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Visual symptoms in McCune-Albright syndrome – case report

Objawy oczne w zespole McCune-Albrighta – opis przypadku

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Summary: McCune-Albright syndrome is a rare complex genetic disorder. It is diagnosed on the basis of bone lesions – fibrous dysplasia, accompanied by at least one additional symptom: hyperactivity of endocrine glands or cafe au lait skin spots. We present symptoms, clinical picture and diagnostic procedure in a 15-year old patient with visual disorders in the course of McCune-Albright syndrome. The ophthalmic disturbances were the part of described syndrome. The active behaviour of the ophthalmologist in multidisciplinary diagnostic procedure led to the establishment of a proper diagnosis and optimal treatment.

Słowa kluczowe: zespół McCune-Albrighta, zaburzenia widzenia, dysplazja włóknista, plamy skórne typu cafe au lait.
Key words: McCune-Albright syndrome, visual disorders, fibrous dysplasia, cafe au lait skin spots.

McCune-Albright syndrome is a rare complex genetic disorder. It is diagnosed on the basis of bone lesions – fibrous dysplasia, accompanied by at least one additional symptom: hyperactivity of endocrine glands or cafe au lait skin spots. Fibrous dysplasia may cause deformity of long bones, spinal column and the skull, leading to pain, pathological fractures and compression of nerves. Intracranial placement of lesions may manifest as decreased visual acuity and visual fields changes. We present symptoms, clinical picture and diagnostic procedure in a 15-year old patient with visual disorders in the course of McCune-Albright syndrome. The ophthalmological disturbances were the part of described syndrome. The active behaviour of the ophthalmologist in multidisciplinary diagnostic procedure led to the establishment of a proper diagnosis and optimal treatment.

McCune-Albright syndrome was first described in 1936 (1). The classical form manifests as fibrous dysplasia of the bones, cafe au lait skin spots and endocrine glands hyperactivity. It is a rare disorder and its occurrence is difficult to evaluate. The skeletal aspect, i.e. isolated fibrous dysplasia is present in the population with the frequency of 1:4000-10000 (2). Recent studies show that McCune-Albright syndrome is closely connected with somatic mutation of the gene encoding protein G_s, which is a transmitter between extracellular receptors and intracellular effector proteins (3).

The leading symptom, i.e. fibrous dysplasia, may cause deformities in the long bones, spinal column, costae and skull, leading to pain and pathological fractures (4,5). Intracranial placement of lesions may manifest as decreased visual acuity and visual fields changes. Cafe au lait skin spots in McCune-Albright syndrome present as large pigmented lesions mainly on the neck, trunk and spinal column (6). The most common form of endocrine glands hyperactivity in this syndrome is precocious puberty (5). Hyperthyroidism, acromegaly or Cushing syndrome are rare (5,7,8). Other abnormalities comprise hypophosphataemia, chronic liver failure or cardiac arrhythmias (5,6,9).

Case description

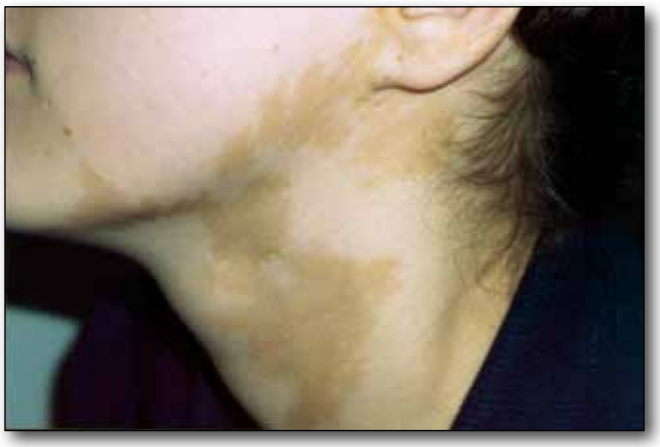
A 15-year old patient was admitted to our Clinic in 2003 because of visual defect in both eyes, progressive for the last few months. Past medical history revealed 8 episodes of bone fractures. For the last 3 years she was treated at the Rheumatology and Bone Metabolism Disorders Outpatient Clinic for osteoarticular pains. Connective tissue diseases, such as rheumatoid process, were excluded. Radiograms of her hands and forearms revealed decreased calcification, and within her right femur irregular bony formation. Densitometry showed decreased bone mineral density of osteopenia type.

On admission decreased visual acuity in both eyes was found: 0.4 and Sn (near vision): 0.75/30 cm, symmetrical position and movements of the eye, normal picture of the anterior eye segment and eye fundus. Refraction study showed normal sight, fluorescence angiography of the eye fundus showed normal condition of the choroid and retina vessels. The visual field was concentrically, irregularly diminished in both eyes to 10°, with peripheral islets of normal vision. In the whole area of the central visual field were present single scotomas– more numerous and more axially placed in the left eye.

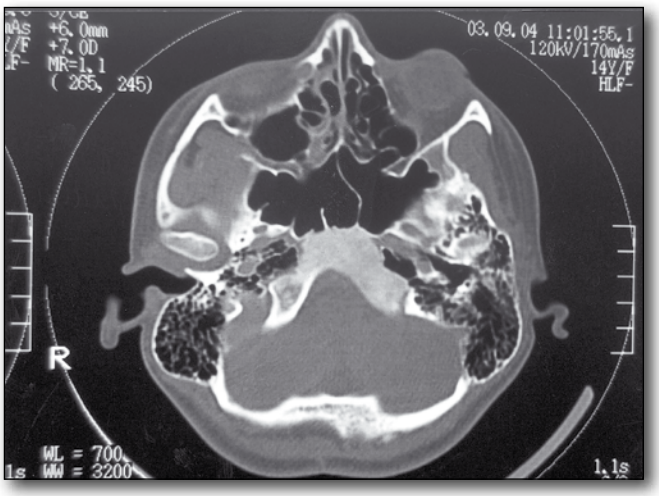
Examination of visual evoked potentials (VEP) using pattern reversal showed in both eyes symmetrical recording of normal amplitude (16N1) and normal latency time (P 103 ms).

General examination of the patient revealed irregular pigmented skin spots, cafe au lait type, on the face, neck and left epigastric area (fig. 1).

Imaging studies, including computerised tomography (CT) and magnetic resonance (MR) of the head, showed pathological areas in the sphenoid bone reaching to the top of the sella and occipital squama, with trabecular structure of the bones destroyed (fig. 2).



Ryc. 1. Plamy skórne typu cafe au lait.
Fig. 1. Cafe au lait skin spots.



Ryc. 2. Tomografia komputerowa: zmiany wewnątrzczaszkowe w obrębie stoku kości klinowej i łuski kości potylicznej.
Fig. 2. Computerized tomography scan shows intracranial changes in the sphenoid bone and occipital squama.

Due to intracranial changes the child was admitted to the Department of Neurosurgery. Histopathology of an occipital bone sample revealed the presence of fibroblasts and no trabeculae. The clinical picture and results of diagnostic tests led to the diagnosis of McCune-Albright syndrome.

Discussion

The diagnosis of McCune-Albright syndrome is established on the basis of visible changes in the bones – fibrous dysplasia, accompanied by at least one of additional symptoms, endocrine hyperactivity or cafe au lait skin spots (6). In Polish literature there are no reports on visual disorders in McCune-Albright syndrome, in foreign literature there are very few reports on this subject. The patient described by us did not have endocrine glands hyperactivity. Cafe au lait skin spots were observed late – at the age of 15 years, while usually they develop already in the neonatal period (6). Histopathology of a sample of occipital bone and typical radiogram of intracranial lesions, described as „ground glass", led to the diagnosis of fibrous dysplasia. The osteoarticular pains and bone fractures in patient's history are most probably connected with the McCune-Albright syndrome.

Fibrous dysplasia usually develops multifocally, within the femur or skull, which was also the case in our patient (2,5,10). Decreased visual acuity in McCune-Albright syndrome may concern one eye or both eyes and is slowly progressing. Usually it is connected with changes within the orbit, which lead to narrowing of the optic nerve canal, causing compression of the optic nerve. In such cases surgical decompression of the optic nerve may improve visual acuity (2,10). Rapid visual loss is observed rarely, and is caused by vascular lesions, usually within the ocular artery (2). We did not observe dysplastic deformations in the optic nerves canals in described patient. The decrease of the visual acuity and visual field defects could have their cause in the fibrous dysplastic lesions within the sphenoid bone demonstrated on CT scans. Prominent distension of the sphenoid bone slope reaching to the top of the sella could be the cause of compression in the region of the optic chiasm. It is possible that further observation will reveal cranial areas of dysplasia requiring surgical intervention. At the moment treatment with bisphosphonate is recommended, which is the treatment of choice in fibrous dysplasia (11). Bisphosphonates may decrease pain, but the results of the treatment in developmental age, in terms of biochemistry, radiology and histopathology, are not satisfactory (12).

In conclusion, it should be stressed that visual disorders in children may be one of the symptoms of a complex group of congenital defects, the McCune-Albright syndrome. The active role of ophthalmologist in multidisciplinary examinations is helpful for establishing proper diagnosis and optimal treatment.

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