

## Wittestofziekten

# Emergency letter VWM

Patient's Name : .....

Date of Birth: .....

Medical Record Number: .....

This patient has been diagnosed with Vanishing White Matter (VWM) disease and is followed by the [.....Hospital Department] at [.....Hospital Name].

VWM is caused by a defect in the cell stress response. Patients with VWM are at risk for an episode of more rapid clinical deterioration following provoking the stress response by physical stresses, including head trauma, fever, illness, amino acid starvation, major surgery and anesthesia. Of potential emotional stress factors, only acute fright is known to sometimes provoke rapid deterioration.

Please contact the [.....Department Name] physician on call at the numbers below.

**Recommended Emergency Room Management:**

- Antipyretics for fever reduction -- treat even mild fever aggressively. Keep the temperature below 38°C.
- Investigate for a source of fever and provide supportive care -- use antibiotics liberally, even if a bacterial infection is not clear.
- Give fluids and start feeding within 24 hours. If unable to eat by mouth, consider NG tube. If not possible, add i.v. amino acids.
- Avoid major surgery and anesthesia as much as possible. If not avoidable, make sure that the nutritional intake, especially calories and amino acids, is guaranteed and consider prophylactic antibiotics and antipyretics. Avoid sevoflurane, which may provoke the stress response and cause decline. Propofol is the first choice, then Ketamine.
- Some antibiotics provoke the cell stress response and can better be avoided: Tetracyclines, Oxazolidinones, and Chloramphenicol

## Patient's usual medications:

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## Patient's usual diet:

The patient follows an unrestricted diet. Adequate caloric intake is important to avoid a catabolic state. **Prolonged fasting should be avoided.**

## General Information:

Vanishing White Matter (VWM) is a progressive neurologic disorder that causes deterioration of the central nervous system's white matter, which permanently affects transmission of brain signals to the rest of the body. VWM is caused by mutations in one of the five genes that collectively code for eIF2B, or eukaryotic initiation factor 2B, which is central in the cell stress response. The age at onset can range from early infancy to adulthood; in most cases affected individuals present between 2-6 years of age with regression of motor skills. MRI findings include symmetric involvement of the cerebral white matter and cystic degeneration over time. Progression of VWM is generally uneven, with periods of relative stability interrupted by episodes of rapid decline. **Affected individuals are particularly vulnerable to stresses such as fever, infection, mild head trauma or other injury, or acute fright. These stresses may trigger the first symptoms of the condition or worsen existing symptoms, and can cause affected individuals to become lethargic or comatose.**

[Department Name]: .....

[Department Phone #]:.....

after hours/weekends: [After-hours Phone #] .....

[Name of Department Head]: .....

[Names of Department Doctors]: .....