Clinicopathological Conference by Dr.Suchart Phudhichareonrat

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History summary:

An 8-year-old Thai boy was brought to the Royal Thai Army Hospital by his mother for surgical correction of his calcaneovarus deformities of both feet. In the clinic, the history of frequent falls while walking was obtained. His past history included that he was born at full term without history of birth asphyxia. He could do neck extension, rolled over and walked at the age of 6, 8 and 18 months respectively. Physical examination revealed a good conscious boy. His pupils were 3 mm and reacted to light normally. He had normal chest movement during respiration. He had mild hypotonia and hyporeflexia (1+) of all extremities. In addition to his calcaneovarus deformities, he also had scoliosis.

Investigation:

CPK was 50 U/L (24-195 U/L). DNA study for SMA was negative. MRI whole spine showed moderate-severe dextroscoliosis at upper thoracic and levoscoliosis at thoracolumbar level. NCV and EMG revealed no F wave response.

Pathology:

Muscle biopsy was done. Findings in the muscle were small type I muscle fibers, an increase in central nuclei as well as endomysial connective tissue, adipose tissue infiltration, type I fiber predominance and hypertrophic muscle fibers.

Diagnosis :

Congenital fiber type disproportion.

Discussion :

Congenital fiber type disproportion (CFTD) is non or slowly progressive muscle disease. Weakness and hypotonia at birth or shortly after is found. Skeletal abnormalities such as congenital hip dislocation, joint contractures, foot deformities and spinal deformities are noted. Small type I muscle fibers (Ø smaller than type II muscle fibers >12%) in the absence of other significant pathologic findings are the pathological changes in muscle tissue. Patients with CFTD have mild to severe respiratory involvement (30%), difficulty swallowing (30%), skeletal abnormalities (25%) and cardiac involvement (<10%).

However, other changes in CFTD muscles such as central nuclei, moth-eaten fibers, nemaline rods (Brooke 1973), central cores, multicores (Iannaccone et. al. 1987) and hypertrophic muscle fibers (Sharma et. al. 2004) had been described.

Genetic involvement in CFTD (Chr. 1) varies from TPM3, ACTA1, SEPN1 and RYR1.

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ในการประชุมวิชาการ 11th Asian and Oceanian Myologic Center's Annual Scientific Meeting
ซึ่งจัดขึ้นระหว่างวันที่ ๖ – ๙ มิถุนายน ๒๕๕๕ ณ ประเทศญี่ปุ่น