Diseases of dysfunctional mitochondria (aka mitochondrial diseases) are relatively rare with prevalence of 1/2000-4000. They predominantly affect children, however adult-onset disorders are also recognized. International collaborative effort of fifteen clinical and/or research centres from the UK, Germany, Ireland, France, Belgium, Austria Italy, Israel and Japan led by Dr. Michal Minczuk from the MRC MBU in Cambridge and Dr. Holger Prokisch from Institute of Human Genetics, Helmholtz Centre in Munich resulted in the discovery that mutations in the GTPBP3 gene cause defects in protein synthesis in mitochondria and are associated with a devastating disease. The patients affected by this disease have deficiency in energy production and suffer from heart and neurological disease.

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