Brain MRIs can help diagnose and classify muscular dystrophies. Above, a T2 image shows muscle changes in a patient with distal muscular dystrophy. Smooth cortex lacks atrophy; abnormalities of the corpus callosum and cerebellum and enlarged ventricles are consistent with Walker-Warburg syndrome (middle). Thick cortex is indicative of muscle-eye-brain disease (MEB) abnormality (right).

**Findings From Brain MRI AID: Muscular Dystrophy Diagnosis**

**by Amy Rothman Schonfeld**

**Contributing Writer**

**Montreal** — Characteristic changes on brain MRI can help diagnose and differentiate muscular dystrophies with brain and eye abnormalities, reported Dr. Jim Vassar. Congenital muscular dystrophies (CMs) with brain involvement share several common features, explained Dr. Vassar at the 16th International Child Neurology Congress. All are autosomal recessive diseases that are characterized by early-onset hypotonia and weakness, delayed motor development, cognitive impairment, and epilepsy. Mutations in genes encoding proteins that are involved in protein trafficking and intracellular transport are responsible for most CMs.

Immunohistochemically, CMs with brain involvement can be grouped into those with a deficiency in laminin-211 chain (also known as merosin) or in α-tubulin, said Dr. Vassar, a neurologist affiliated with the Hospital for Sick Children in Toronto.

MRIs of children with laminin-211 chain deficiency show easily identifiable abnormalities in myelinated areas, although the corpus callosum and optic radiation remain normal. Despite the white matter abnormalities, these children maintain good cognitive function; however, about 30% are prone to seizures. It is more difficult to generalize about the appearance on MRI of α-tubulin-deficient CMs because several CMD phenotypes exist, said Dr. Vassar. As a general rule, MRIs of patients with CMDs show abnormalities in the posterior fossa, such as flattening of the pons, cerebellar hypoplasia or dysplasia, cerebellar cysts, and hypoplasia or absent vermis. The cortex takes on a cobblestone appearance, with disorganized cortical layers due to abnormal neuronal migration, multiple, abnor-

For example, in the most severe type of CMD, Walker-Warburg syndrome (WWS), MRI findings include type II liposchyliphic (cortical smoothening) and cerebral atrophy. Ventriculomegaly and abnormalities of the corpus callosum and splenium are also common. Anterior (e.g., cataracts, microcornea, microphthalmia, lens defects) or posterior (e.g., retinal detachment, optic nerve atrophy) eye abnormalities are also frequent. Clinically, children with WWS are profoundly retarded, have seizures, and usually succumb to death within the first 3 years of life.

MRI findings in children with other CMD types show a spectrum of generally malformations, gray matter abnormalities, and white matter abnormalities, said Dr. Vassar. In muscle-eye-brain (MEB) disease, cortical, cerebellar, and corpus callosum/splenium abnormalities are less prominent than in WWS. Polymicrogyria and thickened cortex may be noted in the frontal and parietal cortices, while agryria, cortical thinning, and liposchyliphic may be evident in the occipital cortex.

MRI in patients with congenital muscular dystrophy (CMD) type 1C may show normal brain with or without cerebellar cysts, said Dr. Vassar. Occasionally, MRI shows other white and gray matter abnormalities, with minor anatomical variations and white matter abnormalities. Findings include locus testae, thalami, anatomic variants, and changes to T2-weighted sequences.