Myofin research: molecular genetics, pathogenetic mechanisms and diagnostic methods of neuromuscular disorders

DATA PROTECTION NOTICE FOR SCIENTIFIC RESEARCH
General Data Protection Regulation of the EU
Articles 12–14
Date: [1/2026]

Information on the processing of personal data in the research project entitled Myofin

The research project entitled Myofin involves processing of personal data. The purpose of this data protection notice is to provide information on the personal data to be processed, from where they are obtained and how they are used. Detailed information on the rights of data subjects will be provided at the end of this notice.

Your participation in the research project and provision of personal data are voluntary. If you do not wish to participate in the project or you wish to withdraw from it, you can do so without negative consequences.

1. Data Controller

Samfundet Folkhälsan i svenska Finland r.f. (”Folkhälsan”)
Address: PL 211 (Topeliuksenkatu 20), 00251 Helsinki

The Data Controller is the organisation defining the purpose and practises of the research.

2. Contact person and principal investigator

Contact person in matters concerning the research project:
Name: Ad. Prof. Vilma-Lotta Lehtokari
Address: Folkhälsan Research Center, Biomedicum Helsinki, P.O. Box 63, Haartmaninkatu 8, 00290 Helsinki
Phone: +358 2941 25000
Email: vilma-lotta.lehtokari@folkhalsan.fi

Principal investigator:

Name: Ad. Prof. Marco Savarese
Address: Folkhälsan Research Center, Biomedicum Helsinki, P.O. Box 63, Haartmaninkatu 8, 00290 Helsinki
Phone: +358 2941 25000
Email: marco.savarese@helsinki.fi

3. Contact details of the data protection officer

You can contact the data protection officer of Folkhälsan via email at dataskydd@folkhalsan.fi.

4. Description of the research project and the purpose of processing personal data

The Myofin research group belongs the research program of molecular genetics at Folkhälsan Research Center. The Myofin research group continues the work of Dr. Carina Wallgren-Pettersson's and Prof. Bjarne Udd's research groups founded in the 1990's on nemaline myopathy and muscular dystrophy, respectively, after the retirement of these two PIs. Hence, the two muscle research groups have been merged to form one group, continuing the work according to the previously approved research plans.

The purpose of the research project is to unravel the inherited (genetic) factors causing neuromuscular disorders (NMDs). The research group in Helsinki has mainly focused on identifying DNA variants causing NMDs and using a variety of different, usually so-called sequencing, methods. Sequencing means reading the unique DNA or RNA code of a person. Sometimes, only a part / parts of the genetic code is analysed, while sometimes the whole genome is investigated. In addition, to improve the identification of the genetic cause leading to an NMD, we continuously develop and optimise new analysis methods. We also may investigate whether the cause is a large duplication or deletion in a person's genome. Moreover, we try to find out how various mutations cause alterations in muscle proteins and how these protein alterations in turn give rise to a muscle disorder.

In this study, we use samples from the research project participants and often from their close relatives also. Sample types used are blood and DNA samples, muscle and skin biopsies, and other biological samples. The muscle biopsies have most often been taken in the context of the diagnostic process.

In addition, we collect information related to the muscle disorder of the participant. Examples of such data are the degree of muscle weakness in different muscles, data on the breathing of the patient, any joint contractures, and the muscle biopsy findings (where one has been performed). The information is collected using a data sheet filled out by the clinician caring for the patient, on request by the person in the research group carrying the medical responsibility for the project. The purpose of this form is for the scientists to investigate the symptoms and /or course of the muscle disorder in relation to, for example, the genes and genetic alterations identified. The long-term goal is to improve the care of patients with NMDs and to help develop specific therapies.

The results gained through the samples and data regarding research participants and their families and the data on their causative genetic variants are handled confidentially. When the samples and clinical data are received by the research group, each participant is assigned a personal identification number. Thereafter, names or other identifiable information are not used. The data are accessed and handled by the research group only using the anonymous

identification number, and only the principal investigators and the research assistant have the right to access and handle personal information when needed. The investigators are acting under professional secrecy.

If needed, consultations may take place with other parties in the interpretation of results. In that case, all results are shared without personally identifiable information, so that the data cannot be linked to the original person. In these cases, only information necessary for the consultation are shared with persons outside the research group.

5. Persons who have the right to process the data

Principal investigator: Marco Savarese

Project leaders (co-PIs) and administrative leaders:
Katarina Pelin (katarina.pelin@helsinki.fi), nemaline myopathy and related disorders
Peter Hackman (peter.hackman@helsinki.fi), muscle dystrophies and other neuromuscular disorders.

Address: Folkhälsan Research Center, Biomedicum Helsinki, P.O. Box 63, Haartmaninkatu 8, 00290 Helsinki
Phone: +358 2941 25000

The research is performed by the researchers and the research assistants of the Myofin group as well as by visiting researchers, and clinicians involved.

Visiting researchers are employees or students of an organisation other than Folkhälsan; University of Helsinki, Helsinki University Central Hospital, University of Turku and / or University of Tampere. All visiting researchers are required to sign the confidentiality agreement and data management agreement with Folkhälsan Research Center.

Expert clinicians responsible for the clinical data are senior physician, neurologist Manu Jokela (Turku University Hospital and Tampere University Hospital Neuromuscular Research Center) and Maria Francesca di Feo, medical doctor specialised in medical genetics (Folkhälsan research center). Both emeriti, Carina Wallgren-Pettersson and Bjarne Udd, continue as consulting experts in the group.

6. Personal data included in the research data

Data collected about the participant include data on the genetic causes possibly affecting their muscle disorder, or their clinical picture, i.e. data derived from the DNA or RNA sample analysed. In this study, such findings are the diagnostic findings in the muscle biopsy and data on the participant's muscle symptoms and/or their progression, and information related to the

heredity of the disease. In addition, the participant's name, reported gender, number of affected and healthy family members, and date of birth are registered.

The clinical and family data are collected using a clinical data sheet filled out by the physician caring for the participating patient, with permission from the participant.

7. Sources of personal data

The clinical data are gained from the physician caring for the patient, using a clinical datasheet.

The samples (see Chapter 2) are collected under the guidance of the referring clinician according to the practises and standards of the unit caring for the patient. The Myofin group isolates DNA, RNA or protein from the tissue samples, and/or sometimes cell lines are established from the tissue samples.

In case previously performed studies include information relevant for the research, the Myofin group may request these results from the original institute or hospital upon the permission of the participant.

8. Sensitive personal data

The following special categories of personal data (i.e., sensitive data), as defined in Article 9 of the GDPR, will be processed in this research:

☒ Ethnic origin
☐ Political opinions
☐ Religious or philosophical beliefs
☐ Trade union membership
☒ Genetic data
☐ Biometric data for the purpose of uniquely identifying a natural person
☒ Health
☐ Sex life or sexual orientation of a natural person

The processing of sensitive personal data is based on Article 9(2)(j) of the General Data Protection Regulation (processing is necessary for scientific research purposes), as well as Section 6, Subsection 1, Paragraph 7 of the Finnish Data Protection Act.

☐ Personal data concerning criminal convictions or offences will be processed in the research project.

The processing of data concerning criminal convictions or offences is based on Section 7, Subsection 1, Paragraph 2 of the Finnish Data Protection Act (the processing of personal data concerning criminal convictions or offences or related security measures is allowed for scientific research purposes).

9. Lawful basis for processing personal data

Personal data are processed on the following basis (Article 6(1) of the GDPR):

In non-commercial scientific research, it is recommended that the lawful basis for processing personal data be 'performance of a task carried out in the public interest: scientific or historical research purposes or statistical purposes'.

☒ Task carried out in the public interest:
- ☒ Scientific or historical research purposes or statistical purposes
- ☐ Archiving of research material and cultural heritage material

☐ Consent by the research subject
☐ Compliance with a legal obligation to which the controller is subject
☐ Legitimate interests pursued by the controller or by a third party
Specify the legitimate interest:

If the processing of personal data is based on the research subject's consent, he or she can withdraw that consent at any time. The withdrawal of consent does not affect the lawfulness of processing based on consent before its withdrawal.

10. Recipients of data

The research group can, if necessary, send a DNA sample (or a tissue sample) from the participant to another laboratory, if Folkhälsan Research Center does not have the methods needed available. The samples are sent pseudonymized, without any identifiers which could be linked to the participant. If the research group need a second opinion or consultation from an expert of another group or clinical department, the data will be shared and discussed without direct personal identifiers.

Collaborators:

Prof. Francesco Muntoni	University College London, United Kindom
Prof. Nigel Laing	University of Western Australia, Australia
Prof. Gina Ravenscroft	University of Western Australia, Australia
Prof. Mathias Gautel	Kings´ College, United Kingdom
Prof. Andreas Roos	University of Essen, Saksa
Prof. Vincenzo Nigro	Telethon Institute Of Genetics and Medicine, Italy
Prof. Coen Ottenheijm	University of Amsterdam, Netherlands
Prof. Alan Beggs	Harvard University, U.S.A.
Prof. Giorgio Tasca	Newcastle University, United Kingdom
Prof. Volker Straub	Newcastle University, United Kingdom
Prof. Alexander Hoischen	University of Radboud, Netherlands

Service providers:

FIMM	Tukholmankatu 8, Helsinki, Finland
CEGAT	Paul-Ehrlich-Straße 23, 72076 Tübingen, Germany

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HUS Clinical Laboratory Geneticist, Dr Kirsi Kiiski, HUS Center of diagnostics, Helsinki

Servers:
Moonhill Ltd (nemaline myopathy database), Finland
FileMaker (sample database), University of Helsinki, Finland
Euformatics, Tekniikantie 12, 02150 Espoo, Finland
Lucid Genomics GmbH ; Machnowerstr 64, 14165 Berlin, Germany
CSC – IT Center for Science, Espoo, Finland

11. Transfer of data to countries outside the European Economic Area

Please select the appropriate option.

During the research project, data may also be transferred to recipients in other countries. The groups of recipients are mentioned in section 10 above.

The transfer of data is based on the European Commission's adequacy decision or standard contractual clauses (https://ec.europa.eu/info/law/law-topic/data-protection/international-dimension-data-protection/standard-contractual-clauses-scc_en), whereby recipients of personal data agree to comply with the data protection requirements outlined in the clauses.

If your research project must transfer data containing personal data to outside the EEA, please contact outi.elomaa@helsinki.fi

12. Automated decision-making

The research project involves no automated decision-making that has a significant effect on data subjects.

13. Protection of personal data

Personal data included in the research dataset will be processed and kept protected so that only those who need the data can access them.

The data processed in data systems will be protected using the following:
☒ Username and password
☒ Registration/log of use
☒ Access control
☐ Encryption
☒ Two-factor identification
☒ Other, please specify: pseudonymization

Physical material (e.g., data in paper form or other tangible form) will be protected using the

following: in a locked facility in a locked cupboard/safe or in a facility that can be accessed by authorised persons alone.

Processing direct identifiers:
☐ The controller collects the personal data without direct identifiers.
☒ Direct identifiers will be removed during the analysis stage and kept separate from the analysed research data.
☐ The data will be analysed using direct identifiers, because (give grounds for preserving the direct identifiers):

14. Duration of the processing of personal data in this research project:

Personal data will be handled until the research has been completed. The personal identifiers are needed for example for reporting the possible findings to the participant (via the referring clinician or a clinician within the Myofin-group). The estimated duration of the research is until 2050.

15. Processing of personal data when the research project ends

☐ The research data will be deleted

☐ The research data will be kept for the purposes of validating or replicating the results of this research project:
☐ without identifiers ☐ identifiers included

☒ The research data will be kept for later, compatible scientific research in accordance with the requirements of the GDPR:
☒ without identifiers ☒ identifiers included

The storage of the research data is based on Article 5(1)(b) and (e) of the GDPR.
Data subjects will receive a new data protection notice on the new use of the research data, unless the controller can no longer identify the subjects from the data.

In addition, the data subjects will not be informed of the new research if delivering this information to them is impossible or involves a disproportionate effort or renders impossible or seriously impairs the achievement of the research objectives (Article 14(5)(b) of the GDPR).

Where and for how long will the data be stored: 2050 in secured in Folkhälsan servers.

16. Rights of data subjects and derogations from those rights

The contact person in matters related to research subjects' rights is the contact person stated in section 2 of this notice.

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Rights of data subjects

Under the General Data Protection Regulation, data subjects have the following rights:

- Right of access to their own data
- Right to rectification of their data
- Right to the erasure of their data and to be forgotten
- Right to the restriction of processing of their data
- Right to data portability from one controller to another
- Right to object to the processing of their data
- Right not to be subject to automated decision-making

However, data subjects cannot exercise all their rights in all circumstances. The circumstances are affected by, for example, the legal basis for processing personal data.

Further information on the rights of data subjects in various circumstances can be found on the website of the Data Protection Ombudsman: https://tietosuoja.fi/en/what-rights-do-data-subjects-have-in-different-situations.

Derogations from rights

The General Data Protection Regulation and the Finnish Data Protection Act enable derogations from certain rights of data subjects if personal data are processed for the purposes of scientific research and the rights are likely to render impossible or seriously impair the achievement of the research purposes.

The need for derogations from the rights of data subjects will always be assessed on a case-by-case basis.

Right to appeal

If you consider that the processing of your personal data has been carried out in breach of data protection laws, you have the right to appeal to the Office of the Data Protection Ombudsman.

Contact details:

Office of the Data Protection Ombudsman
Street address: Lintulahdenkuja 4, 00530 Helsinki
Postal address: PO Box 800, 00531 Helsinki
Phone (switchboard): 029 56 66700
Fax: 029 56 66735
Email: tietosuoja(at)om.fi