McCune-Albright Syndrome with Exophthalmos: A Case Report

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Case Report

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Abstract

Background: McCune-Albright syndrome (MAS) is a rare disease defined by the triad of polyostotic fibrous dysplasia of bone, skin spots, and precocious puberty. No available treatment is effective in changing the course of fibrous dysplasia of bone, but symptomatic patients require the therapeutic support to reduce bone pain and prevent fractures and deformities. We reported 1 case of McCune-Albright syndrome with exophthalmos in clinical practice.

Case presentation: A 35-year-old female was admitted to our hospital who complained about "skin pigmentation for 35 years, vaginal bleeding for 30 years and progressive skeletal deformity for 28 years and exophthalmos for 2 years. And after the examination, she was been diagnosed with "McCune-Albright syndrome with exophthalmos". We highlighted the pathogenesis and development of the disease in this rare condition.

Conclusion: McCune-Albright syndrome with exophthalmos due to multiple fibrous dysplasia is rare but can be seen in clinical practice.

Background

McCune-Albright syndrome (MAS) is a benign osteopathy with unknown etiology and sporadic features, including a series of clinical features, such as multiple fibrous dysplasia, irregular brown pigmented spots on the skin margins, and endocrine disorders [1-2]. The incidence is extremely rare, with an incidence of about 1/100000-1/1000000 [3]. Eye features [4]: Owing to skull and orbital involvement, exophthalmos, papilloedema, optic atrophy and visual field defect may occur. The degree of damage depends on the position of bone lesion, and congenital cataract has also been reported. Now we report 1 case of McCune-Albright syndrome as follows:

Case Presentation

The patient was a 35 years old female. The main cause was skin pigmentation for 35 years, vaginal bleeding for 30 years and progressive skeletal deformity for 28 years and exophthalmos for 2 years.

1. ophthalmology examination after admission

She had an asymmetrical head and face, prominent left zygoma, prominent left eye appearance, showing a "bony lion face" like changes (Fig. 1). Her vision acuity was 3/25 in right eye and 4/25 in left eye and the best corrected visual acuity were 25/25 and 20/25 accordingly. Binocular conjunctiva were without congestion, and the cornea is clear. The anterior chamber depth was normal. The pupil was 3 mm in diameter and the light reflex was normal. The lens was transparent. The Fundus examination showed that the optic disc of both eyes was clear, and the colour of optic disc was pale, retina was orange-red,

and foveal light reflex was blurred(Fig. 2A, Fig. 2B).Exophthalmos examination showed left eye was more prominent than right eye about 2mm(14mm 115mm 17mm). The movement of the eye was not restricted in all directions. The visual field examination revealed a problem in the left eye and the irregular central darkness visual field could be seen in the left eye (Fig. 3A, Fig. 3B); however,There were no significant peripheral visual field reduction could be find. (Fig. 4A, Fig. 4B).While the visual field of the right eye was normal. Orbital CT scan displayed diffuse hyperplasia of craniofacial bones with ground glass appearance, many translucent regions scattered in distributionin in the skull,head and face asymmetry, frontal sinus, ethmoid sinus, maxillary sinus occlusion, orbital cavity smaller and supraorbital fissure, suborbital fission narrow (Fig. 5A, Fig. 5B, Fig. 5C).

2 .Patients detailed medical history

The patient was full term birth, weight 3.0kg at birth, the same height as the same age.Pigmentation was found in various parts of the body, including the mandible, neck, right upper arm, back, bilateral buttocks and left thigh. No skeletal deformity was found.

Walking began at age 1 and 9 months with normal gait. The growth rate of body was faster than that of the same age group before the age of 5, while the height was 6cm higher than that of the same age group.

Irregular vaginal bleeding began at the age of 5 years old, with a limited number of cycles and irregular periods, lasting 4-5 days each time With breast development, pubic hair growth.

The patient presented with walking deviation without bone pain and bone deformity at the age of 7 years old, Pelvic film showed "deviation of pelvis", and the skeletal age was 5-6 years earlier than that of the same age. Blood calcium was normal, ALP rises to 2000U/L.

Between the ages of 8 and 19, the patients underwent osteotomy and bone grafting and intramedullary nailing for hip and upper femur deformities, including 2 time left lower limbs and 1 time right lower limb. Spinal column "S" lateral curvature, left shoulder blade protruding and right shoulder invagination appeared gradually.

Transient ureteral calculi occurred during bed rest after the last operations at the age of 19. Walking was still unstable after operation, and the bone defects are progressively aggravated.

At the age of 21 years old, many teeth flaked off and gradually the whole tooth fell off.

At the age of 30 years old, black spots appeared on the skin of both cheek mucosa and finger of both hands. The area was gradually enlarged and the color was gradually deepened. The diameter was about 0.1-0.2 mm. The patient had severe eczema in both hands and feet, severe spring eczema, progressive frontal contraction and heavy pressure, intermittent vomiting with gastric contents ,non ejection vomiting.

At 33 years old, the skull and mandible began to deform. The zygoma on the right top was thick, the mandibular ramus was invaginated, and the mandible became sharper. Cranial CT showed "multiple cystic shadows in the mandible and zygomatic arch". The patient became unstable in walking and became shorter in height. The highest height was 158cm(20 years old), and then gradually shortened. At present, the height was 139cm.

Discussion

Fibrous dysplasia is a group of tumor-like lesions characterized by fibrous degeneration of bone. Normal bone tissue is replaced by elastic white fibrous tissue. Bone cortex becomes thinner. It is a benign bone disease that can involve single bones or multiple bones. This disease is commonly seen in children, with a slow course, skeletal deformities and pathological fractures. MAS is the most serious type of this disease. It is more common in female [5]. MAS was first described by McCune and Albert Right in 1937, including multiple fibrous dysplasia, irregular brown pigmentation on the skin margins, and endocrine abnormalities. Sexual precocity is the most common endocrine abnormality [6, 7, 8, 9]. The above three main manifestations may occur at the same time or not, skin pigmentation may not occur in some patients, except for sexual precocity, some endocrine abnormalities may not appear until older. Skeletal lesions often involve craniofacial bones, ribs, pelvis and so on except limbs bones, which cause asymmetric deformation of the head and face, and skull changes affect the surrounding tissue structure. About 25% - 30% of these patients have craniofacial bone involvement, most commonly frontal bone, followed by sphenoid and ethmoid bone [10]. Facial skeletal involvement can cause vision loss, exophthalmos, nasal congestion, sinusitis, facial asymmetry, hearing loss. When the sphenoid ridge is involved, the most common symptom is exophthalmos. Lee thinks that the simple optic canal involved or optic canal stenosis would not cause vision loss [5,11]. Our case showed that the visual acuity of the patients did not decrease significantly, but the irregular central dark spot appeared in the visual field of the left eye. This may be related to the development of the condition, which resulted in the decrease of the orbital content and the increase of the orbital pressure. A study by Wong SC [12] and others suggests that patients with McCune-Albright syndrome may also be at risk of developing tumors. Imaging diagnosis is very important for McCune-Albright syndrome, especially for multibony type FD and MAS [13]. The involved craniofacial bone imaging has three features: ground-glass changes, osteosclerosis, and cystic bone changes [14-15].

There is no specific treatment for this disease. Patients must undergo regular Department of Endocrinology examination and follow up. The diphosphate is commonly used to relieve bone pain and improve osteolytic lesions [16, 17]. Calcium and vitamin D may also be effective in some patients, and clinical assessment is being carried out [18]. Since MAS can cause a series of damage, individualized treatment must be carried out for each patient. Generally speaking, with the development of bone, the disease tends to stabilize gradually, so surgery is generally postponed to adulthood. In some cases, surgical intervention can be considered ahead of schedule for functional and aesthetic considerations [19]. Some small lesions can be completely resected, but the operation plan should be

based on the patient's age, lesion location, functional limitations, whether there is an impact on the appearance of comprehensive consideration.

Conclusion

MAS is the most serious type of fibrous dysplasia. It is more common in female and when the sphenoid ridge is involved, the most common symptom is exophthalmos. Individualized treatment should be carried out according to patients' different conditions.

Declarations

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Availability of data and materials

All data generated and analyzed during this study are included in this article.

Authors' contributions

Design of the study (J. Z.); Data collection, analysis and interpretation (J. Y.). J. Z. drafted the article and analyze the data. All authors reviewed the manuscript. All authors read and approved the final manuscript.

Ethics approval and consent to participate

No ethical approval required and the patient have give the written or verbal informed consent to participate in this study.

Consent for publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of written consent is available for review by the editor of this journal.

Competing interests

The authors declare that they have no competing interests.

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Figures

Figure 1

Facial appearance: head and face asymmetry, widened orbital distance, prominent left eyeball, a "bony lion face" like changes

Figure 2

Fundus photography showed that the boundary of the optic disc was clear, the color was pale, the retinal veins and arteries were normal, and the central foveal reflection was not clear

Figure 3

Central visual field examination, normal right eye, and central scotoma of left eye

Figure 4

Peripheral visual field was not significantly reduced

Figure 5

5A: Coronal CT scan of orbit: prominent left zygoma, occlusion of maxillary sinus, ethmoid sinus and sphenoid sinus, scattered skull in multiple areas of light 5B: Coronal CT scan of the orbit: the greater wing and wing of sphenoid bone were obviously thickened, showing a ground-glass appearance, supraorbital fissure and suborbital fissure narrowing 5C: CT scan of the orbit: thickening of the greater wing of sphenoid bone temporal bone, and ethmoid sinus occlusion

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