



ISTITUTO C.S.S. MENDEL
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GENETIC STUDY OF JOUBERT SYNDROME AND RELATED DISORDERS

Dear Colleague,

If possible, we kindly request you to follow this protocol, more “laborious” than a normal dispatch of blood or DNA, since it allows our lab to obtain not only DNA but also RNA from the samples, which use will greatly ease the mutation analysis of JSRD causative genes. In case you are not able to follow the protocol (especially guarantee the samples delivery within 24-36 hours from sampling), when the sample arrives in lab we will perform only DNA extraction. In this case, we will still perform mutation analysis of known genes, but this will require a longer analysis time and will limit the number of detectable mutations.

Thanks for your collaboration!!

PROTOCOL FOR BLOOD SAMPLING

1. Explain to the parents the objectives and possible outcomes of the study: this is a research based project, thus no official response will be issued but we will inform the referring clinicians of any positive results. Ask the parents to sign the INFORMED CONSENT FORM (attached). In case they require further clarifications on the study, please ask them to get in touch directly with the study coordinator (Dr. Enza Maria Valente, Istituto CSS-Mendel, at: e.valente@css-mendel.it).
2. Draw 10 ml of blood in EDTA tube from the patient, parents and any available healthy or affected siblings.
3. Keep the samples refrigerated at +4°C (NOT frozen!!!) until dispatch, that should be planned as soon as possible, within a few hours from sampling.
4. Pack the samples properly for dispatching: place tubes in a close package, make sure that they won't break. If possible, add an ice pack in the package to keep the samples refrigerated (please DO NOT place the ice pack in direct contact with the samples, since the blood would freeze).
5. Dispatch the sample by express courier (we usually use DHL that works well for blood transport, especially for international deliveries). The package should arrive here preferably within 24-48 hours from sampling to allow RNA extraction.
6. Attach a signed declaration stating that the samples are not dangerous or infective (such as the one reported below).



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e-mail: e.valente@css-mendel.it

7. Send the package to the following address:
Dr. Enza Maria Valente / Dr. Lorena Travaglini
CSS-Mendel Institute
viale Regina Margherita 261
00198 ROME
tel. +39-06-4416 0537 or 0513
fax. +39-06-44160548
8. Make sure that the package is delivered to our lab between Monday and Friday morning (the lab is closed over the weekends).
9. Please sent an email (e.valente@css-mendel.it and/or l.travaglini@css-mendel.it) to state that the samples have been dispatched and the expected delivery time. If possible, specify the courier and the expedition code, to allow us to track down the sample in case of unexpected delays.

DECLARATION

I (undersigned), declare that these blood samples are neither pathogenic nor toxic, pose no risk of disease infection or transmission and in no way can be dangerous.

Yours sincerely,

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Place and date:

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