vojta concepts are the most common rehabilitation approaches. Drugs to reduce muscle tone, analgesics and antiepileptic agents are used as an adjuvant therapy. Muscle tension reduction using botulinum toxin is attempted in patients with severe spasticity[23–25]. Anatomical deformities require surgical interventions.

CASE REPORT

A 16-year-old girl was admitted to the Department of Paediatrics, Paediatric Nephrology and Allergology due to chest pain episodes persisting for 3 months and accompanied by numbness and reduced muscular strength in the left upper extremity. The patient experienced compressive and burning pain localised over the left sternal edge and radiating to the left armpit. The pain showed no correlation with physical exercise and responded poorly to analgesics.

At the age of 11 years, the girl developed severe pain in her right knee. Ultrasonography performed at that time revealed a fissure in the patella; therefore, exercises and knee relief were recommended. The pain again increased at the age of 15 years. The patient was consulted by an orthopaedist; swimming and joint relief were recommended. Furthermore, the girl experienced periodic pain in the thoracolumbar spine, temporomandibular joints and feet.

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Due to an eye defect, the patient remained under the care of an ophthalmic clinic.

Due to the unclear clinical picture and coexisting long-term musculoskeletal pain, medical history was supplemented with perinatal and infancy data. The girl was born at 27 weeks gestation with a birth weight of 1,140 g and an Apgar score of 3–6–6. In the postnatal period, the girl was diagnosed with grade 2 and 3 intraventricular haemorrhage, grade 2 respiratory distress syndrome and intrauterine infection. She underwent rehabilitation in the early childhood period, which was discontinued after the infancy period.

The girl was admitted in a good general condition. Physical examination revealed symptoms of overweight, smaller circumference of the left lower limb, and a postural defect in the form of an abnormal lateral curvature of the spine, flat feet and valgus knees (Fig. 1). Laboratory tests showed low inflammatory markers, normal hepatic and renal function markers, ionogram within normal range, and vitamin D below optimal levels (19.9 ng/mL). Due to periodic stabbing chest pain during hospitalisation, creatinine kinase (CK), CK-MB and troponine were measured, and were within normal limits. Cardiac echocardiography was performed and revealed an aneurysmal atrial septum with no evidence of leakage. Twenty-four hour Holter monitoring showed no heart rhythm disturbances, also during the episodes of chest pain. A 24-hour monitoring showed blood pressure normal for age. Once cardiological causes of the reported complaints were excluded, electromyography was performed and showed a positive neuromuscular hyperactivity test (ischaemic test).

Neurological evaluation additionally showed reduced muscle tone and strength in the left upper and lower limb, clonic knee reflexes with no patellar clonus and bilateral positive Babinski sign (Fig. 2). After consulting a physiotherapist, radiography was performed and revealed a sigmoid deformity of the thoracolumbar spine and excessive bilateral lateral patellar compression (with right side dominance). Contrast-enhanced brain MRI showed no abnormalities.

The entire clinical picture, left-sided spasticity, reduced muscle tone and remarkable perinatal history allowed for the diagnosis of mixed CP with minor spastic paresis on the left side. Furthermore, latent tetany and a postural defect in the form of an abnormal lateral curvature of the spine, flat feet and valgus knees were diagnosed. Permanent physiotherapeutic care as well as vitamin D and magnesium supplementation were recommended.

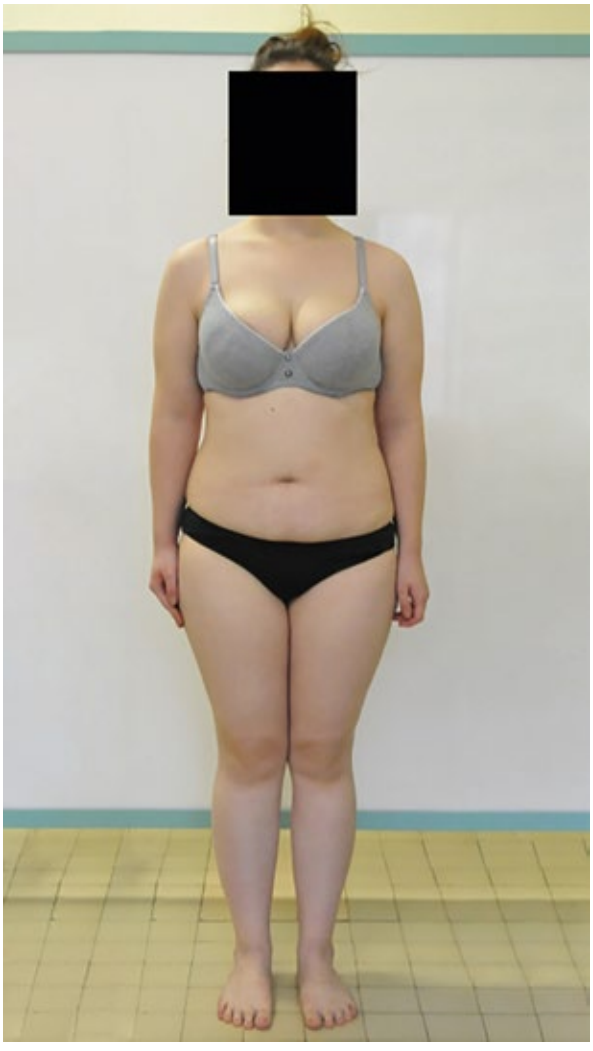

Fig. 1. Valgus knees, flat feet and overweight features in a 16-year-old girl with CP

DISCUSSION

We decided to present this case report due to an extremely late diagnosis of CP in a patient consulted by many doctors with narrow specialties. The clinical picture was not typical. Since bone pain was dominant, help was sought among specialists in orthopaedics. However, it should be noted that persistent pain may be one of the symptoms of CP. Monitoring of the child's development ensured by both a paediatrician and a neurologist is of key importance for the diagnosis.

Prematurely born children with low birth weight are at an increased risk of CP. In the described case, despite remarkable perinatal history and reported pain, the girl was not further examined or monitored by a paediatrician or neurologist.

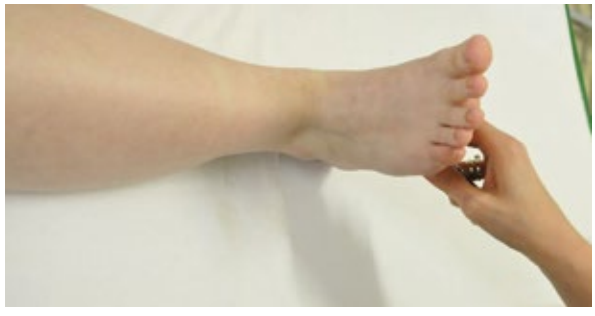

Fig. 2. A positive Babinski sign on the right side in a 16-year-old girl with CP

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Children with complicated perinatal history require detailed assessment during periodic health checkups.

CP should be differentiated from, among others, tetany, which may additionally mask CP symptoms. However, it should be noted that the diagnosis of tetany should not absolve the doctor from the necessity of performing neurological evaluation, especially in patients with complicated perinatal history and long-term clinical manifestations. In the described case, tetany was relatively quickly diagnosed due to easy access to electromyography. Long-term medical history and other symptoms prompted doctors to perform a further, thorough analysis.

The postural defect diagnosed by a physiotherapist is undoubtedly a consequence of neglected physiotherapy. The symptoms associated with this defect masked the final diagnosis of CP.

Conflict of interest

The authors do not report any financial or personal connections with other persons or organisations, which might negatively affect the contents of this publication and/or claim authorship rights to this publication.

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