Molecular Diagnostic Laboratory

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Cardiovascular Genetics Test Requisition Form

Familial dyslipidemia

A- Sample Information

Date : yyyy - mm - dd

Lab reference number : 

Collect sample by (Name) : 

N.B.: DNA samples extracted from Formalin-fixed, paraffin-embedded (FFPE) tissues will not be accepted.

B- Reason for this request (Please check all that apply) 

☐ Genetics test request (Please complete section E or F) 

☐ Patient’s consent was obtained (A copy of the signed consent form must be provided) 

☐ DNA Banking 

☐ Additional testing (Please complete section E or F) 

C- Known or suspected diagnostic 

(Please check all that apply) 

☐ Familial hypercholesterolemia (FH) 

☐ Familial hyperalphalipoproteinemia 

☐ Familial hyperchylomicronemia 

☐ Other : 

☐ Clinical data: 

☐ Pharmacotherapy : 

☐ LDL-C (mmol/L) : 

☐ Before treatment : 

☐ After treatment initiation : 

☐ Tg (mmol/L) : 

D- Patient’s Phenotype

☐ Family History:

☐ Please also complete the HF Dutch Lipid Clinic Network Diagnostic Criteria grid (back).

E- Targeted variant testing only 

☐ Required for family member 

Specific Variant: 

☐ Gene 

☐ Transcript 

☐ Nucleic Change 

☐ Protein Change 

☐ Please attach a copy of a family member’s report.

F- Dyslipidemia Panel Test (Please check all that apply) 

☐ Required for proband 

☐ Please note that sequential analyzes are performed. If no clinically significant variant is identified within the first-line analysis, the second-line panels will automatically be completed.

Familial dyslipidemia

☐ Familial hypercholesterolemia (FH) 

☐ Familial hyperalphalipoproteinemia 

☐ Familial hyperchylomicronemia 

☐ Sitosterolemia 

☐ Addicional gene(s) : 

☐ LDLR (Copy Number Variations - CNV) 

☐ CETP 

☐ LPL 

☐ ABCG5, ABCG8 

☐ APOB, LDLR, LDLRAP1, PCSK9 

☐ - 

☐ APOA5, APOC2, GPHBP1, LMF1 

☐ - 

Addressograph
Dutch Lipid Clinic Network diagnostic criteria for familial hypercholesterolemia

Please circle the criteria that apply

<table>
<thead>
<tr>
<th>Criteria</th>
<th>Points</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Family history</strong></td>
<td></td>
</tr>
<tr>
<td>i. First-degree relative with premature coronary heart disease (M &lt; 55 years; F &lt; 60 years)</td>
<td>1</td>
</tr>
<tr>
<td>OR</td>
<td></td>
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<tr>
<td>ii. First-degree relative with known LDL-C level &gt; 95th percentile</td>
<td></td>
</tr>
<tr>
<td>iii. First-degree relative with tendinous xanthoma and / or arcus cornealis</td>
<td>2</td>
</tr>
<tr>
<td>OR</td>
<td></td>
</tr>
<tr>
<td>iv. Children aged &lt; 18 y/o with LDL-C level &gt; 95th percentile</td>
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<tr>
<td><strong>Clinical history</strong></td>
<td></td>
</tr>
<tr>
<td>i. Patient with premature coronary heart disease (M &lt; 55 years; F &lt; 60 years)</td>
<td>2</td>
</tr>
<tr>
<td>ii. Patient with premature cerebral or peripheral vascular disease</td>
<td>1</td>
</tr>
<tr>
<td><strong>Physical examination</strong></td>
<td></td>
</tr>
<tr>
<td>i. Tendinous xanthoma</td>
<td>6</td>
</tr>
<tr>
<td>ii. Arcus cornealis &lt; 45 years</td>
<td>4</td>
</tr>
<tr>
<td><strong>LDL cholesterol level</strong></td>
<td></td>
</tr>
<tr>
<td>i. LDL-C level &gt; 8,5 mmol/L</td>
<td>8</td>
</tr>
<tr>
<td>ii. LDL-C level 6,5 - 8,4 mmol/L</td>
<td>5</td>
</tr>
<tr>
<td>iii. LDL-C level 5,0 - 6,4 mmol/L</td>
<td>3</td>
</tr>
<tr>
<td>iv. LDL-C level 4,0 - 4,9 mmol/L</td>
<td>1</td>
</tr>
<tr>
<td><strong>DNA analysis</strong></td>
<td></td>
</tr>
<tr>
<td>Causative variant in LDLR, APOB or PCSK9 genes</td>
<td>8</td>
</tr>
<tr>
<td><strong>TOTAL SCORE :</strong></td>
<td></td>
</tr>
</tbody>
</table>

Diagnosis (based on the total number of points obtained):

- Definite familial hypercholesterolemia: > 8 points
- Probable familial hypercholesterolemia: 6-8 points
- Possible familial hypercholesterolemia: 3-5 points
- Unlikely familial hypercholesterolemia: 0-2 points

**Indication for genetic testing :**

**Heterozygous FH**
- Patient with a score > 5 points
- Patient with xanthoma, high cholesterol and family history of coronary heart disease
- First-, second-, and third-degree relative of a proband known with a causative variant in one of the genes associated with familial hypercholesterolemia (Section E-Targeted variant testing only)

**Homozygous FH**
- Patient in whom both parents are known with a causative variant in one of the genes associated with familial hypercholesterolemia
  - OR
- Untreated patient with LDL-C level > 12,0 mmol/L and treated patient with LDL-C level > 8,0 mmol/L
  - AND
- Cutaneous or tendinous xanthoma < 10 years
  - OR
- Both parents with elevated LDL-C levels reflecting heterozygous HF