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Microvillous inclusion disease: What did we know so far

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Microvillous inclusion disease or microvillous atrophy is a rare congenital autosomal recessive disorder of the intestinal epithelial cells that presents with persistent life-threatening secretory watery diarrhea. MVID manifests either in the first days of life (early-onset form) or in the first few weeks of life (late-onset form). MVID is associated with consanguinity and arises mainly in the Middle East. All patients were from consanguineous families (from our study not yet published). The onset of diarrhea was in early neonatal period in all patients. One patient was weaned off PN. One patient underwent SBTx. Four patients are PN dependent and four died between (4 months-7 years of age). Different *MYO5B* mutations were detected in 8 families and a homozygous mutation in STX 3 gene was identified in one patient. Muller, et al. 2008 identified homozygous and compound heterozygous nonsense and missense mutations in *MYO5B*, encoding type-Vb myosin motor protein. Several studies have added to the understanding of pathophysiology of MVID by showing that *MYO5B* mutations were associated with disrupted epithelial cell polarity indicating that *MYO5B* gene has a role in the regulation of intracellular protein trafficking. These findings might have important implications for future treatment options for MVID patients. Moreover, mutations in a second gene, STX 3, causative for MVID-variant, a milder form had been reported recently in 2 patients. Although long-term survival has been achieved upon PN therapy, the prognosis is generally poor due to long-term PN related complications. Small bowel transplantation (SBTx) despite being complicated remains the only hope. However, recently, genome-editing techniques, including the overexpression of the corrected form of the defective gene, or the use of CRISPR (clustered regularly interspaced short palindromic repeats)/Cas9 to selectively correct the monogenic disease-causing variant within the stem cell, make autologous Intestinal Stem Cell (ISC) transplantation a promising future approach.

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