



**MOLECULAR PATHOLOGIC MARKERS OF HIGH RISK AND  
PREVALENCE FOR FAMILIAL HYPERCHOLESTEROLEMIA**

**INDEX PATIENT/ FAMILIAR INFORMATION** (obligatory field, delete as applicable)

Name: \_\_\_\_\_; Date of birth: \_\_\_\_\_

Gender: ☐ M ☐ F Ethnicity and geographical origin: - from index patient

\_\_\_\_\_ ; - from the mother \_\_\_\_\_, - from the

father \_\_\_\_\_ Consultancy Referral Number: \_\_\_\_\_

Identification Label / Barcode

Place the identification label here

**SPECIMEN SOURCE** (obligatory field)

☐ Whole blood

☐ DNA

☐ Saliva

**URGENT** ☐

Reason: \_\_\_\_\_

**PHYSICIAN INFORMATION** (obligatory field)

Physician \_\_\_\_\_

Address \_\_\_\_\_

Institution: \_\_\_\_\_ Department: \_\_\_\_\_

Telephone: \_\_\_\_\_ Fax: \_\_\_\_\_ E-mail: \_\_\_\_\_

**MOLECULAR TEST REQUESTED** (obligatory field)

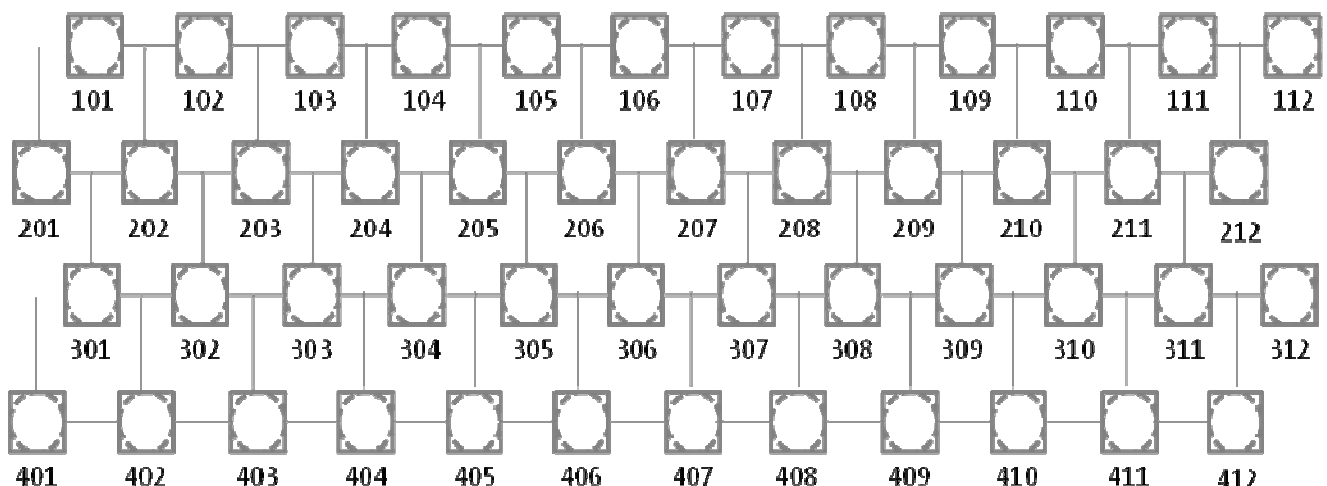
**Molecular pathologic markers of high risk and prevalence for familial hypercholesterolemia** ☐

Evaluation of genetic mutations of *LDLR* gene that cause familial hypercholesterolemia and that are related with 1) high levels of total and LDL cholesterol, 2) severe phenotype, 3) increased risk of premature cardiovascular disease and 4) higher frequency in the population. Evaluation of genetic alterations of *APOE* gene that are associated with increased risk of premature cardiovascular disease.

**PREVIOUS GENETIC CONSULTANCY:** Date \_\_\_\_/\_\_\_\_/\_\_\_\_; **AGE OF DIAGNOSTIC:** \_\_\_\_\_

**FAMILIAR INFORMATION**

Previously studied familial members: identification in genealogical tree. Point out the individual in the present study with an arrow (↗).





**MOLECULAR PATHOLOGIC MARKERS OF HIGH RISK AND  
PREVALENCE FOR FAMILIAL HYPERCHOLESTEROLEMIA**

Name: \_\_\_\_\_

Consultancy Referral Number: \_\_\_\_\_

Position in the tree	Name / Consultancy Referral Number	Clinical information and age of diagnostic

**CLINICAL INFORMATION: COMPLEMENTARY DIAGNOSTIC EXAMS**

By filling these fields you are contributing to improve your future patient diagnostics as we are developing tools to model clinical and genetic data regarding familiar hypercholesterolemia.

Clinical Information	Data
Date and age of diagnostic	_____(day) / _____(month) / _____(year), _____ years
<b>Total cholesterol</b> _____ pv tp mg/dl, _____ af tp mg/dl; <b>LDL-C</b> _____ pv tp mg/dl, _____ af tp mg/dl; <b>HDL</b> _____ pv tp mg/dl, _____ af tp mg/dl; <b>VLDL</b> _____ pv tp mg/dl, _____ af tp mg/dl, <b>non-HDL cholesterol</b> _____ pv tp mg/dl, _____ af tp mg/dl; <b>ApoA1</b> _____ mg/dl, <b>ApoB</b> _____ mg/dl (pv tp - previous therapeutics, af tp - after therapeutics )	
Personal history of cardiovascular diseases	Myocardial Infarction <input type="checkbox"/> _____ (y), Angina <input type="checkbox"/> _____ (y), STENT <input type="checkbox"/> _____ (y), Coronary Bypass <input type="checkbox"/> _____ (y), Aneurysms _____ (y), Carotid Artery Disease <input type="checkbox"/> _____ (A), Stroke <input type="checkbox"/> _____ (y) Premature peripheral arterial disease <input type="checkbox"/> _____ (y), Renovascular Hypertension <input type="checkbox"/> _____ (y)
Signs	Tendon xanthomas <input type="checkbox"/> , Xanthelasmas <input type="checkbox"/> , Arcus senilis <input type="checkbox"/> , Fat liver <input type="checkbox"/>
Associated diseases	Thyroid disease <input type="checkbox"/> , Liver disease <input type="checkbox"/> , Pancreatic disease <input type="checkbox"/> , Autoimmune disease <input type="checkbox"/> , Chronic kidney disease <input type="checkbox"/> , Arterial hypertension <input type="checkbox"/>
Family history	High cholesterol <input type="checkbox"/> , High LDL <input type="checkbox"/> , Premature cardiovascular disease (before 55 in a man and before 60 in a woman) <input type="checkbox"/> , sudden death <input type="checkbox"/>
Associated risk factors	Fast food <input type="checkbox"/> , Lack of physical activity <input type="checkbox"/> , Obesity <input type="checkbox"/> , Overweight <input type="checkbox"/> , Units of alcohol <sup>(1 unit = 1 glass)</sup> / week _____, Smoking <input type="checkbox"/> , nº cigarettes /day_____, nº packs /day _____, stop smoking at _____ years_____
Therapeutics	



**MOLECULAR PATHOLOGIC MARKERS OF HIGH RISK AND  
PREVALENCE FOR FAMILIAL HYPERCHOLESTEROLEMIA**

Name: \_\_\_\_\_

Consultancy Referral Number: \_\_\_\_\_

**ANNEX**

- ☐ Sample tubes labeled with index case / patient / familiar information
- ☐ Whole blood (preferable) (Date obtained: \_\_\_\_ / \_\_\_\_ / \_\_\_\_), Conditions: 4mL or 2 X 3mL in K<sub>2</sub>EDTA or K<sub>3</sub>EDTA collection tube
- ☐ DNA (Date obtained: \_\_\_\_ / \_\_\_\_ / \_\_\_\_); Volume \_\_\_\_ µL; Concentration \_\_\_\_ µg/mL; Purification Method: \_\_\_\_\_; Conditions: minimum 300ng of 35ng/µL,
- ☐ Saliva (recommended kit: Oragene DNA collection kit Genotek)

**INFORMED CONSENT INFORMATION (IT IS MANDATORY TO BE SIGNED)**

I hereby authorize the collection of my/ my child's ..... [name] biological sample for the genetic test specified in this request. I declare that I have been informed about genetic testing features and that I understand the benefits and limitations of the cardiovascular genetic test regarding genetic analysis of familial hypercholesterolemia for which I am giving permission.

I give permission for the anonymously processing of the obtained digital data: yes ☐ no ☐

I give permission for the biological specimen and clinical information to be anonymously used in research studies: yes ☐ no ☐

**Place and Date** \_\_\_\_\_; \_\_\_\_ / \_\_\_\_ / 20\_\_\_\_ **Signature** \_\_\_\_\_

**Physician signature** \_\_\_\_\_