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Titel des Beitrags:
A novel mutation in the MFSD8 gene in late infantile neuronal ceroid lipofuscinosisis.

Abstract:
Neuronal ceroid lipofuscinoses (NCL) are lysosomal storage disorders and constitute the most common group of progressive neurodegenerative diseases in childhood. Most NCLs are inherited in a recessive manner and are clinically characterised by a variable age at onset, epileptic seizures, psychomotor decline, visual impairment and premature death. To date, eight causative genes have been identified to underlie various clinical forms of NCL. We performed a genome-wide linkage analysis followed by sequencing the recently described NCL gene MFSD8 in three affected and three unaffected members of a consanguineous Egyptian family with an autosomal recessively inherited progressive neurodegenerative disorder. The clinical picture of the patients was compatible with a late infantile NCL (LINCL); however, impairment of the visual system was not a cardinal symptom in the respective family. By linkage analysis, we identified two putative loci on chromosome 1p36.11-p35.1 and 4q28.1-q28.2. The latter locus (4q28.1-q28.2) contained the MFSD8 gene, comprising a novel homozygous missense mutation in exon 5 (c.362a>g /p.Tyr121Cys), which segregated with the disease in the three affected sibs. We describe a novel mutation in the previously identified MFSD8 gene in a family with
a common phenotype of LINCL, but no clinical report of vision loss. Our results enlarge the mutational and perhaps the nosological spectrum of one of the recently identified subtypes of NCL, called CLN7.