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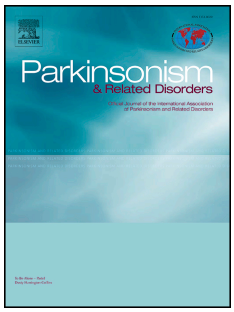

A Klinefelter patient with an additional mitochondrial mutation: Implications for genotype-driven treatment and mitochondrial mutational load in different tissues and family members

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A Klinefelter patient with an additional mitochondrial mutation: Implications for genotype-driven treatment and mitochondrial mutational load in different tissues and family members

Marija Dulovic, MD PhD[a,#], Eva Schäffer, MD[b,#], Frank Leypoldt, MD[b], Alexander Balck, MD[a], Susen Schaake, BS[a], Frauke Hinrichs, BS[a], Henriette Kirchner, PhD[c], Norbert Brüggemann, MD[a], Daniela Berg, MD[b], Katja Lohmann, PhD[a]

a. Institute of Neurogenetics, University of Lübeck, Lübeck, Germany
b. Department of Neurology, Christian-Albrechts University of Kiel, Kiel, Germany
c. Department of Internal Medicine 1, University of Lübeck, Lübeck, Germany

# These authors contributed equally to this work

Correspondence to: Katja Lohmann, PhD, Institute of Neurogenetics; University of Lübeck; Ratzeburger Allee 160, 23538 Lübeck, Germany; mailto:katja.lohmann@neuro.uni-luebeck.de

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Contributors:
Drs. Berg and Lohmann – Study concept and design
Drs. Dulovic, Schäffer, Leypoldt, Balck, Kirchner, Brüggemann, Berg, Lohmann, Mrs. Hinrichs, Schaake – acquisition of data
Drs. Dulovic, Schäffer, Leypoldt, Balck, Brüggemann, Berg, Lohmann, Mrs. Schaake – analysis and interpretation
Drs. Dulovic, Schäffer, Berg and Lohmann wrote the manuscript,
Drs. Leypoldt, Balck, Kirchner, Brüggemann, Mrs. Hinrichs, Schaake – critical revision of the manuscript for important intellectual content
Drs. Berg and Lohmann – study supervision

Many genetic disorders are clinically heterogeneous [1] and thus may be challenging to be diagnosed solely based on the clinical examination. In addition, if two or more genetic disorders are present in the same patient, the phenotypic presentation will be an additive result of both underlying disorders [2]. The scenario gets even more complicated when mitochondrial DNA (mtDNA) mutations play a