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**Muscle glycogen storage disease type 0b presenting recurrent post-exercise loss of consciousness with weakness and myalgia**

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Background: Muscle glycogen storage disease type 0b (GSD0b) is a disease due to muscle-specific glycogen synthase 1 (GYS1) deficiency resulting in profound muscle and heart glycogen deficiency.
Objective: To characterize a Japanese girl with GSD0b clinically, pathologically, biochemically and genetically.
Patient: An 11-year-old girl was admitted to NCNP hospital because of post-exercise loss of consciousness, weakness, and myalgia. At age 5 years, the first episode occurred while climbing upstairs. She fainted away and needed a few hours to recover. One year later, she had a similar episode with limb muscle weakness and pain after running 50 meters. Since then, she repeated similar episodes several times a year. The episodes occurred just after exertion with muscle weakness in lower limbs making her squat down, gradual loss of consciousness, awakening after a few hours, and then strong myalgia in lower limbs. Systematic investigation including electrocardiography, echocardiography, brain imaging, electroencephalography, and screening of metabolic diseases revealed no abnormality. At age 11 years, forearm ischemic exercise test showed lack of lactate elevation.
Muscle biopsy demonstrated remarkable glycogen depletion on PAS staining. Activity of GYS was significantly reduced and GYS protein was absent in her skeletal muscle. *GYS1* gene analysis revealed compound heterozygous mutations in c.1230-2A>G and c.1810-2A>G.
Discussion: GSD0b have been so far reported in two families including two children who presented sudden death after exertion. Our finding further emphasizes that normal cardiac finding does not exclude the possibility of cardiac involvement. Notably, our patient loses consciousness not suddenly but gradually, which is atypical for cardiac syncope, suggesting that metabolic defects, such as exercise-induced hypoglycemia and glycogen depletion in skeletal muscle may also contribute to the syncopal episodes.
Conclusion: We indentified the first Oriental patient with GSD0b.

糖原病 0b 型は、グリコーゲン生成に必要な酵素であるグリコーゲンシンターゼ（GYS）1 の欠損により、骨格筋や心筋で著明なグリコーゲン欠損をきたし、筋力低下や心筋症、不整脈、突然死を起こしうる疾患である。これまで運動後の突然死を生じた 2 例を含む、4 例の小児がスウェーデンとカナダから報告されており、いずれも *GYS* 遺伝子変異が検出されている。今回我々は、運動後の筋力低下と意識消失、筋痛を幼児期より反復する 11 歳日本人女児例を経験し、糖原病 0b 型と診断した。骨格筋の病理所見では著明なグリコーゲン欠損を認め、骨格筋における GYS 活性低下、GYS1 蛋白欠損、および *GYS1* 遺伝子変異を検出した。