Supplemental data

Supplemental Table 1

MOLECULAR DIAGNOSIS OF GENETIC DISEASES
PRENATAL DIAGNOSIS
INFORMED CONSENT FORM

The undersigned _____, born in _____ on _____
resident in _____ address _____
partner _____, born in _____ on _____
Telephone _____, Mobile _____
doctor or department of origin
_____

authorises Prof./Dr. _____ of the Clinical Department of Laboratory Medicine (DAsMeLab)–CEINGE Biotecnologie Avanzate to make DNA extraction from the following fetal tissue:

☐ Chorionic villi ☐ Amniocytes ☐ Cord blood

and to carry out the prenatal diagnosis of the following disease _____ according to standardised protocols and reported in the international literature.

– I have been informed that if the quality and quantity of fetal DNA obtained be sufficient, analyses for the molecular diagnosis of _____ will be conducted on this material. These analyses consist of:

☐ Detection of previously identified molecular alterations in the _____ gene previously found
☐ In the index case ☐ In both parents ☐ In one of the parents (indicate which _____)
☐ Linkage analysis
☐ Presence of contamination by maternal DNA

– I have been informed that these analyses are restricted to this specific disease (_____) and that they cannot give information on other diseases possibly present in the unborn child. In addition, the accuracy of the molecular analysis is based essentially on the accuracy of the clinical diagnosis carried out elsewhere, therefore the Clinical Department of Laboratory Medicine (DAsMeLab)–CEINGE Biotecnologie Avanzate cannot be held responsible for erroneous clinical diagnoses.

Signature _____

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– In case of diagnoses based on linkage analysis, I declare that I have been informed in detail about the principle and the limits of this type of analysis. I have been informed that in this case the accuracy of the result could be 95%-99% due to events of recombination that could determine a diagnostic error.

Signature _____

– I have been informed that the molecular analysis can take from one to three weeks, starting from the day of the fetal tissue sampling. If it is necessary to prepare a cell culture with the fetal material collected, the result of the analysis may be available from three to five weeks after the sampling.
– I have been informed that should the fetal material be insufficient, I may be asked to undergo another sampling.
– I have been informed that should the fetal material be contaminated by maternal material, it will not be possible to carry out the analysis and/or I can be asked to undergo another sampling.

Signature _____

– I have been informed that, in the case of a prenatal diagnosis carried out without a previous molecular characterisation of the family, this analysis could be not informative.
– In addition I have been informed that in case of direct analysis of mutations, the analysis of fetal DNA is limited to the detection of parental mutations, therefore this analysis is based on the premise that **the family relationships declared are truthful.** Should this not be the case, the undersigned relieves DAsMeLab and its personnel from any legal or personal responsibility. I am also aware that DAsMeLab-CEINGE reserves the right to ask for a second blood sampling from the parents in order to avoid sampling errors.
– I have been informed that the type of analysis used for the detection of possible contamination by maternal DNA in the fetal tissue is based on a test that allows one to define the fetal alleles derived from the mother and father. I have been informed that this result, of which I will be made aware, is covered by professional secrecy. Other people will be informed only if I give my written consent or if required by the legal authority.
– I have been informed that in the case of dissonance between the declared and the biological paternity, the accuracy of the test may not be 100%.

Signature _____

– I have been informed that the results of the analyses will be given, in a written report and during multidisciplinary counseling, only and exclusively to me, the expectant mother requesting the analysis, and eventually in the presence of my partner, and not, for any reason, by telephone. The results of the analyses will not be given to anyone without my written permission, and my data will not be made public, according to the personal data privacy code. In addition I agree that the fetal sample that is no longer required for the diagnosis can be used anonymously for clinical research and/or epidemiology records.

Signature _____

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Other possible consequences of the prenatal diagnosis

- I have been informed, I understand and I accept that the analysis:
  Can take longer than scheduled
  May not provide useful results
- I have been made aware of all the above that I declare I understand and accept as possible complications of the entire analytic procedure.

Signature _____

- I have been informed and I accept that the procedures used for these analyses can entail the following **risks**:

Risks of laboratory error

In all laboratory examinations there is a probability, albeit low, of a laboratory error. I have been informed that DAsMeLab-CEINGE takes all the possible precautions to prevent and avoid a laboratory error that, however, could occur, as in any human activity.

Risks of sample loss

One of the risks of the laboratory analysis of samples from human tissues is that the sample could be lost, damaged or made useless for the analysis. I have been informed that DAsMeLab-CEINGE takes all possible precautions to prevent this eventuality. Nevertheless, I am aware that there is always a possibility that this could happen as a possible risk of this examination. In this case, I will be informed and eventually I will undergo a new sampling.

Risk of stress and emotional damage

One of the effects of this examination is that I can suffer from stress during the execution and the completion of the analysis and while awaiting the result. Neither the institute nor the personnel in charge of the analysis can be held responsible for this effect.

Signature _____

Information related to the prenatal diagnosis

I declare that I have received and examined the information regarding the prenatal diagnosis and I undertake to examine it in detail.

Signature _____

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This informed consent has been collected by Prof. / Dr.:

Name _____ Signature _____

In the presence of Drs.:

Name _____ Signature _____

Name _____ Signature _____

Additional questions asked by Ms _____

_____

_____

_____

_____

_____

_____

_____

Signature _____

The answers to the above-mentioned questions have been provided by Prof./Dr.:

Name _____ Signature _____

In the presence of Drs.:

Name _____ Signature _____

Name _____ Signature _____

Naples, _____