

Laboratory of Neurogenetics and inherited eye disorders

Director | Prof. Doutora Catarina Resende Oliveira

Head of Laboratory | Doutora Maria do Rosário Almeida

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SAMPLING AND TRANSPORTATION CONDITIONS

1. SAMPLE COLLECTION

- Per patient, two 3 ml EDTA blood samples should be provided.
- For the quantification of the progranulin concentration in serum, a clotted blood tube should also be provided.
- The sample tube must contain the following information: Full name of the patient and Date of birth of the patient.

2. INFORMATION REQUIRED

- All samples should be accompanied by the respective Request form completed and signed by the referring clinician
- Request forms for genetic testing can be ordered by email (mralmeida2008@gmail.com) or download from the website (http://www.cnbc.pt/services/serv_cnc06.asp)
- For predictive tests, a copy of the signed informed consent is required to be sent along with samples. A proof of genetic counselling is also required.
- Please note that payment must be received prior to testing. Please contact Rosa Fernandes; email: rosafolhasfernandes@gmail.com, for more information on payment

3. TRANSPORTATION MATERIAL

The samples can be sent by regular mail in an appropriate container at room temperature (preferable should reach our lab within 48hours) to:

Maria Rosário Almeida or Maria Helena Ribeiro
Hospitais da Universidade de Coimbra (piso +1)
Laboratório de Neurogenética
Praceta Prof. Mota Pinto
3000-75 Coimbra
Portugal

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4. TESTS AVAILABLE

Disease/ Test Available	Methods	Price (Euros)	Time referral
Alzheimer Disease			
All coding exons of Presenilin 1 gene (<i>PSEN1</i>)	PCR+sequencing	310 €	10 weeks
All coding exons of Presenilin 2 gene (<i>PSEN2</i>)	PCR+sequencing	310 €	10 weeks
Exons 16 and 17 of the Amyloid precursor protein gene (<i>APP</i>)	PCR+sequencing	125 €	4 weeks
Apolipoprotein E (<i>ApoE</i>) genotype	PCR+RFLP	105 €	4 weeks
Parkinson Disease			
All coding exons of Parkin gene (<i>PARK2</i>)	PCR+sequencing	450 €	10 weeks
G2019S mutation in exon 41 of the Leucine-rich repeat kinase 2 gene (<i>LRRK2</i>)	PCR+sequencing	105 €	4 weeks
Exons 9 and 10 of the Glucocerebrosidase gene (<i>GBA</i>)	PCR+sequencing	150 €	4 weeks
Frontotemporal lobar degeneration			
Exons 1, 9-13 of Microtubule-associated protein tau gene (<i>MAPT</i>)	PCR+sequencing	250 €	4 weeks
All coding exons of Progranulin gene (<i>PGRN</i>)	PCR+sequencing	450 €	4 weeks
Determination of Progranulin serum concentration	ELISA	60 €	8 weeks
Hexanucleotide Repeat Expansion in <i>C9ORF72</i> gene	PCR+sequencing	190 €	8 weeks
All coding exons of sequestosome 1 gene (<i>SQSTM1</i>)	PCR+sequencing	275 €	8 weeks
Familial British dementia / Familial Danish dementia			
Exon 6 of the <i>BRI2</i> gene	PCR+sequencing	125 €	4 weeks
Fatal familial insomnia			
D178N mutation in prion protein gene (<i>PRNP</i>)	PCR+sequencing	125 €	4 weeks
<i>PRNP</i> M129V polymorphism genotype	PCR+RFLP	115 €	4 weeks
Retinitis pigmentosa			
All coding exons of Rhodopsin gene (<i>RHO</i>)	PCR+sequencing	350 €	6 weeks
Nanophthalmia			
All coding exons of <i>MFRP</i> gene	PCR+sequencing	490 €	8 weeks
Anophthalmia / Microphthalmia (A/M)			
All coding exons of <i>OTX2</i> gene	PCR+sequencing	250 €	4 weeks
All coding exons of <i>RAX</i> gene	PCR+sequencing	250 €	4 weeks
All coding exons of <i>SOX2</i> gene	PCR+sequencing	200 €	4 weeks
Test for known familial mutation	PCR+sequencing	105 €	2 weeks