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LETTER TO EDITOR

Hunter Disease in Children (MPS II): New Aspects and Treatment Options

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Morbus Hunter is a life-threatening and inherited metabolic disorder, difficult to diagnose, difficult to treat. It is present in 1 in 162,000 newborn. Hunter's disease belongs to the group of lysosomal storage diseases. Due to X-linked inheritance, the disease affects almost only boys. In a few cases, female cases were described. In Germany, this is about 4-5 new cases per year. The cause of Hunter's disease is a defective gene on the X chromosome (Xq27.3-q28) These codes for iduronate-2-sulfatase. Due to the mutation, the degradation of dermatan and heparan sulfate is impaired. It belongs to the mucopolysaccharidoses (MPS), a group of diseases in which the lysosomal degradation of mucopolysaccharides is disturbed.

The manifestation ranges from severe forms with mental retardation (formerly type A) to very mild manifestations with little or no mental developmental delay (formerly type B). The transitions are fluid. The diagnosis is established by the increased excretion of mucopolysaccharides (dermatan and heparan sulfate) in the urine (electrophoresis). In addition, the defective enzyme is determined in leukocytes or fibroblasts. Molecular genetic analysis is possible, as is prenatal diagnosis in amniotic cells or chorionic villi (cells).

Specific for MPS type II are skin symptoms with pale, nodular, mostly grouped thickenings ("peau d'orange"). Other symptoms are facial changes: thick eyebrows, flat, sunken nasal bridge, fleshy, wide lips, enlarged tongue, prognathism, deep and hoarse voice, cardiac involvement up to heart failure, middle ear and sensorineural hearing loss, optic atrophy, early onset progressive joint contractures, distended abdomen due to developing hepatosplenomegaly, umbilical hernia growth retardation. Severely affected patients, after a period of aggressiveness and erectile dysfunction, may develop gross motor disturbances with gait instability and frequent falling, eventually tetra spasticity with dysphagia and respiratory disturbances. The signs of Hunter's disease are

different for each patient, can vary in severity, and can progress at different rates. The first symptoms usually appear between the ages of 2 and 4 and are usually an unusual combination of normal childhood symptoms, such as umbilical or inguinal hernias, middle ear infections, enlarged tonsils, recurrent infections, as well as organ enlargements, coarse facial features, and irregular teeth. It is important to remember that many of the disease features may resemble typical childhood complaints. Therefore, it is the combination of the various signs that may indicate Hunter's disease. Not all patients are affected by all disease features. Therefore, there is no classic disease course that is the same for all patients. Many signs and disease characteristics (symptoms) that may indicate Hunter's disease overlap with common childhood complaints. This is why Hunter's disease is so difficult to diagnose. This fact, combined with the extremely rare occurrence of the disease, means that it is often years before Hunter disease is diagnosed. There are a number of tests that can be performed to confirm or rule out the diagnosis of Hunter's disease.

There is no causative treatment available. The possibility of stem cell transplantation is possible in individual cases. Idursulfase (Elaprase) was the first enzyme replacement therapy to be approved in Europe in January 2007. Characteristics are the many, non-specific disease features (symptoms) that can occur and usually worsen with disease duration. Intravenous elaprase over 3 hours weekly is indicated for the long-term treatment of patients with Hunter syndrome (mucopolysaccharidosis II, MPS II). Elaprase is given weekly at a dose of 0.5 mg/kg body weight by intravenous infusion over a 3-hours period. This period may be gradually reduced to 1 hour if no infusion-related reactions are observed. Elaprase does not enter the brain barrier completely, so treatment should focus on research in this application.

No clinical studies have been conducted in heterozygous female patients. This treatment should be supervised by a physician or other healthcare professional experienced in the care of patients with MPS II disease or other inherited metabolic disorders. Further research is based on diminishing cognitive impairment and to develop a medication of Elaprase to enter the brain-barrier to prevent possible long term cognitive and neurodevelopmental impairment in Hunter disease patients. Further research in this extremely rare disease is necessary.