INTRODUCTION

Congenital muscular dystrophy (CMD) is a rare heterogeneous muscle disorder characterized with muscle weakness, hypotonia, joint contractures and delayed gross motor development after the delivery. CMD is the most common neuromuscular disease inheriting as autosomal recessive. Fukuyama (FCMD), a type of congenital muscular dystrophy, is a rare type of CMD. It is characterized with cranial, cerebellar, ocular and congenital malformation muscular dystrophy. There is a mutation on 9Q31-33 in affected patients. Early started hypotonia, severe growth retardation and convulsions are seen in approximately half of patients at the first year of life.

ABSTRACT

Muscular dystrophy is an inherited group of disorders that affects skeletal and many other systems. It is transferred to the next generations with autosomal recessive trait. Congenital muscular dystrophy is a rare disorder characterized by findings emerging from birth. There are 12 different forms of mutation according to defects. Fukuyama syndrome is a rare form of congenital muscular dystrophies in our country. There is FKTN gene mutation. Because it is a rare disease in Turkey, we find this case to be worthy of presentation. The delivery, patients with recurrent convulsion and hypotonia were admitted to pediatric emergency department. Patients were diagnosed as Fukuyama congenital muscular dystrophy after evaluation based on clinical findings, imaging techniques and gene analysis. Congenital muscular dystrophy should be considered, whereas it is a group of disease in which hypotonia and recurrent convulsions are seen in early infancy period.

KEY WORDS: Fukuyama syndrome, Autosomal recessive disorder, Hypotonia, Convulsion.

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Therefore, the brain is also affected in Fukuyama syndrome. At the last group, it results from mutations in the gene of SEPN1, encoding an endoplasmic reticulum protein whose function is unknown. Because it is a rare case in the world this 6 month-old female patient was considered to be worthy of reporting.

**CASE REPORT**

A 6-month baby girl was brought to our emergency department with the complaints of recurrent convulsions by her family. The baby was born on time and did not have any problems at her history; breast-feeding and food supplement were given together, but she was poor-sucking, weak-crying and unable to get weight and her hands and arms were loose. She was not able to keep her head constant and got over convulsion four times without fever since she was born. The baby was seen as pale-looking and had a general hypotonia on physical examination. The baby could not keep her head and showed growth retardation (weight: 4.800gr, height: 57cm, head circumference: 39cm, weight and height below the 3rd percentile). There was no evidence for active convulsion when the patient arrived.

The patient was hospitalized after blood analysis, urine analysis and the imaging. Creatine kinase was 412 U/L on laboratory analysis, as abnormality. Blood and urine amino acids and tandem mass metabolic screening tests were found as negative. Blood gases were normal. On magnetic resonance imaging, there were hydrocephalus, diffuse signal increase in white matter (Fig.1), polymicrogria in occipital lobe and subcortical band type heterotopy in occipital again splenium agenesis of corpus callosum (Fig.2), cerebellar polymicrogria (Fig. 3 and 4), brainstem hypoplasia and vermian hypoplasia (Fig.5). Eye examination revealed the chorioretinal degeneration. Muscle biopsy showed dystrophic changes. FCMD was diagnosed on chromosome analysis via detecting mutation at 9Q31.

Fig.1: Axial T2 image of hydrocephalus, diffuse signal increase in white matter polymicrogria in occipital lobe and subcortical band type heterotopy in occipital.

Fig.2: Axial flair image of occipital horn expansion and parallelism due to splenium agenesis of corpus callosum.

Fig.3-4: Coronal T2 and sagittal flair image of disorganized cerebellar folia (cerebellar polymicrogria).
and 33 (Fukutin gene). Hydrocephalus was treated with ventriculo-peritoneal shunt by neurosurgery, and physiotherapy treatment was given. She was recalled for the purpose of control and follow-up for the physical therapy recommendations about hypotonia.

**DISCUSSION**

Muscular dystrophy is an inherited group of disorders that affects many systems, as well as muscular and skeletal system. Whereas some of the patients show immediate symptoms at delivery that result in death progressively, some of them may not show up any symptoms until late adult.5 Muscular dystrophy (MD) is separated from other neuromuscular diseases by primary myopathy that is genetically transitive, shows progressive outward, becomes degeneration of muscle fibers and death.5 Mutations that occurs in 12 different genes create CMD forms.6-8 These forms are collected under three groups. There is a mutation on FKTN gene that is responsible from production of protein called fukutin.9 Skeletal muscles which are used for movement of the body are affected.

The first symptom of the disease appears in early period of infancy, and contains weak cry, poor nutrition and the reduction in muscle tone (hypotonia).10 The elevation of transaminase and creatine kinase levels were remarkable in laboratory findings. It is usually diagnosed in suspected patients with their clinical and laboratory findings after further examination as a result of being seen related to radiological imaging and genetic analysis. Findings of lissencephaly were seen on cranial MRI. Dilated lateral ventricle, abnormalities of white matter, brainstem hypoplasia and cerebellar polimikrogiri were seen.11

The characteristic findings which possess to dystrophic muscle are seen on EMG. Fifty percent of patients respond to anti-epileptic therapy.12 Our patient was referred to us with recurrent convulsions, and typical symptoms were seen on laboratory findings and cranial MRI during further examination that was requested due to suspicion to hypotonia. In these types of cases suspecting some diseases have great importance. Although cases, who have recurrent convulsions accompanied by hypotonia, are uncommon in our country, CMD should be considered.

**REFERENCES**


**Contribution of Authors:**

Ibrahim Silfeler, Vefik Arica and Ramazan Davran: Literature search and collecting data from patients. Ibrahim Silfeler, Murat Tutanc, Fatmagul Basarslan: Design and support for the article in English. Ibrahim Silfeler: Prepared the final draft for publication.