UK National Screening Committee

Screening for Familial Hypercholesterolaemia in Children

12 February 2016

Aim

1. To ask the UK National Screening Committee (UK NSC) to make a recommendation, based upon the evidence presented in this document, on whether screening for familial hypercholesterolaemia (FH) in children meets the UK NSC criteria for the introduction of a population screening programme.

   This document provides background on screening for FH in children.

Current recommendation and background

2. The current recommendation is that a systematic population screening programme for FH in children is not recommended.

3. The UK NSC has not published a formal review on *childhood* FH screening, but the UK NSC's adult FH screening recommendation for cascade testing would be applicable to children. The most recent UK NSC external review of familial hypercholesterolaemia in adults, conducted in 2011, concluded that:

   "Universal screening for FH is not cost-effective and therefore a universal screening programme is not recommended. Best evidence currently supports cascade testing; tracing family members to identify affected relatives of known FH patients. However the NHS Health Check programme will also be testing all adults for cholesterol levels and will inevitably detect more people with FH which will complement cascade testing. It is doubtful whether existing lipid clinics could cope with the extra workload without investment."

4. This is consistent with the current NICE recommendation for cascade testing of relatives of FH cases.

5. The 2011 UK NSC evidence review was informed by previous modelling undertaken as part of a 2000 HTA, which concluded that cascade testing following identification of cases was the most cost effective strategy in adults. Universal population screening was the least cost

effective. However, this HTA did find that universal screening may be cost effective when targeted at young people (16 year olds).

6. The current review therefore aimed to review the evidence for universal screening for FH in childhood or adolescence.

Review

7. This condition is being reviewed as part of the UK NSC's three year review cycle (http://legacy.screening.nhs.uk/familialhypercholesterolaemia-child) and has been undertaken by Bazian Ltd. The review focuses on:

    i. the test
    ii. evidence looking at the effectiveness of a screening programme in reducing mortality and morbidity
    iii. acceptability of screening and treatment to patients, families and clinicians
    iv. cost-effectiveness of a universal screening programme in comparison with a cascade screening programme.

8. The conclusion of this review is that universally screening for FH in children should not be recommended. The key reasons to support this conclusion are:

    a. No studies were identified that have examined the performance of universal screening in practice. **Criterion 14 not met**

        One systematic review of case-control studies aimed to determine a) the age and b) the serum cholesterol concentration that would give the best discrimination between people with and without FH. This review suggested 1-9 years as the optimal age category.

        A UK prospective study due for completion this year is currently evaluating universal child (-parent) FH screening at the time of routine child immunisation at 1-2 years using blood spots.

    b. No studies were identified that assessed whether child screening (either universal or cascade testing) reduces morbidity or mortality from FH. **Criterion 13 not met**

    c. There remain many unanswered questions relating to the ethics and acceptability of universal screening at 1-2 years

d. No studies were identified which assessed the cost-effectiveness of universal screening in childhood. The HTA which informed the previous review suggested that universal screening at 16 years would be cost effective; no further studies were identified which evaluated the cost effectiveness of child screening at any age. **Criterion 16 not met**

Consultation

9. A three month consultation was hosted on the UK NSC website. Direct emails were sent to stakeholders of whom 26 organisations and individuals were contacted directly. **Annex A**

10. Six Responses were received from the following stakeholders: HEART UK, Paediatric Familial Hypercholesterolaemia service in Wales, Genetic Alliance UK, a joint response from the British Inherited Metabolic Diseases group (BIMDG) and UK Paediatric Familial Hypercholesterolaemia Register, a response from Professor David Wald on behalf of the British Cardiovascular Society and a response submitted by Dr Eurodiki Dragari **Annex B**

11. Overall the small number of responses acknowledged the absence of evidence relating to the key criteria discussed in the review. However respondents emphasised that screening in the early years should not be ruled out. The responses noted that the ongoing study of blood spot screening in 1 – 2 year olds (Wald, D. Study of child-parent screening for familial hypercholesterolaemia) may provide a point of departure for consideration of further evidence requirements in terms of the types of study and the outcomes.

Recommendation

12. The committee is asked to approve the following recommendation:

    A systematic population screening programme for screening the screening of familial hypercholesterolaemia in children is not recommended.

This is because:

- A suitable and feasible strategy for general population screening has not yet been identified.
- No studies have been identified that assessed whether child screening reduces illness or death from FH.

- There remain many unanswered questions about the ethics and acceptability of universal screening at 1-2 years, relating to the management of screen-detected cases
- no cost-effectiveness studies have been published on screening all children between 1-2 years old.

Based upon the 22 UK NSC criteria to recommend a population screening programme, Familial hypercholesterolaemia screening in children did not meet the following primary requisites:

	Criteria	**Met / Not met**
The Test		
5	There should be a simple, safe, precise and validated screening test.	Not Met ✗
The Screening Programme		
13	There should be evidence from high quality Randomised Controlled Trials that the screening programme is effective in reducing mortality or morbidity.	Not met ✗
14	There should be evidence that the complete screening programme (test, diagnostic procedures, treatment/intervention) is clinically, socially and ethically acceptable to health professionals and the public.	Not met ✗
16	The opportunity cost of the screening programme (including testing, diagnosis and treatment, administration, training and quality assurance) should be economically balanced in relation to expenditure on medical care as a whole (i.e. value for money). Assessment against this criteria should have regard to evidence from cost benefit and/or cost effectiveness analyses and have regard to the effective use of available resource.	Not met ✗

Annex A

List of organisations contacted:

1. AntiCoagulation Europe
2. British Cardiac Patients' Association
3. British Cardiovascular Society
4. British Heart Foundation
5. British Inherited Metabolic Disease Group
6. Cardiac Risk in The Young
7. Cardio & Vascular Coalition
8. The Cardiomyopathy Association
9. Children Living with Inherited Metabolic Diseases
10. Children's Heart Federation
11. Circulation Foundation
12. David Wald
13. Faculty of Public Health
14. Genetic Alliance UK
15. HEART UK
16. Institute of Child Health
17. MetBio
18. Royal College of General Practitioners
19. Royal College of Nursing
20. Royal College of Paediatrics and Child Health
21. Royal College of Physicians
22. Royal College of Physicians of Edinburgh
23. Royal Society for Public Health
24. Scottish Lipid Forum
25. UK Genetic Testing Network
26. Wolfson Institute of Preventive Medicine

UK National Screening Committee

Screening for Familial Hypercholesterolaemia in children- an evidence review

Annex B

1.

Name:	Simon Williams	**Email address:**	**xxxx xxxx**
Organisation (if appropriate):	HEART UK		
Role:	**Head of Communications & Policy**		
Do you consent to your name being published on the UK NSC website alongside your response? **Yes** ☒ **No** ☐			

Section and / or page number	**Text or issue to which comments relate**	
		HEART UK consulted with its FH Implementation Team and Medical, Scientific & Research Committee and has the following observations and comments: 1. HEART UK considers screening at 16 years of age too late to introduce beneficial lifestyle changes. Children of this age will need the infrastructure and psychological support if tested positive for FH, which can be very difficult at a crucial time for their education and advancement into adulthood. 2. HEART UK agrees that there is insufficient evidence yet on the impact of parent-child screening. 3. HEART UK considers that it may be premature to judge Universal Screening for FH in childhood as ineffective until the Wade study is completed in June 2016. We would accept the

		need for a larger pilot study for 5 to 10 years with all the necessary support for children, should the study return with a positive benefit. 4. Children from some families with CVD in 3rd and 4th decade of live and those with particularly high LDL-cholesterol may benefit from an introduction of statin therapy before age of 16.

2.

Name:	Dr P J Dale	**Email address:**	**xxxx xxxx**
Organisation (if appropriate):	Paediatric Familial Hypercholesterolaemia service in Wales		
Role:	**Consultant Paediatrician & Gastroenterologist,** **Lead for Paediatric FH Service in Wales**		
Do you consent to your name being published on the UK NSC website alongside your response? **Yes** ☒ **No** ☐			

Section and / or page number	**Text or issue to which comments relate**	
		Comments regarding consultation on familial hypercholesterolaemia screening in childhood document (draft 2, March 2015) Thank you for sending us the links for the screening for familial hypercholesterolaemia in childhood NSC document. We have reviewed this document in detail. As the paediatric consultant leading the paediatric FH service in Wales I am responding on behalf of our group. Dr Ian MacDowell, Chemical Pathologist, and colleague who jointly undertakes

		the paediatric clinic with me (and is Chair of the Wales FH Professional Advisory Group in Cardiff) and I have discussed this document in detail together with other members of our FH service including specialist nurses. We have comments which are both for and against the potential screening programme in childhood which we hope will be helpful for you. We fully recognise that screening children earlier in life than currently undertaken would potentially pick up a great number of paediatric patients with FH than currently takes place when screening through the cascade system. Picking up more patients with FH therefore would result in better population management of FH from childhood. On the negative side, if screening is undertaken early in childhood (for example in the first year of life) we would be recognising children who have been diagnosed with FH but in whom we would not be offering treatment with statins until the age of 10 years. We suspect that parents may find it difficult for their child to be given a diagnosis of FH but yet may not be started on medical treatment for potentially eight or nine years. The other concern raised by early diagnosis may be that any potentially over-strict dietary changes made by the family may result in restriction of some parts of the diet in both the children and family which may lead to other additional problems and potential deficiencies. I hope these comments are helpful for you. Yours sincerely

		Dr P J Dale Consultant Paediatrician & Gastroenterologist Lead for Paediatric FH Service in Wales

3.

Name:	Alastair Kent	**Email address:**	**xxxx xxxx**
Organisation (if appropriate):	Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 180 patient organisations. Our aim is to ensure that high quality services, information and support are provided to all who need them. We actively support research and innovation across the field of genetic medicine.		
Role:	**Director**		
Do you consent to your name being published on the UK NSC website alongside your response? **Yes** **No**			

Section and / or page number	Text or issue to which comments relate	
5	"This review searched the literature between January 2004 and January 2015 to answer key questions related to universal FH screening, specifically in children"	The current methodology used by the UKNSC when making decisions about whether the benefits of introducing a screening programme for a condition outweighs the risks places a premium on peer reviewed literature to the exclusion of all other forms of evidence.
		Additionally, relying solely on peer reviewed literature excludes the direct contribution of the patient voice to the process. While information from clinicians and patients may not be published, it represents the most recent and relevant information on a condition coming from those that either directly manage or are affected by the condition today. We note with concern that in this instance half of the literature referenced is more than five years out of date.

		Additionally, relying solely on peer reviewed literature excludes the direct contribution of the patient voice to the process. While information from clinicians and patients may not be published, it represents the most recent and relevant information on a condition coming from those that either directly manage or are affected by the condition today. We note with concern that in this instance half of the literature referenced is more than five years out of date. It is also unfortunate that the literature review is a year out of date, as a number of papers directly relevant to this NSC review have been published since January 2015. These include an article relating to the outcomes of the Slovenian universal childhood screening programme (Klančar G, Grošelj U, Kovač J, et al. Universal Screening for Familial Hypercholesterolemia in Children. J Am Coll Cardiol. 2015;66(11):1250–1257) which is referred to in the review. The review also acknowledges that the UK prospective study evaluating universal child(-parent) screening at the time of routine immunisation is due to report very shortly. While inclusion of these studies in the evidence review may not necessarily have changed the conclusions reached, they do address some of the unanswered questions which were identified, and may indicate that it would be appropriate to review this decision sooner than the standard three years.
5-6	“Whether universal child screening reduces morbidity and mortality associated with FH”	While we understand that this is a standard question asked as part of UK NSC’s reviews, framing the question in this way is inappropriate as it sets an unrealistically high evidence requirement. The report itself acknowledges that long term follow up of

		several decades is required to examine cardiovascular outcomes in children diagnosed with FH, and it is not clear how this information is likely to be generated within a reasonable time frame. Should sufficient evidence be generated to show that universal child screening improves shorter term objective measures of atherosclerosis development, it would be inappropriate to delay implementation of a screening plan until direct proof of reduced mortality had been obtained, as this would lead to significant numbers of unnecessary deaths. It is important that the criteria against which a proposed screening programme are assessed are realistic and relevant to the specific goals of the proposed screening. Adhering uncritically to a standard set of questions does not lead to balance and fairness, but rather the opposite.

Royal Free London NHS
NHS Foundation Trust

Royal Free Hospital

Consultation on Screening for Familial Hypercholesterolaemia in Children External Review Against programme appraisal criteria for the UK National Screening Committee (UK NSC)

Comments on behalf of British Inherited Metabolic Diseases Group (BIMDG) & UK Paediatric Familial Hypercholesterolaemia (FH) Register

Brief Summary of the review:
Bazian Ltd. on behalf of the UK National Screening Committee conducted this review. The review found that the evidence is currently insufficient to answer the key questions around universal screening of children for FH, including child-parent screening. This review searched the literature between January 2004 and January 2015 to answer key questions related to universal FH screening, specifically in children:

1. Whether there is a reliable universal screening test for FH in children and how it performs compared with cascade testing of relatives of index cases
2. Whether universal child screening reduces morbidity and mortality associated with FH
3. Whether universal child screening and subsequent treatment of screen-detected cases would be ethical and acceptable to professionals and the public
4. 4. Whether universal child screening would be cost effective compared with cascade testing

The review found no studies that examined how well a screening test covering all children performed in practice; there was some evidence suggesting the screening test performs best in children between 1-9 years. (Wald et al, a UK study currently evaluating FH screening being carried out at the same time as child vaccinations at 1-2 years, following which the parents of any children detected with FH are themselves tested. This may help identify additional people with the condition. Study completion date: June 2016) No studies were identified that assessed whether child screening reduces illness and death from FH.

The review concluded that it is unclear whether screening all 1-2 year old children at the same time, as vaccinations would be acceptable. The review highlighted concerns with this approach, as it commented that statin medication recommended for FH is not usually given to children under 10 years old, as per NICE guidelines. The review states that there are no studies assessing the balance between benefit and cost of screening all 1-2 year olds, or in children of any age.

The review describes the current UK NSC recommendation as not to carry out population screening for familial hypercholesterolaemia (FH). This follows the 2011 review of FH in adults that concluded that universal screening is not a cost-effective strategy. The current recommendation is cascade testing, where both child and adult relatives of index cases (probands) diagnosed with FH are screened. The review also mentions that modeling undertaken as part of a 2000 HTA suggested that universal screening of children aged 16 years could be more cost effective than cascade testing.

The review states that it found no studies that examined the performance of universal screening in practice. The child-parent screening study in the UK has not been completed; the ethics of this approach and management of screen-detected cases is not clear; no studies of cost effectiveness and clinical benefits of universal screening for FH; previous HTA study explored the evidence of screening aged 16 years but concluded that universal screening not cost effective and that best cost effective option currently available is cascade

screening plus using opportunistic pick of new probands from GP note searching or NHS Health checks for cholesterol.

The review describes a recent meta-analysis of eight studies demonstrating that carotid intima media thickness (CIMT), as a surrogate measure of atherosclerosis, is significantly thicker in children with FH compared to those without, demonstrating that atherosclerosis is already in development in children. Such outcome measures could possibly be examined in screening studies to give an indication of whether universal screening might reduce morbidity in children compared with cascade testing. The review states that however, no screening studies were identified, which have looked at these outcomes.
It also states that such measurement of subclinical atherosclerosis requires specialist expertise, and there are also no accepted thresholds of CIMT for defining the presence or progression of atherosclerosis in children.

Regarding cost effectiveness: one review mentioned is that of Marks et al 2003, which use modeling to estimate the number of deaths prevented over 10 years with universal screening at 16 years compared with case finding/family tracing. This study used previous estimates of the number of undiagnosed people with FH in Oxfordshire and applied it to the England and Wales population. Universal screening at 16 years was estimated to prevent 9.8 deaths in males and 1.9 in females over a 10-year period with a screening cost of £9,766 and £9,773 per male and female case, respectively, identified. The total cost per case identified and treated over 10 years was £13,141, with a cost of £527,919 per death averted. By comparison, case finding/family tracing (ages 16 to 54 years) prevented 377 male deaths and 182 female deaths over 10 years with a testing cost of £135 for each case identified. The total cost per case identified and treated by family tracing over 10 years was £3,505, with a cost of £3,187 per death averted. Therefore over the 10-year period, the results of this modelling study favoured the current strategy of family tracing. Uptake during cascade testing is likely to be higher than universal FH screening at 16 years. In this study Marks et al. (2003) assumed that population uptake of the initial screen at 16 years would be 55%, with 75% subsequently attending for diagnostic confirmation. However, a lower uptake would reduce the number of cases detected and so decrease the benefit of universal screening.

The review found no randomised or non-randomised controlled studies (prospective or retrospective) that looked at whether universally screening children for FH is associated with reduced morbidity and mortality. Potential feasibility issues when examining the effects of screening children due to the long duration of follow-up that would be required to look at cardiovascular outcomes and mortality was highlighted.

The review concludes that future prospective studies could potentially examine the effects of universal screening but no clearly established threshold measures that indicate the presence or progression of atherosclerosis in children. The review recommends randomised controlled studies (RCTs) to evaluate the benefits/cost effectiveness of universal screening for FH.

Comments on behalf of BIMDG and the UK Paediatric FH Register Steering Group

1. We agree that the consultation document is a fair and accurate summary of the available published evidence on the topic.

2. In support of early identification of children with FH, there have been several studies from the Netherlands indicating Carotid Intima Media Thickness in affected patients

being significantly increased compared to unaffected cohorts and that this increase was apparent from early childhood. However, in our view, screening at 16 years is too late to be introducing the recommended life style changes (diet, exercise and avoiding smoking) and it is likely to deny the options to commence statins sooner unless identified by cascade screening. This is also an important and probably stressful time during the young person's life with respect to education, social and other aspects of transition from child to adulthood. Unless there is an adequate infrastructure and psychological support for screen positive cases, screening at this age is not ideal.

3. We agree that currently there is insufficient evidence on the impact of child-parent screening. Wald et al. (2007) estimated a positive predictive value (PPV) of 64% for testing in the 1-9 age category using a Fixed False Positive Rate (FFPR) of 0.1% and estimated population prevalence of 1 in 500. Wald et al also indicate that "Child-Parent screening" modelling will take approximately 30 years to attain a critical mass of families.

4. We acknowledge that Wald et al 2011 is the only study of relevance to universal screening in children, assessing the feasibility of the proposal to screen children at the time of routine immunisation at 1-2 years; small sample size - single centre study with 200 children and their parents and 7 health professionals/GP staff. Acceptability for this study was very high; and there was likely to be an ascertainment bias, as it was a simple procedure administered at the same time as immunisation, and none of the children screened were positive. This study does not include the numbers of patients that refused screening, and hence the possible effects of positive screening are unclear.

5. However, Wald et al in our view had rightly addressed the issue with a pilot study in the first instance but we acknowledge the limitations of that study. However, a larger study across 80 centres is currently underway by the same author and his colleagues with a completion date of June 2016. Until the results of this study are reported it would be premature to judge Universal Screening for FH in childhood to be ineffective. The report does acknowledge that the best delineation of positive and negative screening based on cholesterol is in the age group 1-9 years.

6. With respect to the cost effectiveness of universal screening, based on the study by Marks et al, the report concludes that the current strategy of family tracing is favourable and that the uptake by cascade screening would be higher than universal screening at 16 years. This does not discuss universal screening at a younger age – i.e. below 10 years of age. We are aware that positive screening with cascade testing is disappointingly low at approximately 17% and therefore universal screening will certainly be more beneficial in combination or on its own when compared to cascade screening of 1st and 2nd degree relatives.

7 Despite the limited evidence on feasibility, acceptability, cost-effectiveness and the huge logistical exercise for management and follow up of screen positive cases, cascade screening plus child-parent screening is conceptually a good idea. The universal screening model will capture the vast proportion of patients with FH. We recognise that non-paternity would reduce the efficiency of positive screening and are concerned that the low estimates for non-paternity suggested by Wald are likely to be too low and are generally reckoned to be around 10%.

9. We agree that RCTs for FH are not a feasible option, as FH is a slowly progressive condition with the first clinical presentation of Heterozygous FH often being a life threatening cardiac event in early to mid adulthood. RCT's are likely to take 20 to 40 years at least to address the question of benefit vs. cost effectiveness, which is unacceptable and in itself not cost effective or ethical. We emphasise that IMT has been used successfully as a surrogate marker of slowly progressive atherosclerosis and in addition to total cholesterol and LDL cholesterol levels could be used effectively in Universal screening models for FH.

10 We suggest that the Wald et al UK universal screening study is studied prior to rejecting the model of Universal Screening for FH. We suggest that a larger pilot study with appropriate infrastructure and psychological support for screen positive cases is rolled out for a period of 5 to 10 years, should the Wald study demonstrate even a small benefit with respect to child-parent screening for FH. Most metabolic clinicians in the UK would support such an initiative.

Yours Sincerely

Uma Ramaswami

Dr. Uma Ramaswami FRCPH, MD
Consultant Metabolic Paediatrician
Royal Free London NHS Foundation Trust
&
Clinical Lead and Steering Group member, UK Paediatric FH Register;
Member of British Inherited Metabolic Disease Group;
Member of BHF Familial Hypercholesterolaemia Steering Group;
Member of NICE Familial Hypercholesterolaemia Quality Statements Group.

6.

British Cardiovascular Society

(Separate response with same content additionally sent in by Professor David Wald)

Consultation for UK National Screening Committee

"Screening for familial hypercholesterolaemia in childhood"

This review is in response to a personal request by the NSC on this consultation and a society request to respond on behalf of the British Cardiovascular Society.

Author: Professor David Wald

Questions posed to Bazian Ltd (authors of Consultation document) in relation to Screening for familial hypercholesterolaemia in childhood.

1. (i) Whether there is a reliable universal screening test for FH in children and (ii) how it performs compared with cascade testing of index cases.

The answer to part (i) of the question is uncertain. As the authors of this report point out, a study of child-parent screening, a population screening method, has recently been completed in England, involving cholesterol and DNA analysis in about 10,000 children and the parents of children positive for FH. The results are needed to answer this question and are expected later this year.

(ii) Cascade testing is not a population screening method and so is not an appropriate comparator to methods that do aim to screen the population. As stated in the report (by reference to the paper by Morris et al.) cascade testing would miss a large proportion of people with FH in the population because it is self-limiting; once all available relatives have been tested, it stops. Cascade testing relies on a systematic population screening method to identify new index cases.

2. Whether universal child screening reduces morbidity and mortality associated with FH.

The lack of randomised trials of screening showing a reduction in mortality from identifying and treating individuals identified with FH in childhood should not be regarded as an obstacle.
Randomised trials are not needed to identify those at risk of cardiovascular disease, but they are

needed (and have been done) for assessing the efficacy of lowering cholesterol in reducing the risk of cardiovascular disease. If screening were effective and feasible in identifying children and their parents with FH then it can be assumed that treatment with statins or other cholesterol lowering drugs will reduce their risk of having a cardiovascular disease event because, the excess risk is mediated through serum cholesterol and randomised trials show that such treatment is effective.

3. Whether universal child screening and subsequent treatment of screen-detected cases would be ethical and acceptable to professionals and the public.

There are no ethical concerns relating to screening for FH that would override the benefit of preventing premature death in people identified. The results of the Child-parent screening study is expected to provide evidence on the acceptability of screening children.

4. Whether universal child screening would be cost effective compared with cascade testing.
Cascade testing is not a means of reaching the whole population and so it is not a comparator for population screening methods like Child-parent screening (see point 1 above).

Summary

The report by Bazian is correct in concluding that further evidence is needed before reaching a decision on screening for FH in childhood and that the results of the Child-parent screening are expected to clarify this.

7.
(*Response submitted by Dr Eurodiki Drogari*)

I am a member of BIMDG and I received the below e mail about Paediatric FH screening in UK.

Several years ago I had my training for Metabolic Paediatrics at the GOS. Now I am working as a Professor of Metabolic Paediatrics at the University of Athens.

I would like to write to you my brief comments from my 25 years experience on Paediatric FH in Greece. You will see how I collected the FH patients on my own, because unfortunately we do not have as yet well organized system in our Ministry of Health for Screening.

1.We know now that Hetero FH has a frequency of 1:200-250 births of the general population, whilst Homo FH occurs in 1: 200.000-300.000 births of the general population!

2.You can collect quickly doing cascade screening. If you screen for three continuous generations you will find that 1 child will give you at least
3-5 new FH members (adults and children).

3.According to the guidelines you should suspect FH using the Simon Broomes or Dutch criteria. I did not find it extremely helpful because Paediatricians are dealing with very young ages and the parents of the children are quite young and therefore they may not have yet a heart attack.

4.If you screened a child according to American guidelines at 8 or 10 years etc, you will miss the opportunity to inform the family and it will be too late for the child to change lifestyle. I find it extremely difficult to change dietary habits in a child of 8 years of age. It is much easier to teach the whole family when the parents are younger and the children are at the infantile period.

5.You should do selected screening only, but this has some disadvantages because as I wrote above the close relatives are very young in some families and they do not know that they have high cholesterol levels.

6.There is good evidence that IMT studies are abnormal in children as young as 6-8 years of age.

7.There is good evidence that you can use treatment to lower LDL –C even from the age of 6 years and certainly after 8 years.

8.Cholesterol levels are quite stable at the age of 3 years and afterwards. Therefore I would like to suggest, if I may, that infantile period is the best time to screen children for FH.

9.Of course you can use the other way which is to know that a young adult-parent has high cholesterol and check if this happens to one of his parent and after this to screen his child.

Using all the above I am glad to let you know that for the last 25 years I run a Clinic with about 15.000 FH children and adolescents molecularly diagnosed and picked up using the above ways. It is the largest FH single Clinic in the world. Knowing the FH children from very young age you can also study the Natural History of FH which is not well known, to plot on the UK map the place of origin of the eldest FH members and find out the frequency of the mutations in each part of the UK.

FINALLY TO SUCCEED IN ALL THESE YOU NEED TO TRAIN AND INFORM THE PAEDIATRCIANS AND GP's IN YOUR COUNTRY. They can do the first screening and send to you they highly suspected children.

I wish and hope that you will find some of the above a little useful, based in my 25 year experience.

I wish to the UK people involved to the Paediatric FH Sceening the best of success. The disorder is greatly under diagnosed, it is much more frequent than we thought before, it causes Atherosclerosis at very young age which is preventable by changing the lifestyle and using drugs when necessary.

Kind Regards,
Euridiki Drogari
MD, PhD, MSc (Biochem), MSc (Nutr), DCH, MRCP, FRCMetabolPed