JUSTIFICATION FOR WAIVER OF INFORMED CONSENT

Study Description

This study is a retrospective research project based exclusively on the analysis of clinical data that was previously collected as part of the routine clinical care of patients with CHKB-related myopathy. No additional interventions, examinations, procedures, or direct contact with patients will be conducted for the purposes of this research.

The data used for analysis are pseudonymized prior to extraction and processing. Investigators performing the analyses do not have direct access to patient identifiers. Data processing is conducted in accordance with the applicable requirements of the General Data Protection Regulation (GDPR), relevant national data protection legislation, and institutional policies governing the use of health data for research purposes.

The requirement to obtain individual informed consent is waived for the following reasons:

1. The study is strictly retrospective and involves only pre-existing data collected during routine clinical care.

2. No intervention, modification of patient management, or additional procedure is performed as part of the study.

3. All data are pseudonymized prior to analysis, thereby minimizing risks to patient privacy and confidentiality.

4. The research presents no more than minimal risk to the individuals concerned.

5. The study concerns a rare disease, and retrospective re-contact of all eligible patients would be impracticable and could introduce significant selection bias, thereby compromising the scientific validity and feasibility of the research.

6. Given the rarity of the condition, the historical nature of some of the data, and the potential geographical dispersion of patients, obtaining consent from every individual would represent a disproportionate effort relative to the minimal risk posed by the study.

The processing of health data is carried out for scientific research purposes and is subject to appropriate safeguards in accordance with Article 9(2)(j) of the General Data Protection Regulation (GDPR).

Where required by applicable regulations, patients are informed that their health data may be used for research purposes and retain the right to object to such use.