Family Analysis
(Segregation Analysis) Form Epilepsy & Migraine

General Information

Dear consulting family member,

Dear referring clinician,

You have requested molecular diagnostics from CeGaT for a family member.

In molecular diagnostics, there are often **several variations in genes** identified, for which the relevance for pathogenesis of the disease of the patient might be inconclusive. These variations can be either **disease causing mutations** or only **rare variants**, which are not causative for the disease. If the variation is also present in a healthy parent or other non-affected family member, it would suggest that the variation only represents a rare variant.

Therefore, we would like to conduct a **family analysis (segregation analysis)**, in which we only determine the presence of these rare variants identified in the patient in additional family members.

We ask you to **accurately provide the following information (neurological medical history, page 3)** for each consulting family member. This enables us to create a targeted assessment of the molecular diagnostics results.

- **For family analysis (segregation analysis) we require the following:**
- A **declaration of consent** for genetic analyses (as listed on page 2, or equivalent), as required by German law, for each consulting family member (signed by the family member (or by legal guardian) and the referring clinician)
- **Material**: EDTA blood (min. 5 ml) or DNA (min. 1µg)
  Samples can be sent by mail in cardboard box or air cushion envelope. Samples should not be exposed to direct sunlight.
- The **medical history** form (page 3), completed for each consulting family member
- Invoice recipient
  - ❑ Patient
  - ❑ Sender / clinic

Contact
To discuss the diagnostic strategy
please do not hesitate to contact us.
Phone: +49 7071 565 44 55
Email: diagnostic-support@cegat.de

CeGaT is accredited by DAkkS according to DIN EN ISO 15189:2014 and by the College of American Pathologists (CAP).

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General Information

Family analysis (segregation analysis)

Concerning the following **patient** (for which a molecular diagnostics was requested from CeGaT):

Surname: _____ First name: _____

Date of birth: _____ CeGaT-No. (if known): _____

Information about yourself (consulting family member)

Surname: _____ First name: _____ Sex: ❑ male ❑ female

Date of birth: _____ Date of sample collection: _____

Transplants (bone marrow, tissue, stem cells) ❑ No ❑ Yes, (please specify) _____

Relationship to the patient

❑ Father ❑ Mother ❑ Brother ❑ Sister ❑ _____

Symptoms

Does this persons currently suffer or has suffered from the same or a similar disease as your patient?

❑ No ❑ Yes; symptoms?: _____

Inquiry (gene variant, familial mutation, Index ID, etc.)

Declaration of consent

By signing this form, I declare that I have received comprehensive information about the genetic background related to the disease in question as well as the possibilities and limitations of molecular genetic testing. I understand that I have the right to withdraw my consent to genetic analyses.

I have been informed, and agree, that the data obtained in the analysis will be recorded, evaluated, or stored in an anonymised form in scientific databases, and further, in accordance with data protection and medical confidentiality, that the request, or parts thereof, may be transmitted to a specialised cooperating laboratory.

If you do not check these boxes, your answer will be recorded as "no".

I consent to the storage of my genetic material for additional tests and/or quality control (for max. 10 years). ❑ Yes ❑ No

I consent to the storage of my test results beyond the timespan of 10 years (as required by German law). ❑ Yes ❑ No

I consent to the anonymous storage and use of surplus genetic material and/or test results for scientific research. ❑ Yes ❑ No

In certain cases, medical findings occur that are not connected to the inquiry. About these findings,

❑ I do not want to be informed ❑ I definitely want to be informed

❑ I only want to be informed when opportunities for treatment could be developed for me or my relatives

This declaration of consent can be completely or partially withdrawn at any time. I have had sufficient time to consider giving my consent.

Doctor's stamp / Barcode

_____ _____

Patient / legal representative (block letters) Doctor (Surname, First name)

X _____ X _____

Patient / legal representative (Signature) Doctor (Date, Signature)

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Anamnise

Neurological medical history

Name of consulting family member: _____

	No	Yes	
I currently suffer from **epilepsy**	❑	❑	if yes, which kind: _____
I have already had a **seizure**	❑	❑	if yes, when (age): _____
I had **febrile seizures** as a child	❑	❑	
I have or had **migraine(s)**	❑	❑	if yes: with auras / hemiplegia / other: _____
I have or had a **learning disability**	❑	❑	
I had **delayed speech / language development** as a child	❑	❑	
I had **delayed motor development** as a child	❑	❑	
I am affected by a **movement disorder**	❑	❑	if yes, which kind: _____
			❑ Persistent
			❑ Intermittent (duration: seconds / minutes / hours / days)
			❑ After physical exercise
I have or had **cardiac arrhythmias**	❑	❑	
I have frequent **muscle cramps**	❑	❑	
Are there further persons in your family who suffer or have suffered from symptoms listed above??	❑	❑	if yes, who: _____ which symptoms: _____

<u>Neurological examinations</u> performed

EEG	❑	❑	Abnormalities: no / yes (if yes, please include a copy of existing reports)
MRI	❑	❑	Abnormalities: no / yes (if yes, please include a copy of existing reports)

Additional comments

For further information and advice please do not hesitate to contact our Diagnostic Support team at any time.

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diagnostic-support@cegat.de
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