**FAMILIAL HYPERCHOLESTEROLAEMIA GENE TEST REQUEST**

**Laboratory Genetics, Polwarth Building, Foresterhill, Aberdeen AB25 2ZD**

Email gram.molgen@nhs.scot Tel 01224 553893

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| --- | --- | --- | --- | --- | --- |
| **Essential Patient Demographics (Patient label can be used)** | | | | | |
| Forename: | | Surname: | | Date of Birth: | |
| CHI No.: | | Pedigree no. (if relevant): | | Male / Female (Circle as appropriate) | |
| Address (**must** include postcode): | | | | | |
|  | | | | | **Postcode:** |
| **Essential Referrer Details** | | | | | |
| Referring Clinician(s): | | Department & Address for report: | | | |
| Email address (generic departmental where possible): | | | | | |
| **Essential Sample Information** | | | | | |
| Sample Type: | Date Taken: | | Time Taken: | | |
| High Risk: YES / NO  If yes, please state risk \_\_\_\_\_\_\_\_\_\_\_\_\_\_  ***Notify lab in advance if high risk*** | Urgent analysis required: YES / NO | | DNAStorage only: YES / NO | | |
| **INDEX CASES:** see [Diagnosing familial hypercholesterolaemia (guidelines) | Right Decisions](https://rightdecisions.scot.nhs.uk/tam-treatments-and-medicines-nhs-highland/adult-therapeutic-guidelines/lipids/diagnosing-familial-hypercholesterolaemia-guidelines/)  **Indicate how patient meets FH Wales Criteria:** <https://fhwalescriteria.co.uk/assistant>  Choose the highest relevant score in each box and add together.  **Score of 6 or above is eligible for genetic testing. Score of 5 or less not eligible unless exceptional circumstances.** | | | | | |
| |  |  |  | | --- | --- | --- | | 1 | **Family history ie 1st or 2nd degree relative with**  CHD aged <60 yrs  CHD aged <45 yrs  Adult: LDL cholesterol > 4.9 mmol/L (or total cholesterol > 7.5 mmol/L)  Age <18 yrs: LDL cholesterol > 4.0 mmol/L (or total cholesterol > 6.7 mmol/L) | 1  2  1  2 | | 2 | **Physical examination**  Tendon xanthomata (in patient or 1st /2nd degree relative)  Premature corneal arcus (<50 yrs) | 6  4 | | 3 | **Patient clinical history**  Patient with premature CHD (<45yrs)  Patient with premature CHD (<50yrs)  Patient with premature CHD (<60yrs)  Patient with premature (<60yrs) strokes and/or peripheral vascular disease | 4  3  2  1 | | 4 | **Patient untreated or corrected LDL-cholesterol** (<https://fhwalescriteria.co.uk/ldl_estimator>)  Consider age of patient. Secondary causes should be excluded prior to genetic testing.  LDL-cholesterol 8.5 mmol/L or higher  LDL-cholesterol 6.5 to 8.4 mmol/L  LDL-cholesterol 5.0 to 6.4 mmol/L  LDL-cholesterol 4.0 to 4.9 mmol/L | 8  5  3  1 | | 5 | **Patient fasting triglycerides**  Triglycerides 2.5 to 3.4 mmol/L  Triglycerides 3.5 to 4.9 mmol/L  Triglycerides 5.0 mmol/L or higher | -2  -3  -4 | |  | **Choose ONE score only per category**  **(Score ≥6 eligible for genetic testing) Total** |  | | | | | | |
| **Additional details if applicable** | | | | | |
| **Discussed with Clinical Genetics or Lipidologist?** Y / N / NA  If yes name of contact: | | | | | |
| **CASCADE TESTING OF RELATIVES: complete where family variant has already been detected** | | | | | |
| Pedigree reference no.:  Name of relative with known variant:  Variant details including lab. report reference:  ***If index variant found in another lab please supply a copy of the report where possible*** | | | | | |
| ***CONSENT: It is the responsibility of the referring clinician to obtain informed consent from the patient / carer for the test and for the sample to be stored for future diagnostic testing.***  ***Referring Clinician Signature: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Print name: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Date: \_\_\_\_\_\_\_\_\_\_\_\_\_*** | | | | | |