HYDROCEPHALUS

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Communicating hydrocephalus (increased fluid in the ventricles of the brain) is a common, but often undiagnosed, problem in individuals with MPS I, MPS II, MPS VI and MPS VII and may be associated with some cortical atrophy (loss of brain cells). Children with greater neurological involvement have a greater risk of developing hydrocephalus. Early detection and treatment of hydrocephalus is believed to improve quality of life. However, neurosurgeons are often unfamiliar with the unique aspects of diagnosing communicating hydrocephalus in MPS and related diseases, creating a frustrating situation for parents.

Hydrocephalus was once known as “water” on the brain. The “water” is actually cerebrospinal fluid (CSF), a clear fluid surrounding the brain and spinal cord. The CSF protects the brain and spinal cord from injury by providing a liquid cushion, and is continually being produced, circulated and absorbed. Communicating hydrocephalus (non-obstructive hydrocephalus) is caused by inadequate reabsorption of CSF. The excessive accumulation of CSF results in an abnormal enlargement of the spaces in the brain called ventricles. This causes potentially harmful pressure on the tissues of the brain.

In infancy, the most obvious indication of hydrocephalus is often the rapid increase in head circumference or an unusually large head size. In older children and adults, symptoms may include one or more of the following: headache, vomiting, nausea, downward deviation of the eyes (called "sunsetting"), problems with balance, poor coordination, gait disturbance, urinary incontinence, slowing or loss of development, lethargy, drowsiness, irritability, or other changes in personality or cognition, including memory loss.

Hydrocephalus in individuals with MPS may develop very slowly over months or even years, therefore, the typical signs and symptoms of hydrocephalus are not commonly seen, such as ventriculomegaly (enlarged ventricles) or papilledema (swelling of the optic disk). The lack of either of these symptoms does not rule out hydrocephalus in individuals with MPS.

Hydrocephalus is diagnosed through clinical neurological evaluation and by using cranial imaging techniques such as ultrasonography, computer tomography (CT), magnetic resonance imaging (MRI), or procedures like lumbar puncture (spinal tap) or placement of a brain wire monitor to measure pressure, called inner cranial pressure monitoring, or ICP monitoring.

It is recommended that children with MPS have a “baseline” head scan (CT or MRI) at the time of diagnosis with a follow-up scan yearly by a neurologist to evaluate the size of the ventricles. If there is evidence of a progressive increase in ventricular size, a shunt should be considered. Because some individuals with MPS who are symptomatic for hydrocephalus do not have progressive ventricular enlargement, measuring intracranial pressure can be diagnostic and is recommended in these situations. Intracranial pressure is measured in millimeters of mercury (mm Hg) and the upper limits of normal are around 18 to 20 mm Hg. Intracranial pressure may

be measured at various points in time by performing a lumbar puncture or may be monitored over a short period of time by surgically attaching an ICP monitoring device.

Hydrocephalus is most often treated with the surgical placement of a shunt system, which is a pressure valve and flexible plastic tube. This system diverts the flow of excessive CSF from the ventricles to another area of the body, typically the abdominal cavity, where it can be absorbed as part of the circulatory process.

Prior to shunt surgery, the cervical (around the neck) region should be evaluated to determine if there is evidence of obstruction at the foramen magnum (base of the brain). Such an obstruction can place the spinal cord at risk of life-threatening injury. If an obstruction is identified, the surgeon may wish to consider a laminectomy (removal of a portion of the back of the vertebra) at the time of the shunt procedure. If a shunt is placed, specialists recommend a high-pressure shunt (10-15 mm Hg) to prevent rapid decompression (reduction of fluid in the ventricles). In other instances, surgeons choose to use a programmable shunt, where the pressure setting may be adjusted electronically if necessary.

Children with MPS III routinely develop enlarged ventricles, but elevated CSF pressure may not present. In MPS III, the enlarged ventricles are believed to be due to cortical atrophy and not elevated CSF pressure. Robertson et al, European Journal of Pediatrics 157:653-655, 1998 reported that 6 children with MPS III with shunts had improvement in behavior and a decrease in level of agitation.

As with any surgical procedure in a child with an MPS condition, it is important to meet with the anesthesiologist prior to the surgery. For more information about hydrocephalus and shunts, please refer to the following websites:

http://www.ninds.nih.gov/health_and_medical/disorders/hydrocephalus.htm
http://www.hydrocephalus.org/ and http://www.hydroassoc.org