Results: We found 367 samples from 288 patients. The majority (233/367, 64%) originated in general practice and most were female (222, 60%). Mean age of tested patients was 49±15 years (F:M 51:46, p<0.01). No medical history data were available. In 2018 we invited all test request originators to refer their patients to a hospital clinic for FH assessment including genetic testing. To date, 69 of 288 patients have been seen in Metabolic and Lipid clinics between two sites, 24 are awaiting assessment, 2 have consented to genetic testing only, 14 had diagnostic mutations established and in 23 hypercholesterolaemia was secondary to other conditions.
Conclusions: While this strategy will not identify all FH patients, we believe it is a proactive first step in using available laboratory data to identify hitherto unknown FH patients and alert clinicians to this underdiagnosed condition. It is planned to extend this strategy to other hospitals in the Dublin region.

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EAS19-1016.
COMPLETE REGRESSION OF XANTHOMATA IN 2 YEARS OLD GIRL WITH HOMOZYGOUS BETA-SITOSTEROLEMIA TREATED WITH EZETIMIBE 5 MG/DAY

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Background and Aims: Beta-sitosterolemia (BS) is a rare autosomal-recessive monogenic lipid disorder, with elevation of plant sterols due to genetic defect of transporters ABCG5 or ABCG8. Phenotypically these patients demonstrate features of FH and respond well to diet and ezetimibe therapy.
Methods: Initial examination of patient M.I, girl 2 years old, revealed high level of LDL (15 mmol/l), low level of heamoglobin and extensive yellow -orange planar xanthomatosis of the buttocks, wrists, knees and coccyx area. Secondary causes of hyperlipidemia were ruled out.The diagnosis of BS in girl was confirmed by DNA test (mutation in ABCG8 transporter chr2:44102511T>C, rs769576789). Both parents were normolipideamic, but heterozygous to BS by DNA test.
Results: Treatment with ezetimibe 5 mg a day was started in November 2016, since when it has been continued without side-effects for 6 months without side effects. Total and LDL-C levels averaged as 5.1 mmol/l (198 mg/dl) and 3.32 mmol/l (129 mg/dl), subsequently. Hemoglobin level increased from 98 to 123 g/l. Latest clinical chemistry results was normal (total cholesterol 131 mg/dl, trigs- 31 mg/dl, AST-16.6 IU/l, ALT-29.4IU/l. The xanthomata at all sites (both knees, coccyx area, extension area of wrists showed demonstrable early regression in 2 months and complete resolution after 6 months of ezetimibe 5mg/day.
Conclusions: We demonstrated the first case of rapid complete regression of xanthomata in 2 years old girl with homozygous BS treated with ezetimibe 5 mg/day over 8 months. Further long-term follow up is needed to assess efficacy and safety this treatment modality in small children with BS.

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EAS19-1030.
FAMILIAL HYPERCHOLESTEROLAEMIA IN MALAYSIAN COMMUNITY IS MORE COMMON THAN THAT IN MOST EUROPEAN COUNTRIES

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Background and Aims: Familial hypercholesterolaemia (FH) is the most common autosomal dorminantly inherited metabolic disorder causing severe elevation of low-density lipoprotein cholesterol (LDL-C), leading to premature coronary artery disease (CAD). Furthermore, the incidence of premature CAD in Malaysia is increasing with the average age of CAD onset being lower compared to neighbouring countries and western population. Worldwide, the estimated prevalence of FH is approximately 1:200-1:500. However, the prevalence of FH in Malaysia is unclear.
Methods: In this study, 4821 Malaysians (males:1771, females:3040, age range:18 – 75 years) were recruited from Community Health Screening Programmes organised throughout all states in Malaysia to investigate the prevalence of FH. Personal and family data, medical history and blood samples were obtained from all participants. Physical examination was conducted on site. Serum was obtained from the blood samples for fasting lipid profile analysis. FH patients were clinically diagnosed using Dutch Lipid Clinic Network (DLCN) Criteria, using LDL-c threshold of ≥4.9 mmol/L.
Results: From 4821 participants, 440 participants (9.1%) had severe hypercholesterolaemia (LDL ≥4.9 mmol/L). The number of participants with DLCN categories of Unlikely, Possible, Probable and Definite FH were 37, 352, 38 and 13, respectively.
Conclusions: Based on the consensus of accounting Probable and Definite FH as "Potential FH", the prevalence of FH in this study cohort was 1:95, which is more common than most that have been reported in European countries. A more rigorous community and hospital-based screening for FH has to be conducted in future where early FH detection may potentially reduce the incidence of premature CAD in Malaysia.

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EAS19-1058.
ASSESSMENT OF THE PREVALENCE OF FAMILIAL HYPERCHOLESTEROLEMIA IN PATIENTS WITH PRIMARY HYPERLIPIDEMIA IN MOSCOW LIPID CLINIC

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Background and Aims: To estimate the prevalence of familial hypercholesterolemia (FH) in patients (pts) of different ages with primary hyperlipidemia from 1990 to 2017.
Methods: Analysis included 4379 pts (46% men) with primary hyperlipideamia, who was examined in lipid clinic from 1990 to 2017. All pts were Caucasian, aged from 1 year to 85. Cascade screening included lipid and / or genetic testing of relatives. Patients were studied by calculation of FH probability according to Dutch Lipid Clinic Network (DLCN) and the Simon Broome (SB) criteria.
Results: We detected a lower prevalence of FH in patients with primary hyperlipidemia aged 1–25 (1,2%-5,9%). However, study showed variation in FH frequency with age, with an increase in prevalence that peaked between ages 45 and 54 (8,3%) and declining thereafter. The prevalence of FH in patients of different ages with primary hyperlipidemia was 7.63% which corresponds to a frequency of 1 in 13 individuals.
Conclusions: Our findings may be explained by insufficient dyslipidaemia screening in children and adolescents. Screening programmes (cascade screening) are necessary to increase the number of cases identified and treated. Despite the ongoing work on primary prevention, it is advisable to recommend sending such patients to specialized clinics for more qualified consultations.

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EAS19-0781.
CHILD WITH FAMILIAL HYPERCHOLESTEROLEMIA PHENOTYPE

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