Patient data

Last Name: _____

First Name: _____

Date of Birth: _____ □ male □ female

Please enclose signed informed consent form!

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GENETIC TEST REQUISITION FORM FOR "CLINICAL EXOME SEQUENCING":

Clinical Diagnosis:

□ Hereditary neuromuscular disorder

□ Hereditary epilepsy syndromes

□ *other*, please specify: .....

This type of analysis can only be performed as "type 2" genetic analysis according to Austrian Law ("Gentechnikgesetz" §65, genetic diagnostics limited to medical purposes in order to confirm an existing disease caused by a germline mutation).

Explanations:

The planned genetic analysis of "whole exome sequencing" will employ a screening technique that allows to a analyze a multitude of human genomes in parallel (construction of a DNA library with Agilent SureSelect Clincial Research Exome v2 and "150 bp-paired end" sequencing employing Illumina's next generation sequencing technology, mean per base coverage 90-fold). Data analysis will be performed by (1) read mapping to the human reference genome (2) variant detection and (3) annotation of these variants followed by comparison with reference databases. The bioinformatic analysis will be **limited to genes with causal relationship to the presumptive clinical diagnosis according to literature and the OMIM database**. Detected mutations, which are formally supporting the clinical diagnosis, will be confirmed by targeted PCR followed by conventional DNA-sequencing.

It is important to note that (1) "whole exome sequencing" **cannot detect or may miss certain genetic mutations**, such as "deep intronic" mutations or structural variants (larger deletions / duplications) and (2) **some gene sequences** might be underrepresented or not captured at all and therefore **cannot be analyzed by this technique**.

This screening technique will generated a huge amount of data, interpretation of which will not be possible without clinical information. **In order to reach a final conclusive molecular genetic diagnosis, the integration of clinical data and possibly further examinations and analysis steps will be necessary.**

Therefore, we kindly ask you to contact us in advance!