Kids with autism may have gene that causes muscle weakness

13 April 2008

Some kids with autism may have a genetic defect that affects the muscles, according to research that will be presented at the American Academy of Neurology 60th Anniversary Annual Meeting in Chicago, April 12–19, 2008.

The study looked at 37 children with autism spectrum disorders who were evaluated for mitochondrial disease, which causes muscle weakness and prevents a child from being able to participate in physical activities and sports. Mitochondrial disease occurs when genetic mutations affect the mitochondria, or the part of the cell that releases energy.

A total of 24 of the children, or 65 percent, had defects in the process by which cells produce and synthesize energy in the muscles, or oxidative phosphorylation defects in the skeletal muscles.

“Most children with autism spectrum disorders do not have recognizable abnormalities when you look at genetic tests, imaging, and metabolic tests,” said study author John Shoffner, MD, owner of Medical Neurogenetics, LLC in Atlanta, GA, and member of the American Academy of Neurology. “But a subset of these children does have significant defects in this area. Identifying this defect is important for understanding how genes that produce autism spectrum disorders impact the function of the mitochondria.”

Source: American Academy of Neurology