Neurologic, Endocrine, and Pancreatic Disease, Multisystem, Infantile-Onset

Alternative Names
IMNEPD

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities > Other congenital malformations

Incidence per 100,000 Live Births
0-1

OMIM Number
616263

Mode of Inheritance
Autosomal recessive

Gene Map Locus
17q23.1

Description

Infantile multisystem neurologic, endocrine, and pancreatic disease (IMNEPD) is an extremely rare progressive multisystem disease characterized by intellectual disability, progressive cerebellar atrophy, postnatal microcephaly, failure to thrive, hearing impairment, polyneuropathy, and organ fibrosis with exocrine pancreas insufficiency. Patients invariably show exocrine pancreatic insufficiency with reduced pancreas elastase levels.

IMNEPD has been reported in only a handful of patients from less than 10 families. This disease is transmitted as an autosomal recessive pattern.

Molecular Genetics

Mutations in PTRH2 gene are the cause of infantile multisystem neurologic, endocrine, and pancreatic disease (INMEPD). This gene plays an important role in regulating cell survival and death; by an integrin-signaling pathway for cells attached or lost their attachment to the extracellular matrix (ECM). It interacts with transcriptional regulator amino-terminal enhancer of split (AES) to promote apoptosis in a process called anoikos.

Epidemiology in the Arab World

Saudi Arabia
Picker-Minh et al., (2016) descried five patients from two consanguineous Tunisian and Saudi families. All patients presented with neurological features including: intellectual disability, ataxia, motor delay, severe speech delay, and sensorineural hearing loss. In addition, they all had exocrine pancreatic insufficiency with reduced pancreas elastase levels that was partly associated with failure to thrive in the first years of life and consecutive deficiency of lipophilic vitamins. A homozygous missense mutation (c.254A > C) in the PTRH2 gene was detected in all affected patients.

Tunis
See [Saudi Arabia> Picker-Minh et al., (2016)]

References
Picker-Minh S, Mignot C, Doummar D, Hashem M, Faqeih E, Josset P, Dubern B, Alkuraya FS, Kraemer N1, Kaindl AM. Phenotype variability of infantile-onset multisystem neurologic, endocrine, and pancreatic disease IMNEPD. Orphanet J Rare Dis. 2016; 11(1):52. PMID: 27129381

Related CTGA Records
Peptidyl-tRNA Hydrolase 2

External Links
http://www.malacards.org/card/infantile_onset_multisystem_neurologic_endocrine_and_pancreatic_disease

Contributors
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