Mitochondrial neurogastrointestinal encephalomyopathy

Mitochondrial neurogastrointestinal encephalomyopathy , known as MNGIE syndrome, is a very rare autosomal recessive inherited multisystem disease that mainly affects the digestive and nervous systems. The disease can develop at any time during childhood or adulthood, but usually its main symptoms appear by the age of 20. So far, about 70 patients with this disease have been described worldwide.

Etiopathogenesis

The cause is a mutation in *the TYMP gene* (formerly known as ECGF1). This gene is responsible for making an enzyme called thymidine phosphorylase , which breaks down the nucleoside thymidine into smaller molecules, thereby regulating its amount in cells. Mutations in the TYMP gene reduce thymidine phosphorylase activity, resulting in increased cell concentrations. This results in an imbalance in the number of nucleotides required for mtDNA replication . As a result, mutations accumulate in the mtDNA and thus become very unstable. These genetic changes disrupt the normal function of mitochondria and result in impaired mitochondrial energy metabolism .

Clinical picture

Gastrointestinal system

Most patients with this disease suffer from gastrointestinal dysmotility , in which the muscles and nerves of the digestive tract do not move the digested food effectively. The result is a feeling of fullness even after eating a small amount of food, dysphagia, nausea and vomiting after eating, abdominal pain, diarrhea and pseudoobstruction. These difficulties lead to gradual weight loss and reduction of muscle mass (cachexia).

Nervous system

Manifestations of the nervous system are milder. These include tingling, stiffness and weakness of the limbs (peripheral neuropathy). Other neurological manifestations include eyelid ptosis , ophthalmoplegia and hearing loss. MRI shows leukoencephalopathy . _

Diagnostics

The diagnosis of MNGIE should be considered especially in patients with failure and intestinal motility disorders, who also develop neurological symptoms. The method of choice in diagnosis is to determine the level of thymidine in the blood, but for genetic counseling in the affected family, diagnostics at the enzymatic and molecular level is necessary.

Links

Resources _ _ _ edit source]

- BURGETOVÁ, Andrea, Pavel JEŠINA and VANĚČKOVÁ MANUELA. Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE) - a case report of a rare disease with radiological findings. *Czech Radiology* [online] . 2010, vol. 64, vol. 4, pp. 295-300, also available from < http://kramerius.medvik.cz/search/nimg/IMG_FULL/uuid:14838850-69b8-11e3-93fe-d485646517a0#page=1 >.

- Genetics Home Reference. *Mitochondrial neurogastrointestinal encephalopathy disease Print All* [online]. Last revision 2008-06, [cited. 2016-04-23]. < https://ghr.nlm.nih.gov/condition/mitochondrial-neurogastrointestinal-encephalopathy-disease >.

Related Articles _ _ edit source]

- Mitochondrial diseases
- Disorders of pyrimidine metabolism

External links _ _ edit source]

- Genetics home reference
- Journal of Czech Physicians - MNGIE Syndrome

Hereditary metabolic disorders (DMPs)	
In general	DMP of complex molecules • DMP of small molecules • Neonatal screening • Screening of hereditary diseases • Examination methods at DMP
DMP amino acids	Alkaptonuria
Organic aciduria	-
DMP urea cycle	Alcaptonuria • Ornithine transcarbamylase deficiency • Prolidase deficiency • Phenylketonuria • Glutaric aciduria • Hyperphenylalaninemia • Hyperornitinemia • Isovaleric aciduria • Leucinosis • Non-ketotic hyperglycemia • Cystinosis • Tyrosinemia
DMP propionate, biotin and cobalamin	Biotinidase deficiency • Methylmalonic acidemia • Propionic acidemia
DMP purines and pyrimidines	Liver porphyria • Skin porphyria • Mitochondrial neurogastrointestinal encephalomyopathy
DMP sugars	Glycogenoses • Fructosealdolase deficiency • Fructose-1,6-bisphosphatase deficiency • Essential fructosuria • Galactokinase deficiency • Galactose-1-phosphate uridyltransferase deficiency
DMP mitochondria	Phosphoenolcarboxykinase Deficiency • LCHAD Deficiency • MCAD Deficiency • Pyruvate Dehydrogenase Deficiency • Pyruvate Carboxylase Deficiency • SCAD Deficiency • Chronic Progressive External Ophthalmoplegia • Leber's Hereditary Optic Neuropathy • Leigh Syndrome • Maternally Hereditary Diabetes and Deafness • SayLC Syndrome
DMP peroxisomes	Neonatal adenodystrophy • Refsum's disease • Rhizomelic chondrodystrophia punctata • X-linked adrenoleukodystrophy • Zellweger syndrome
DMP of lysosomes	Fabry disease • Gaucher disease • Krabbe disease • Danon's disease • Mucolipidosis II • Metachromatic leukodystrophy • Mucopolysaccharidosis III • Niemann-Pick disease • Cystinosis • Tay-Sachs disease
Portal: Pathobiochemistry	

Category :

- Pediatrics
- Genetics
- Pathobiochemistry