

Mutational Analysis for Congenital Neutropenia

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Patient:

Date of Birth:

Requested molecular genetic analysis:

1. Single gene analysis for patients with positive family history:

ELANE *HAX1* *G6PC3* *JAGN1* *SRP54* *CSF3R* other

2. For patients with unclear etiology:

Neutropenia Panel Whole Exome Sequencing (*only after consultation*)

Material: 1 - 5 ml heparinized bone marrow **OR** 5 - 10 ml EDTA blood **OR** DNA

Date Drawn:

Sender stamp:

Physician's name in block letters

Phone number for consultation

For a better assessment of the relevance of molecular genetic findings for severe chronic neutropenia, we ask you to answer the following questions:

1. Is the patient registered in the SCNIR? Yes No

2. Has the SCNIR been consulted? Yes No

3. Clinical neutropenia: permanent cyclic unclear

4. Etiology: congenital acquired unclear

5. Since when is the neutropenia known?

6. How low have been the levels of granulocytes on average?

<500/ μ l 500-1,000/ μ l 1,000-1,500/ μ l number of documented CBCs

When cyclic neutropenia is suspected: Have 3 CBCs per week been collected and has a cycling been documented?

Yes No

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7. Have anti-neutrophil antibodies been detected?

Yes No not tested

8. Has a bone marrow examination been performed? Yes No

If yes, has a maturation arrest been detected? Yes No (please attach report)

9. Therapy: G-CSF: Yes No other:

10. Did the following infections occur?

Otitis media Tonsillitis Pneumonia Skin abscesses
 Aphthae Sepsis other:

11. Have organ abnormalities been detected? (e.g. cardiac defects, kidney malformations)

Yes No If yes, which?

12. Are additional family members affected from neutropenia?

Yes No If yes, who?

13. Are the parents of the patient related by blood? (e.g. Cousins)

Yes No

Location, Date

Signature

Samples should be taken at the beginning of the week and sent off immediately by overnight express!

Please notify the laboratory prior to sending the sample:

Phone: +49 7071 2986014 or +49 162 2052224; Fax: +49 7071 2925161

E-Mail: Labor-SCNIR@med.uni-tuebingen.de

This analysis requires written consent of the patient or legal guardian (e.g. parents). Please attach the signed consent form. Without a signed form we are not allowed to perform the analysis. Please find the consent form on www.severe-chronic-neutropenia.org