Adult Niemann-Pick disease type B with myositis ossificans: a case report

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ABSTRACT

Niemann-Pick disease (NPD) is a rare autosomal recessive lysosomal lipid storage disorder. It is caused by mutations of genes which products are involved in the metabolism of sphingolipids. Their dysfunction causes sphingomyelin to accumulate in different organs which leads to progressive multisystemic disorder. Type A and type B NPD are caused by mutations in sphingomyelin phosphodiesterase-1 gene with deficiency of acid sphingomyelinase (ASM). Type C and type D NPD have normal or reduced sphingomyelinase activity but differ pathogenetically from type A and B. The various types share common clinical features and the severity of the disease varies depending on the gene mutation, enzyme deficiency and the system involved. The estimated incidence of type A and B NPD is 1:250000 and of type C is 1:150 000 live births.

ANAMNESIS

His past anamnesis and family history revealed:
- failure to thrive, walking and talking difficulties
- hepatosplenomegaly diagnosed at the age of 2;
- psychomotor retardation and deterioration of neurological development;
- diagnosis of paranoid schizophrenia at the age of 32;
- family history of a brother who died from pneumonia, a death sister with a history of asthma, a brother with hepatosplenomegaly and a cousin with Niemann-Pick disease type B.

LABORATORY DATA

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PLAIN RADIOGRAPHS

- dysmorphic face with gothic palatum;
- chest deformity, inspiratory wheezing with normal breath sounds;
- hepatosplenomegaly – up to 3cm for the liver and 8 for the spleen;
- difficulties walking, limited range of motion in the hip joints, internal rotation of the right hip joint, pain and limited range of motion in the right knee joint with a flexion contracture at 45°, pain and limited range of motion in the lumber spine;
- cerebellar syndrome, adiadochokinesis, quadriplegic syndrome, no abdominal reflexes, polyneuropathy syndrome;
- cognitive dysfunction.

PHYSICAL EXAMINATION

Pic. 1. Denser hilus, reticular interstitial pattern of lung involvement, without infiltrates;
Pic. 2, 3, 4. Massive ossifications with amorphous character around the hip joints; right hip joint in internal rotation with internal rotation in the knee joint, high foot arches, a heel-spur on the right side. Hip, knee and ankle joints – with normal joint spaces.

CT IMAGES

Pic. 5, 6, 7, 8. Multiple massive ossificans and exostoses around the iliac bones and greater trochanter bilateral, in the internal and external obturator muscles, gluteus medius and minimus and quadratus femoris muscle. The images were suggestive of myositis ossificans.

CONCLUSION

It is important to raise the awareness of this debilitating condition and the need of a multidisciplinary management of such patients. As there is no recognized effective treatment for this disorder the possibility for prenatal diagnosis through amniocentesis or chorionic villus sampling especially in familial cases is of great importance.