Patient informed consent for PEDIA study

(Prioritization of Exome Data by Image Analysis)

Proband data
Last Name: _____
First Name: _____
Date of Birth: _____ (DD/MM/YYYY)
Genetic variant: _____
(HGVS format: e.g. PIGU; NM_080476.4;c.209T>A;p.(Ile70Lys))

I acknowledge that the analysis purpose is to improve the genetic diagnosis of a patient's health issue. I have discussed the benefits, risks, and limitations of genetic tests and computer-assisted analysis of phenotype data that might enhance the interpretation of molecular diagnostic tests. I confirm that I understood the proband information and received a copy thereof.

I consent to the recording, processing and storing of digitized photographs, clinical information, family history information in a pseudonymous form.

Furthermore, I consent to share the data in pseudonymous form with colleagues of the study team and collaboration partner to improve quality of automated image analysis.

I am aware that I may withdraw consent at any time without giving reasons and with no adverse consequences. I understand that in such case all data will be deleted and not further processed.

_____
Place, date

_____
Signature of study participant or Guardian's signature

(optional)

☐ I consent for these photographs to be used in medical publications, including medical journals, textbooks, and electronic publications, I understand that the image may be seen by members of the general public, in addition to scientists and medical researchers that regularly use these publications in their professional education. Although these photographs will be used without identifying information such as name, I understand that it is possible to recognize the depicted person.

_____
Place, date

_____
Signature of study participant or Guardian's signature

Principal investigator's statement: I have explained computer-assisted image analysis and data interpretation to this individual. I have addressed the limitations of the test and have answered all stated questions.

_____
Place, date

_____
Signature of principal investigator