New Research in Hunter Syndrome (Mucopolysaccharidosis II) – The Signs and Symptoms a Neurologist Needs to Know.

European Neurological Review, a peer-reviewed, open access, bi-annual neurology journal published a cutting-edge article by Hernan Amartino.

(PRWEB UK) 4 June 2015 -- European Neurological Review, a peer-reviewed, open access, bi-annual neurology journal published a cutting-edge article by Hernan Amartino.

Abstract: Hunter syndrome (mucopolysaccharidosis II) is a rare X-linked lysosomal storage disease caused by deficiency of the enzyme iduronate-2-sulfatase. The condition is one of a group of disorders, the mucopolysaccharidoses, which all result in accumulation of glycosaminoglycans. Hunter syndrome is a chronic progressive disorder whose clinical manifestations vary widely in severity and involve multiple organs and tissues. In addition to developing somatic symptoms, patients having the neuropathic form of the disease also display developmental delay and cognitive impairment in early childhood that progressively worsens and that is severely life-limiting. Patients are at risk of developing secondary neurological manifestations, including hydrocephalus, vision and hearing loss, carpal tunnel syndrome and spinal cord compression. Common findings from brain magnetic resonance imaging (MRI) scans and at autopsy include neurodegenerative changes in white matter, the corpus callosum and basal ganglia; enlargement of periventricular spaces; ventriculomegaly; closed cephaloceles; and tissue atrophy. Though at present there is no specific treatment for the neurodegenerative aspects of the disease, hydrocephalus, carpal tunnel syndrome and spinal cord compression can be managed surgically. Patients who have Hunter syndrome should receive coordinated care from a multidisciplinary team: in light of the extensive neurological symptoms of the disease, neurologists play an important role in the diagnosis and management of this condition.

Hunter syndrome (mucopolysaccharidosis II, OMIM 309900), is a rare progressive X-linked lysosomal storage disease caused by deleterious mutations in the iduronate-2-sulfatase (I2S) gene, leading to a deficiency of the enzyme. I2S is required for the catabolism of the glycosaminoglycans (GAGs) dermatan sulphate and heparan sulphate; in the absence of I2S, these GAGs accumulate in tissues and organs. Hunter syndrome occurs with a reported incidence of 0.3–0.71 per 100,000 live births and almost exclusively affects males, though rare cases of females having the disease are known. Patients suspected of having Hunter syndrome are often first screened by assessing urinary GAGs (quantitative and qualitative tests are available). However, a definitive diagnosis requires enzyme assay in leukocytes, fibroblasts, dried blood spots or plasma, using substrates specific for I2S. Molecular testing can confirm the diagnosis and may be used to screen family members when the type of MPS and the family mutation is known.

Charles Hunter first described the condition in 1917 in two brothers presenting with developmental delay. Hunter syndrome is associated with multiple somatic symptoms affecting nearly every organ, including the cardiovascular, respiratory, gastrointestinal and endocrine systems. Patients also develop characteristic dysmorphic facial features and stunted growth (see Figure 1).

To continue reading this peer-reviewed article in full for free please go to: http://www.touchneurology.com/articles/hunter-syndrome-mucopolysaccharidosis-ii-signs-and-symptoms-neurologist-needs-know

Note to Editors:
touchNEUROLOGY (a division of Touch Medical Media) provides independent, cutting-edge, peer-reviewed content from world renowned physicians, designed to lead the debate on health and to engage, inform, and support physicians in improving patient outcomes globally.

touchNEUROLOGY.com provides an international platform for peer-reviewed content from industry-leading journals alongside other carefully selected sources and aims to support physicians, clinicians and leading industry professionals in continuously developing their knowledge, effectiveness and productivity within the field of neurology.

Our portfolio of peer-reviewed journals, European Neurological Review and US Neurology comprise of concise review articles which are designed to keep busy physicians up-to-date with the latest developments in their field and serve as a key reference resource for the international neurology community.

http://www.touchneurology.com/
Contact Information
Barney Kent
Touch Medical Media
http://www.touchoncology.com
+44 2071933009

Online Web 2.0 Version
You can read the online version of this press release here.