Erasmus MC
Universitair Medisch Centrum Rotterdam

Department of Clinical Genetics

Postbus 2040
3000 CA Rotterdam

General terms and conditions

1. Requests for testing

Requests for testing by the Department of Clinical Genetics can only be accepted when accompanied by a completed application form. The following application forms have been specifically designed for this purpose:

- Enzyme diagnostics
- Tumour cytogenetic testing
- Postnatal cytogenetic testing
- Prenatal cytogenetic testing
- Metabolic/basic diagnostics
- DNA diagnostics

If possible, make an imprint of the patient identification card in the appropriate space on the form. The form should be filled in completely and clearly and materials should be clearly identified to avoid errors and delays. Incomplete forms and unclear or incompletely identified material may be refused to avoid error. The sender shall be informed as quickly as possible.

2. Confidentiality

The Department of Clinical Genetics employs Privacy Regulations to guarantee data confidentiality. A copy of these regulations shall be sent upon written request.

3. Sampling protocols

Sampling and all related activities such as storage and transportation fall beyond the scope of the Department of Clinical Genetics. For sampling protocols (with the exception of prenatal diagnostics), please refer to the relevant application forms. For DNA diagnostics, please refer to the website **www.dnadiagnostiek.nl**. Each sample should be clearly identified, as indicated on the application form.

4. Sending and packaging

For sending and packaging sample material, please refer to the instructions on the relevant application form. Rush orders shall be handled after consultation by telephone (see the application form for the telephone number).

Packaging
The container should be well protected against breakage to minimise the risk of contamination when unpacking samples after receipt. Accompanying application forms should be packed separate from the samples, but in the same outer package.

5. Methodology and implementation

The Department of Clinical Genetics defines the method and equipment to perform the work according to applicable norms, standards and rules. The Department of Clinical Genetics can provide the applicant with this information upon request. For work in areas that the Department of Clinical Genetics has little or no knowledge or experience, the Department of Clinical Genetics retains the right to outsource testing within the established procedure.

6. Results

Results in the form of test results, advice, information or any other form are provided in writing. Rush work is possible upon request by phone, telefax or otherwise and remains the responsibility of the applicant.

7. Duration of the testing

The average duration of testing by section and by type of testing is reported below:

Prenatal cytogenetic testing
Chorionic villi: 1-2 weeks
Amniotic fluid: 2-3 weeks

Postnatal cytogenetic testing
8 weeks

Tumour cytogenetic testing
1-3 months (depending on the complexity of the karyotype)
Upon abnormal findings, an interim report shall be provided.

Prenatal enzyme diagnostics
Testing directly on chorionic villi: upon receipt on Monday or Tuesday: 1 - 4 days (if no further testing is required and with prior consultation about sending).
Testing of **cultured cells, chorionic villi or amniotic fluid**: 3 - 4 weeks (sub-testing can be completed sooner).

Postnatal enzyme diagnostics
Blood testing:
Lysosomal storage diseases and biotinidase: 2 - 3 weeks
Glycogenoses, CDG, galactosemia (GALT): 3 - 4 weeks
Others: 5 weeks
Testing of skin biopsies (incl. cell culture of 6 weeks):
Urea cycle defects: 6 - 12 weeks
DNA repair defects: 6 - 16 weeks
Organic/ (amino) acid urea: 6 - 12 weeks
Note: Sending cultured skin fibroblasts shortens the testing time by 4 weeks.
Testing of tissue biopsies (muscle/liver): 3 - 6 weeks
On average, results for mitochondrial diagnostics can be expected within 3 months..

DNA diagnostics
See website **www.dnadiagnostiek.nl**.

Metabolic / Basic diagnostics
Complete basic screening in urine: 6 weeks
Testing on plasma, spinal fluid, amniotic fluid and hair: 2 - 6 weeks
In the event of abnormal findings, an interim report shall be provided.

Note: For rush orders, other deadlines can be arranged. The Department of Clinical Genetics shall inform the applicant if testing is delayed due to special circumstances.

8. Use of patient material

If no agreements have been made when submitting the application for testing, the Department of Clinical Genetics shall either destroy or save the samples or their remnants after testing in accordance with its regulations.
The use of anonymised patient material may be desirable for developing and improving methods of analysis (checks and validation) and for further testing of the relevant illness. For further use of bodily samples, the Department of Clinical Genetics shall conform to the guidelines of the Code of Practice of the Dutch Federation of Biomedical Scientific Societies and the local Medical Ethics Committee, if applicable.
The Department of Clinical Genetics kindly requests that the applicant inform the patient about this. Patient objections to the anonymous use of bodily materials may be submitted in writing.

9. Quality assurance

The Department of Clinical Genetics uses a quality assurance system based on the ISO 15189-2012 guideline, M105.

TESTEN
RvA M 105