Elucigene FH20 and LIPOchip for FH - data extraction form

Reviewer ID: Date:

Administration details

Study ID: Publication status:

Other papers this study may link with:

Aim of the study

Test(s) reported

	Index cases	Cascade testing
Elucigene	☐	☐
LIPOchip	☐	☐
LDL-C	☐	☐
Targeted gene sequencing		☐
*CGA	☐	☐

* includes DNA sequence analysis+ test for deletion/duplication+ analysis of APOB p.Arg3527Gln and PCSK9 p.Asp374Tyr using various techniques.

Outcomes reported

Diagnostic accuracy ☐ Mutation detection rate ☐ Clinical effectiveness ☐

Study details

Cross-sectional comparative ☐ RCT ☐ Case control study ☐

Cross-sectional single test ☐ Other, please specify: ☐

Multicentre study? Yes ☐ No ☐ If Yes, number of centres:

Consecutive recruitment? Yes ☐ No ☐ Not stated ☐

Country:

Study dates:

Length of follow up:

Source of funding:

Inclusion criteria:

Exclusion criteria:

Baseline characteristics of participants

Adults ☐ Children ☐

Criteria used for clinical diagnosis:

Simon Broome ☐

Dutch ☐

Medped ☐

Other ☐ specify the LDLC cut offs used and definition

Type of FH:

Possible ☐ Unclassified FH ☐

Definite ☐ Not stated ☐

Heterozygous FH ☐ Homozygous FH ☐

Diagnosis of the Index cases confirmed by:

Clinical test ☐ Genetic test ☐

Who perform the clinical diagnosis?

Number of participant/sample

give detail of each type of FH if reported	All	Index cases	Relatives
Eligible			

Enrolled			
Analysed			
Received index test(s)			
Received comparator test(s) for index cases			
Received cascade test(s) 1st degree relatives 2nd degree relatives 3rd degree relatives			
Received comparator test(s) 1st degree relatives 2nd degree relatives 3rd degree relatives			
Age (mean/ median, SD, range)			
Receiving treatment for hyper- cholestorelaemia (specify treatment)			
Ethnicity			
Gender	**M:** **F:**	**M:** **F:**	**M:** **F:**
Tendon xanthomas			
Coronary Heart Disease			

Intervention tests

Elucigene FH20 (Tepnel molecular diagnostics)

If not FH20 which version and how many mutations was it designed to detect?

Gel-based analysis ☐ Fluorescent analysis: ☐

Who carried out the test?

Where was the test undertaken?

Time taken to obtain test results:

Additional information on the test:

LIPOchip (Progenika Biopharma)

If not version 10 which version and how many mutations was it designed to detect?

Samples processed at: LIPOchip laboratory ☐ Other ☐ If other, please give details:

Methodology used:

DNA array ☐

Analysis for large gene re-arrangements ☐

Automated sequencing of the LDLR ☐

Who carried out the test?

Where was the test undertaken?

Time taken to obtain test results:

Additional information on the test:

Comparator tests

CGA (as defined on page 5 of the protocol)

CGA should include following:

☐ DNA sequence analysis of the promoter, all exons, the exon/intron boundaries and into 3' untranslated region of the LDLR gene

Manufacturer and any other technical characteristics of the test:

☐ MLPA for each exon and the promoter region of the LDLR gene to detect deletions and duplications

Manufacturer and any other technical characteristics of the test:

☐ Analysis for the common APOB p.Arg3527Gin and PCSK9 p.Asp374Tyr gene mutations

Manufacturer and any other technical characteristics of the test:

Who carried out the test?

Where was the test undertaken?

Time taken to obtain test results:

Additional information on the test:

LDL-C concentration

Estimated from a fasting blood sample using the Friedwald equation? Yes ☐ No ☐

If No please specify method used:

For cascade test please specify age and gender specific LDL-C cut offs:

No. of times LDL-C was measured? Once ☐ Twice ☐ Not stated ☐

Criteria used to define a positive test result:

Who carried out the test?

Where was the test undertaken?

Time taken to obtain test results:

Additional information on the test:

If targeted gene sequencing of relatives was undertaken, please give details:

Reference standard test

Was there a reference standard test that consisted of either of the followings?

CGA in combination with Simon Broome criteria ☐

CGA only ☐

Simon Broome only ☐

Results for Index cases

1. Genetic test **……..**

	LDLR	*APOB*	*PCSK9*	*MLPA*	*sequencing*	*Total*
Number of participants						
Number of samples analysed						
n/N (%) with mutation detected						
n/N (%) with no mutation detected						

2. Genetic test **……..**

Number of participants						
Number of samples analysed						
n/N (%) with mutation detected						
n/N (%) with no mutation detected						

3. LDL-C as a clinical test						
Number of participants						
Number of samples analysed						
Number of FH diagnosed						
Number of FH not diagnosed						
Results for Cascade test						
Specify the genetic test……..						
Number of participants (Index cases)						
Number of samples analysed						
Number of families tested						
n/N (%) with mutation detected						
n/N (%) with no mutation detected						
LDL-C age and sex specific test						
Number of participants (Index cases)						
Number of samples analysed						
Number of families tested						
Number of FH diagnosed						
Number of FH not diagnosed						

Record data on each level of analysis containing 2x2 tables of true and false positives and negatives for

Test accuracy of genetic test (Elucigene/Lipochip) vs genetic test (CGA)

Test accuracy of genetic test (Elucigene/Lipochip) vs clinical test (LDL-C-SB criteria)

Subgroup analysis reported (e.g. ethnicity)? Yes ☐ No ☐

If Yes please give details:

Give details of any clinical effectiveness outcomes reported, e.g. cholesterol levels following treatment, CHD events etc or probability of true FH:

Give details of any information reported on acceptability and/or interpretability of the tests

Additional information: