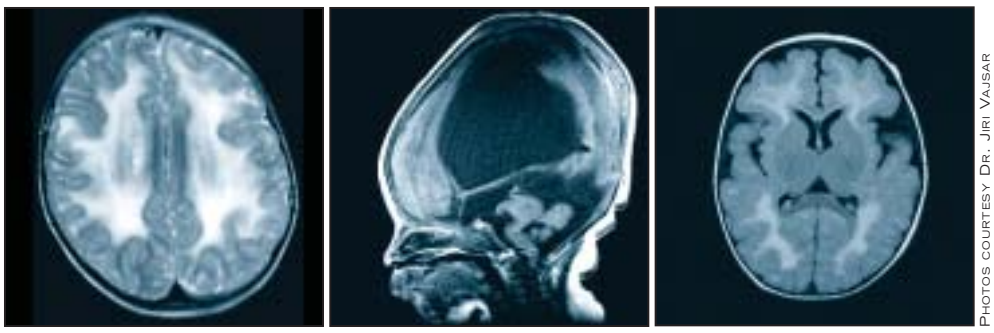

Photos courtesy Dr. Jiri Vajsar

Brain MRIs can help diagnose and classify muscular dystrophies. Above, a T2 image shows abnormal high signal in white matter (left) in congenital muscular dystrophy. Smooth cortex lacks sulcation; 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 migrational 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 congenital muscular dystrophies with brain and eye abnormalities, reported Dr. Jiri Vajsar.

Congenital muscular dystrophies (CMDs) with brain involvement share several common features, explained Dr. Vajsar at the 10th International Child Neurology Congress. All are autosomal recessive diseases that are characterized by early-onset hypotonia and weakness, delayed motor development, and elevated creatine kinase levels. Some CMD types also manifest mental retardation, delayed global development with subsequent regression, progressive contractures, seizures, variable ophthalmic findings, and cardiac and respiratory involvement.

Immunohistochemically, CMDs with brain involvement can be grouped into those with a deficiency in laminin-α2 chain (also known as merosin) or in α-dystroglycan, said Dr. Vajsar, a neurologist affiliated with the Hospital for Sick Children in Toronto.

MRIs of children with laminin-α2 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-dystroglycan deficient CMDs, because several CMD phenotypes exist, said Dr. Vajsar. As a general rule, MRIs of these disorders show abnormalities in the posterior fossa, such as flattening of the pons, cerebellar hypoplasia or dysplasia, cerebellar cysts, and hypoplastic or absent vermis. The cortex takes on a cobblestone appearance, with disorganized cortical layers due to abnormal neuronal migration; multiple, abnormal, and coarse gyri with agyric regions; and variable thickness.

For example, in the most severe type of CMD, Walker-Warburg syndrome (WWS), MRI findings include type II lissencephaly (cortical smoothing) and cerebellar malformation. Ventriculomegaly and abnormalities of corpus callosum and splenium are also common. Anterior (e.g., cataracts, microcornea, microphthalmia, lens defects) or posterior (e.g., retinal detachment, optic nerve atrophy, glaucoma) 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 milder gray and white matter abnormalities, said Dr. Vajsar. In muscle-eye-brain (MEB) disease, cortical, cerebellar, and callosum/splenium abnormalities are less prominent than in WWS. Polymicrogyria and thickened cortex may be noted in the frontal and parietal cortices, while agyria, cortical thinning, and lissencephaly may be evident in the occipital cortex.

MRI in patients with MDC type 1C may show normal brain with or without cerebellar cysts, said Dr. Vajsar. Occasionally, MRI shows other white and gray matter abnormalities, from cortical migrational anomalies and white matter changes to MEB-type or WWS patterns of abnormalities. ■