



**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)

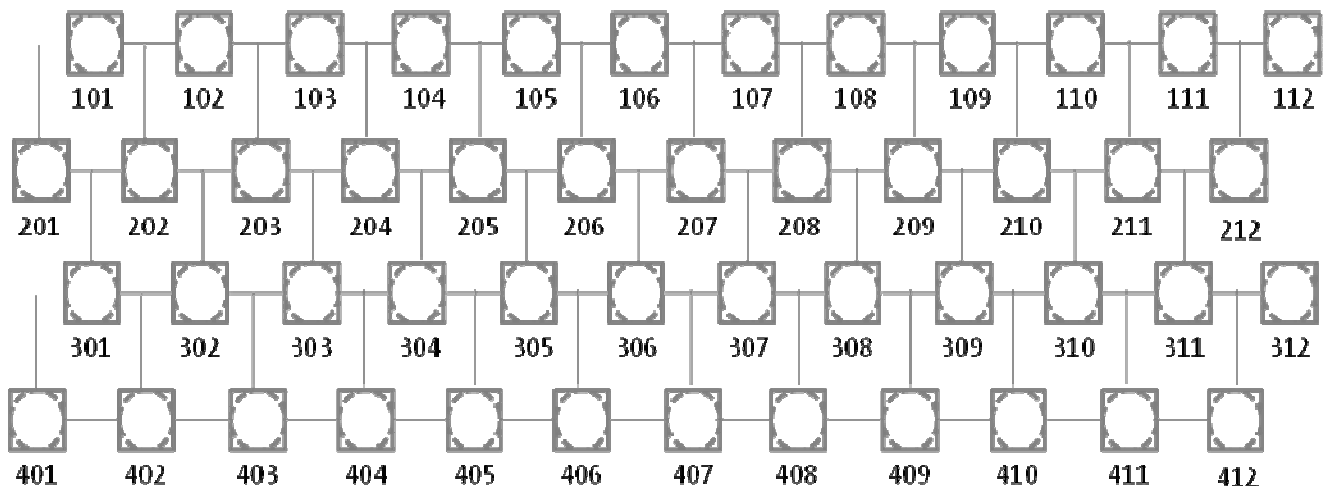
Molecular pathologic markers of high risk and prevalence for familial hypercholesterolemia ☐

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





PREVALENCE FOR FAMILIAL HYPERCHOLESTEROLEMIA

Name: _____

Consultancy Referral Number: _____

| Position in the tree | Name / Consultancy Referral Number | Clinical information and age of diagnostic |
|----------------------|------------------------------------|--|
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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 Arterial Hypertension risk factors.

| Clinical Information | Data |
|---|--|
| Date and age of diagnostic | ____ (day) / ____ (month) / ____ (year), ____ years |
| Total cholesterol _____ pv tp mg/dl, _____ af tp mg/dl; LDL-C _____ pv tp mg/dl, _____ af tp mg/dl; HDL _____ pv tp mg/dl, _____ af tp mg/dl; VLDL _____ pv tp mg/dl, _____ af tp mg/dl, non-HDL cholesterol _____ pv tp mg/dl, _____ af tp mg/dl; ApoA1 _____ mg/dl, ApoB _____ 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 ^(1 unit = 1 glass) / week _____, Smoking <input type="checkbox"/> , nº cigarettes /day____, nº packs /day _____, stop smoking at ____ years____ |
| Therapeutics | |



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 K₂EDTA or K₃EDTA 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 hypertrophic cardiomyopathy 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 _____