

Data Collection Form for FCS/MCS Service Assessment/Quality Improvement project 2020 UK (V5)

Centre who have genetically confirmed FCS patients (or if the patient had genetic testing but FCS was not confirmed ie no mutation or one mutation in FCS related genes) are invited to take part.

Please complete one form for each patient to be included in the project.

We are collecting anonymised data of the following cohorts from different centres/ countries:

1. Confirmed Familial Chylomicronaemia Syndrome (FCS) with genetic testing (homozygous, compound and double heterozygous)
2. Genetic testing showed one pathogenic mutation/variance only (heterozygous)
3. Severe hypertriglyceridaemia with FCS phenotype caused by antibodies (eg LPL, apoC2,...) or any other potential novel mechanism.
4. Data for patients who are phenotypically FCS but have novel mutations (heterozygous or homozygous) for example CREB3L3.
5. Data for patients who are phenotypically FCS but genetic test did not show a pathogenic mutation/variance
6. Patients diagnosed as MCS (genetic test performed but no mutation or no genetic testing performed).

Please complete the following:

- Principal Investigator name, title and qualifications
- Name of the centre and full affiliation
- Contact details
 - Email
 - Tel
 - Fax
- Country
- Patient ID (please use 001,002 etc) _____
- Gender
- Current age
- Consanguineous marriage (parents are first or second cousin...)
 - If yes, please provide details
- Ethnicity
- Age at formal diagnosis
 - Age when symptoms started
 - Years between symptoms and diagnosis
- Body weight (Kg) at diagnosis or first available
- Height (meter) at diagnosis or first available
- BMI

Data Collection Form for FCS/MCS Service Assessment/Quality Improvement project 2020 UK (V5)

- Genetic testing result
 - Result is consistent with FCS (homozygous, compound and double heterozygous).
Details of mutation(s):
 - One mutation only, details of mutation:
 - No pathogenic mutation/variance found:
 - A novel mutation/variance in a gene other than the established 5 genes (LPL, APOC2, GPIIIBPI, APOA5, LMF1).
If yes give detail:
- Type 3 hyperlipoproteinaemia excluded by apoE genotyping? Yes No
if yes give details of apoE genotype result
- Type 2 diabetes
 - Yes give age at diagnosis of diabetes
 - No
- Cardiovascular disease and state age of diagnosis
 - CHD
 - Stroke/cerebrovascular disease
 - Peripheral arterial disease

If yes to any please give age at diagnosis in years _____
- Acute pancreatitis (AP)
 - Yes give age when first AP occurred
 - More than one AP
 - Number of AP episodes if known
 - No AP
- Lowest serum triglycerides ever recorded _____
- Peak serum triglycerides ever recorded _____
- Fasting triglycerides > 10 mmol/L on 3 consecutive occasions
 - Yes
 - no
- Fasting triglycerides >20 mmol/L at least once
 - Yes
 - no
- Onset of symptoms
 - at age <10 years
 - <20 years
 - <40 years
- Secondary causes for hypertriglyceridemia Yes No
If yes please state _____
- History of unexplained recurrent abdominal pain Yes No
- Evidence of peripheral neuropathy
 - Yes, please give details
 - No

Data Collection Form for FCS/MCS Service Assessment/Quality Improvement project 2020 UK (V5)

- Triglycerides not responding to lipid lowering medications (less than 20% reduction in TG)
 - Yes
 - no
- History of Familial Combined Hyperlipidaemia
 - Yes
 - No
- Current treatments for severe hypertriglyceridaemia/lipid lowering therapy
 - 1.
 - 2.
 - 3.
 - 4.
 - 5.
 - 6.

- Current other treatments
 - 1.
 - 2.
 - 3.
 - 4.
 - 5.
 - 6.
 - 7.
 - 8.

- Is this patient on lipoprotein apheresis (LA) or plasma exchange (PE) (if yes give details of the method)?
 - Yes regular apheresis Type of treatment (PE or LA and frequency)

 - Occasional LA or PE to abort acute pancreatitis (please give details type and frequency) _____
 - Only during pregnancy (LA or PE and frequency) _____
 - Never received apheresis
 - Other (give details) _____

- Pregnancy history (females)
 - How many pregnancies
 - Acute pancreatitis during pregnancy
 - Yes
 - No
 - Not known
 - How many miscarriages _____
 - Related to acute pancreatitis?
 - Yes

Data Collection Form for FCS/MCS Service Assessment/Quality Improvement project 2020 UK (V5)

- No
 - Not known
- Has the patient avoided pregnancy because of possible complication?
 - Yes
 - No
 - Not known
- Ever been advised by health care professional to avoid pregnancy?
 - Yes
 - No
 - Not known
- Do you use the diagnostic scoring system for FCS proposed by Moulin et al? See below [https://www.atherosclerosis-journal.com/article/S0021-9150\(18\)31126-2/fulltext](https://www.atherosclerosis-journal.com/article/S0021-9150(18)31126-2/fulltext)
 - For diagnosis Yes No
 - For patient selection for genetic testing? Yes No
 - Other, please state:
 - If you use the FCS score , please indicate the score _____
- Do you have a dedicated dietitian in your clinic/lipid clinic (delete as appropriate)?
 - Yes
 - No
- Do you have a Lipid specialist nurse? Yes No
- Any other medical conditions?
- Any other relevant data/notes

Please send completed form(s) to:

Dr See Kwok, Senior Research Fellow,
c/o Cardiovascular Trials Unit, Cardiovascular Research Clinical Trial Management
Office,
North Road, Manchester Royal Infirmary, Oxford Road, Manchester M13 9WL, UK
Or by email to: see.kwok@mft.nhs.uk

**Moulin et al diagnostic scoring system for FCS. Moulin *et al.*
Atherosclerosis; 275; 265-272**

Data Collection Form for FCS/MCS Service Assessment/Quality Improvement project 2020 UK (V5)

Recruitment phase

Severe primary HTG (fasting TGs >10 mmol/L or 885 mg/dL)

Patient pre-selection in non-acute setting

1. Fasting TGs >10 mmol/L for 3 consecutive blood analyses (+5)^a
 - Fasting TGs >20 mmol/L at least once (+1)
2. Previous TGs <2 mmol/L (-5)
3. No secondary factor^b (except pregnancy^c and ethinylestradiol) (+2)
4. History of pancreatitis (+1)
5. Unexplained recurrent abdominal pain (+1)
6. No history of familial combined hyperlipidaemia (+1)
7. No response (TG decrease <20%) to hypolipidaemic treatment (+1)
8. Onset of symptoms at age:
 - <40 years (+1)
 - <20 years (+2)
 - <10 years (+3)

FCS score:
≥10: FCS very likely
≤9: FCS unlikely
≤8: FCS very unlikely