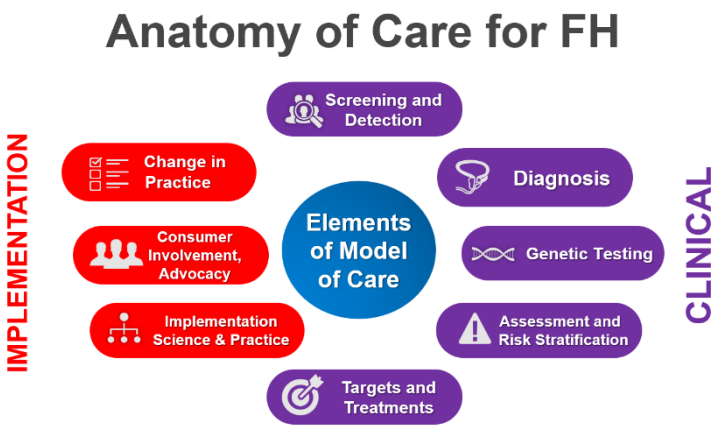
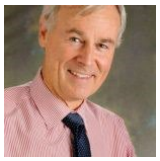
Report: FH Summit 2023 September 9, 2023

The Familial Hypercholesterolaemia Australasia Network (FHAN) brought together a panel of Australian experts for a face-to-face Summit in September 2023. Building on the previous Familial Hypercholesterolaemia (FH) Summits and the State-based workshops, this Summit aimed to identify ways to improve the management of FH and to coordinate the improved treatment of this condition throughout Australia said A/Professor David Sullivan in welcoming everyone to Sydney. He also acknowledged the increased interest in FH amongst paediatricans, adding that they are quickly becoming a key driving force in the quest for optimal management of FH.

Professor Gerald Watts (Winthrop Professor of Cardiometabolic Medicine, University of Western Australia and chair of the FHAN) reminded everyone that FH is a tier one, genetic condition which accelerates the risk of cardiovascular disease by up to 30 years. The key to better management is early detection and early treatment, including in the paediatric setting. There has been an explosion of knowledge in the FH area over the last 10 years including a significant contribution from Australia, including the Integrated Guidance for FH paper (Heart Lung Circ 2021 Mar;30(3):324-349). The approach outlined in this paper can be summarized in this schematic.



Interest in this paper led to a collaborative publication focusing on core implementation strategies - the International Atherosclerosis Society guidance for implementing best practice in the care of familial hypercholesterolaemia (Nat Rev Cardiol. 2023 Jun 15).



A/Professor David Sullivan expanded on the evolving Models of Care for FH. One of the most important areas of endeavour is increased case detection. Most approaches involve the identification of an index case and associated high-risk patients through both opportunistic detection and increasingly the use of digital tools. This usually leads onto cascade screening although universal screening in young children is increasingly discussed.

The primary element within cascade screening is the communication between primary and tertiary care, however, the workforce is simply not large enough to manage the anticipated number of families which need to be evaluated and managed. Healthcare resources and issues such as data privacy and management need to be addressed. The momentum being created from the paediatric sector is encouraging clinicians to take a "Whole of Life risk perspective" but it is also important to understand the consumer perspective and provide the appropriate support. Other areas to consider are gender, special needs such as family planning, age, the risk spectrum and ways to improve adherence. Registries will play a central role but there are issues related to accuracy of diagnosis, the ability to record cases from both the public and private sectors, the use of data for clinical versus research applications and the sustainability of registry management.

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Management of FH has been enhanced by the interest in FH shown by paediatricians and a shared-care model which views FH from the chronic disease perspective. It is important to address adherence in the setting of misinformation and to empower consumers as they are an effective voice for change. Treatment involves an escalating range of medications often with aggressive therapies. Cost issues will need to be watched especially with some of the newer treatments. Trends in healthcare systems are complicated with many inputs such as consumers, geographical considerations, state and federal systems and even international influences. Prof. Sullivan concluded by emphasizing the broad FH consumer experience which we need to both support and encourage to represent the sector.

Professor Gerald Watts outlined the aims and eligibility criteria of the web-based FH Registry, which has 50 sites within Australia encompassing children and adults and some private sector practices.

Aims of the FH Registry

1. Promote awareness of FH
2. Evaluate real-world clinical practice
3. Contribute to scientific knowledge
4. Inform clinical practice guidelines
5. Enable implementation strategies
6. Inform health care policy
7. Improve health outcomes

Eligibility Criteria

Adult INDEX with:

- Genetic mutation that is causative of FH
- Dutch Lipid Clinic Network Score (DLCNS) ≥ 6 (ie. **probable** or **definite** FH)
- Familial elevation of Lp(a)
    - Lp(a) > 0.5g/L with a DLCNS ≥ 3 (ie. **possible** FH)

Child/adolescent INDEX with:

- Genetic mutation that is causative of FH
- Classification of **highly probable** FH
    - LDL-cholesterol > 5.0 mmol/L alone
- Classification of **probable** FH
    - LDL-cholesterol > 4.0 mmol/L with a family history of premature heart disease
    - LDL-cholesterol > 4.0 mmol/L with hypercholesterolaemia or tendon xanthoma or arcus in the first-degree relative
- Familial elevation of Lp(a)
    - Defined as Lp(a) > 0.5g/L with a family history of premature heart disease

All FH RELATIVES of the above

- Classified according to age- and gender-specific LDL-cholesterol cut-offs (Starr et al, 2008)

There are currently 2,356 registrants, most from Western Australia (1,225) with 373 from New South Wales, 365 from Victoria, 217 from Queensland, 130 from Tasmania, 42 from South Australia and 4 from the Northern Territory. Professor Watts encouraged greater participation from all States. Only 32% of the registrants are genetically diagnosed. This equates to roughly 3% of adults and 1.5% of children and confirms the low rate of detection amongst the general population. A very low rate of cascade testing is recorded. Co-morbidities in the registrants are evident with around 30% having hypertension, 8% diabetes and 25% obesity. Pleasingly there was an increase in treatment between enrolment and follow-up (median 2.5 years) which led to more people achieving treatment goals.

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Dr Michell Sarkies, Senior Lecturer, NHMRC Emerging Leadership Fellow, Co-Lead, Sydney Health Partners Implementation Science Academy, Discipline of Physiotherapy at the University of Sydney, described the outputs from the State-based workshops supported by the National Heart Foundation and Amgen which aimed to develop an implementation strategy for enhancing the detection of familial hypercholesterolaemia in Australia, especially important given the 2020 introduction of the MBS item to cover diagnostic genetic testing and predictive testing of family members. Dr Sarkies reminded all of the barriers and enablers to FH detection discussed at the 2021 FH Summit and which formed the basis of discussion at each of the State-based Workshops.

	Barriers	Enablers
Patient-level	• Experiencing care gaps • Lack of insurance coverage • Lack of awareness of treatment options • Side effects from medications • Competing personal or family demands	• Great medical team • Good understand of FH • Useful resources for FH
Clinician-level	• Lack of sufficient evidence • Lack of awareness of FH • Difficulty convincing patients to adhere to treatment regimen • Busy clinicians • Lack of cohesive medical record systems	• Good knowledge of available treatment • Good understanding of genetic results • Clear diagnostic criteria for FH

Several models for FH index case screening were identified across pathology providers, digital screening of electronic health records, interaction with coronary care, cardiac rehabilitation and outpatient clinics, universal screening and options withing primary care and the community. Cascade testing opportunities were identified within pathology providers, a patient-augmented approach and withing general practice with support from tertiary providers. Implementation strategies to support detection were varied. A publication providing details is planned.

Discussion followed.

Phil Aylward opened the discussion by commenting that is clear people with an interest in the area are needed and so the challenge is to get adult physicians involved. *Stephen Nicholls* agreed that this is essential. Preventive Cardiology is an area where jobs of the future will be. *Karam Kostner* emphasized that endocrinologists are also key in this area. *Shubha Srivinasan* added that both Endocrinology and Cardiology Curriculum are both undergoing review and we should be advocating for FH's inclusion. *Stephen Nicholls* added that lack of awareness is a major issue – every medical graduate will see someone with FH and it needs to be in the medical undergraduate curriculum. As well as the basic need to treat high levels of cholesterol early. *Kanga* commented that from the Northern Territory a multi-disciplinary approach is mandatory as most patients have several co-morbidities. *Paul Lacaze* suggested working with the NHF re FH or as he calls it, genetic cholesterol. *David Sullivan* suggested awareness would increase if universal testing were to be introduced.

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PL also suggested that FH Champions with a high public profile would also be an asset.

SN wondered if there is an over-emphasis on the genetic component – it's expensive, genetic counselling can be frightening, mutations are often quite rare, and both patients and GPs often find it too confusing and complex.

JDV said that most consumers call it "family cholesterol".

SS if we don't have familial or genetic we won't capture the children.

KK echoed SN's comment, emphasizing that discussion with patients should not be over-complicated. In addition, several diagnostic tests allow a clearer analysis of patient risk which can be used to

KWS: met with GP Liaison officers. Athero-FH website – untreated LDL – no equivalent on the RACGP literature

Gerald Watts called for all in attendance to encourage and create the Preventive Health experts, whatever their specialty. Genetic testing is important as it very clearly delineates people for risk stratification. GPs are excellent genetic counsellors and patients generally prefer information from them.

SN: there are 400 generally part time genetic counsellors so they are over-burdened. They will not be able to manage the 1 in 200 FH patients in the population, GP is a better way to go for FH.

DS: added that FH is not the disaster that some other genetic diseases are, such as Huntington's Disease.

All agreed that high level genetic counselling for FH is overkill. PL added that they have developed video-based information for patients involved in genetics-based research and finding this very effective and well-received by the participants.

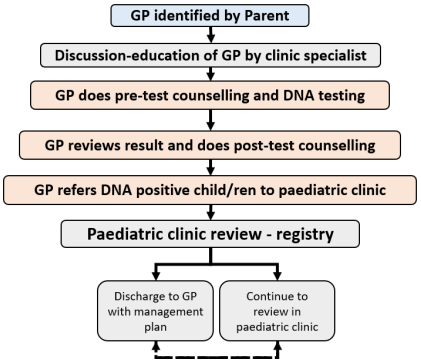
SV: GPs are looking for simple information: Who is the person I should be looking at closely? Agreed that GPs are good at counselling. The word 'genetic' is not a problem – it can be motivating. GP Network health Pathways are really useful. The borderline patient is the problem patient – being able to talk with a specialist or get them an appointment is a real problem.

In closing this session, Gerald Watts asked "How can we combine greater detection with inclusion into the registry? Have the patients been identified and if yes, why are they not being entered into the registry? "

State Updates

NSW	David Sullivan and Shubha Srinivasan summarised the NSW State workshop, which was combined with a Case Detection workshop. A number of opportunities were identified: • Digital: "Lumos", pathology reports, "virtual" clinics, GP Mx software, targeted digital marketing • Social Media: Rx reminder, test reminder, public awareness posts, "Care Monitor". • Patient-facing: Waiting room education, data collection, genetic counsellor/nurse access, point-of-care tests • Systematic: CCU dedicated staff, ICD-10 capture, protocols, home visit/funded family reunion, "Track Gene" or similar • Primary Care: Special interest group in RACGP, Chronic Disease MBS, potential NDIS items QI practice incentives • Community-based: Public awareness campaigns, Centre for Genomics Education, Clarity of message, positive (wellness) message focusing on an increased life expectancy The workshop has led to a pilot assessment of FH Cascade testing in primary care, the assessment of facilitated DNA sample collection using saliva samples, collaboration with paediatric services, interest in early neonatal genetic screening initiatives and a Medicine Today Professional awareness article. A Paediatric FH clinic has been established at the Children's Hospital Westmead for many years. A lipid clinic dietitian sees patients first, the lipid profile is repeated after 6 months and then seen by specialist. The aim is to genotype all patients and enroll them in the FH registry and then consider shared-care. In 2022 there were 50 active patients, 5 HoFH and 4 sitosterolaemia. The clinic accepts referrals from most of Sydney but is starting to upskill specialists in other centres through use of the 2022 guidance (Horton et al JPCH) • General paediatricians • Paediatric endocrinologists • Paediatric cardiologists • Shared care with general paediatricians and GPs Future developments: • NSW TRGS project - cascade testing in primary care, primary-tertiary shared care model, which means paediatric referrals are anticipated • Phenotype test: working with Enzo Ranieri (HOD NSW NBS) who has a strong metabolomics interest and research grant towards a pilot study in 2024 to look at bloodspot cards of known FH children at CHW lipid clinic • Explored Newborn Screening of FH with NSW NBS/ MRFF grant for WES on NBS – decided to wait for Qld pilot study (Natalie Taylor/ Glenn Bennett et al) to generate data for a National NBS Framework submission • Discussed at Paediatric FH working group meetings – families are more open to child being tested as index case but adult item number not entirely suitable for paediatric index case testing. Need to identify how best to lobby for this.

QLD	Tony Hunyh and Karam Kostner commented that new paediatric referrals are being managed by the paediatric endocrinology team (aave 3 homozygous patients). Newborn screening is now underway. An adult awareness program in rural and outback Queensland has commenced using the Heart of Australia Trucks, one which has a cardiac CT machine. An online FH resource for QLD with one for professionals, including a list of those with FH expertise and a patient portal is being developed. 
VIC	Ari Horton and Stephen Nicholls announced that the adult and paediatric lipid clinics will be merged at the Victorian Heart Hospital (VHH) to provide a "Whole of Family Care" approach. This will probably also lead to better recruitment for clinical trials. They added that there can be an over-emphasis on genetics for an ubiquitous disease such as FH. A better approach would be to make it normalised, enhancing education and treatment. There are more individual sites in Victoria in the Registry than in other States. The VHH may become a referral-hub and has a strong prevention section. They are also developing a broader cardio-metabolic program. The Victorian Heart Institute plays both research and teaching roles, which provide opportunities to take FH into community settings such as a collaboration with Wesfarmers and Novartis which undertook risk factor screening in Gippsland (highest risk factors in Australia). Aim to scale that up and that will lead to a greater number of FH diagnoses.
TAS	Andrew Black noted that a dedicated public FH service has recently been established with one clinic a fortnight. Pathology alerts are now in place and a nurse practitioner has been appointed and is making a huge difference in registry enrolments and enhancing adherence. eReferral pathways are now commonplace and cascade screening is being encouraged. Both community and GP education is increasing and early discussions are being held around newborn screening. Index cases are being identified through the cardiothoracic database audit: post-bypass patients with FH need better treatment and the cardiology team have determined to improve management of these patients.
SA	Phil Aylward and Kathryn Waddell-Smith reported that all at the Workshop agreed FH management in SA was a less than ideal. Automated calculations for discharge documentation was discussed. General practitioners emphasized the positive role the GP nurse can play. Communication between specialists and GPs was also identified as needing improvement. Cascade testing is sub-optimal in SA. A state-wide GP education day is planned for November and this will utilize specialists across the spectrum and includes cardio-metabolic and genetics sessions. They are aiming to increase registry data entry through a wider group of clinicians. There is a Lipid Group in SA with both adult and paediatric specialists which may be a conduit to spread education throughout the State and some established protocols to encourage cascade testing.
WA	Gerald Watts and Andrew Martin commented that the real gap identified at the WA workshop was the identification of children with FH. This spurred the clinicians to really consider current practices and how to get cascade testing to work and aiming to identify at least 25% (ie 500) of children with FH in WA over 3 years. These decisions have led to: • Implementation of the first stages of a primary-tertiary shared-care model for genetic cascade testing • GP facilitator employed at tertiary centre • Patient advocate employed as counsellor • Advocating a centralised co-ordination of cascade testing

	• Development of Health Pathways for FH and Cascade Testing for FH and advised the local GP networks. The package includes template letters and family brochures for cascade testing and advocates a quick phone call by the specialist to the GP. • Perth Children's Hospital has established links with all adult tertiary services (Royal Perth Hospital, Sir Charles Gairdner Hospital and the Fional Stanley Hospital)  *Health Pathways for FH and Cascade Testing for FH materials are available for other groups to use as a starting point when developing their own materials.
NT	Nadarajah Kangaharan commented that the Northern Territory was the last area to join the FHAN. It is a very large area (about 18.5% of Australia's land mass) with a population of 246,500. Aboriginal people are over-represented in the health system: 50-60% vs a population of 30%. Geographical and workforce issues are significant. FH Registry Update: The support from the FH registry has allowed a 0.2 FTE nurse to be employed. In turn, all ACS patients coming through RDH CCU have been screened (n= 511) with 12 consented and added to the register and 8 pending consents. Other patients have been identified in Central Australia, through paediatric cardiologists, adult general physicians and genetic testing. Limited Cascade Screening has commenced, and GP education sessions are planned. A MDT Preventive Cardiology Clinic (GP + Cardiac CNC + Cardiologist) has been established along with CTCA + CACS services in Darwin. A Lipid Audit of the previous 12 months was conducted. Of the ~300 patients reviewed, 22 have definite FH and 13 probable FH based on the Dutch Lipid Score calculator. None of the patients identified had received any further investigation of their FH status. Also developed locally and soon to be launched widely is the Territory Kidney Care (TKC) program which will be able to identify all patients with high lipids, assess cardiovascular risk and Dutch Lipid Score and present the data appropriately for each specialty, including GPs. The NT detection effort is progressing well, enabled by the NT Integrated Electronic Medical Record – CDS. The next step is to formally engage with the primary care team to assist with optimizing treatment and for the genetic and cascade testing process to be established.
ACT	Tony Lafferty advised that following on from the ACT workshop, the Hospital Working Group is facilitating the development of FH flags in the Electronic Medical Record. There is a plan to provide a letter to index cases to promote cascade screening. The Primary Care Working group plan is to build a FH pathway into GP software, develop a Primary Healthcare Network Health Pathway, continue relationship development with Sonic re genotyping and for pathology companies to flag pathology likely to be FH. To address these aims there have been discussions aiming to set up parallel public/private pathways from pathology to clinicians for patients with elevated LDL-C. In addition, Sonic has employed a genetic counsellor for those having screening through their lab. A well-received presentation to Paediatric Grand Rounds has led to ongoing discussion with general paediatrics

	team. Discussion with hospital biochemistry continues on topics such as obtaining deidentified data to estimate prevalence in the outpatient cohort, options to filter secondary causes with existing data, and developing an algorithm to flag LDL thresholds and alerts to referrers. There have been preliminary discussions to establish a public lipid clinic with cross-referral between adult and paediatric services. A pathway and interface needs to be established between primary, secondary and tertiary services to make this achievable. Such a clinic would also provide an opportunity to participate in clinical trials. Discussions have touched on increasing community awareness within healthcare professionals: hospital- and community-based as well as a parallel process to increase community awareness. Industry support would be valuable in this space. It is also considered important to 2-3 paediatric consultants with an interest in FH. It would also be useful to increase awareness of the Medicare Codes for identification and cascade screening. In terms of awareness, perhaps a parliamentary friends group or targeting medical practitioners in federal parliament, or an FH Champion would help any awareness campaign. The patient information on the AAS-FH website is excellent, but perhaps needs to be in more languages.

DISCUSSION

KK: CABG patient data fascinating. Measuring Lpa --- answer not recorded. Lpa in indigenous patients would be very interesting. Combine paed/adult clinics a very interesting idea. SS: Geographical coordination in Sydney is a hurdle. SN: at the VHI, both clinics are within the one service which helps with administration, but the relatives will be spread across the country. PL: Familial Cancer groups are almost all now running virtually which may be a model to follow.

PL: How reluctant are indigenous people to undergo blood collection/genetic testing and what are the canonic mutations involved? KN: It is a more complex process involving lots of family members and interpreters but doable. AH: Working with an indigenous partner and going to the families (house to house) with a first-language interpreter are needed for a successful project. No knowledge of specific FH gene panel appropriate for the Aboriginal population. GW added that there is a reluctance to genetic testing but not phenotypic and that a culturally-sensitive approach is required. DS: Would neo-natal screening be an issue? AH: babies have the right to make their own decisions from birth and so much more work about cultural ramifications is needed. Whole community screening is more successful.

SS: Nationwide newborn screening is currently running at ~95%. Adding in an extra test at regular childhood vaccinations does not attract the same success rate. MS: If there is newborn screening for FH some years in the future, how could cascade screening be scaled up? SH: Adding any test into newborn screening would need careful planning as paediatric clinics would be overwhelmed. SS: Identifying the newborn is probably more about identifying the parent and their relatives who don't know they have FH and managing the reverse cascade-screening. Treatment of the child will generally be limited to diet until they are able to commence statins (8 years old). MS: There are 300,000 births in Australia annually, which suggest 1200 will be identified with FH. How will this workload be managed? AH: It won't – the workforce resource requirement is too great. It will need AI or some other digital process to address the need. PL: The cancer-based video could be adapted for a FH-patient information/consent process. SN: The AI-digital process is appealing but will struggle outside the research setting, but there are opportunities. GW: The NIH have funded several studies which show an increase from 2 to 30% when digital resources are utilised. However, in WA their success rate

in consenting patients is around 65-70%. It's important to integrate the systems and monitoring effectiveness. ZA: DNA testing needs to cost around $250 per person for people 18-25 to be cost-effective.

SN: What can we learn from national screening processes? Would a national registry with a link to the Department of Health work to automate family notifications? GW: This is similar to what happens in southern Spain. AH: The legality of notification of at-risk family members is important. An app or link which can be forwarded to family members may be more effective than a paper letter. KH: developed a GP package for families of index cases allowing them to communicate via email or a QR code and while this is only a month-old program, there gave been 2 families expressing interest. DS: We must also remember that recipients will have different levels of interest. JDV: The GP is incredibly important in the cascade-testing process. AB: Pathology databases show LDL >5mmol/L on multiple times, so we do actually know who the patients are but we don't seem to address it. PA: Patients seem to think diet will work, and many people are really reluctant to take a statin. At the specialist level it may be possible to show them their blocked arteries via echo, but at the GP level it can be really difficult to convince them to start treatment.

NR: Would a national testing program in teenage years be an answer? GW: In the US recommendations are that children should be screened between 9 and 11, and then again at 18 years of age. Need to look to the NHF and Government to address awareness. AM: 90% of the children who should be on a statin are not taking them. Would a family clinic to address management of each family member assist? GW: In some countries, non-adherence for children is 30%.

TL: Chronic disease healthcheck (aged 45-49) – could this be adapted for FH screening and remove the age limit.

SS: Needs to be a public awareness campaign managed by public health experts rather than already convinced clinicians. Involving a high-profile person is very successful.

TH raised concerns about the impact of this trial NBS screening for FH with regard to lack of formal consultation with paediatric services, and the likely significant pressure on paediatric services in terms of new referrals for which there is no funding. SS agreed that awareness campaigns and screening programs would significantly impact paediatric services.

Dr Glenn Bennett, a founder and Chief Medical Officer of Genepath outlined their approach for Targeted Gene Sequencing for FH newborn screening. Their platform involves Targeted probes for 132 genes and allows customisable high-volume screening for up to100 treatable conditions in a short time frame. It uses the blood spot which is routinely collected from newborns and its development has been supported by both NSW and Federal Governments. There is a 4-year program to expand newborn screening and FH is one of the conditions which could be considered for inclusion. The merits of including FH in newborn screening have been identified in the scientific literature since the 1970s – a key positive is the early identification of young parents with FH.

The Genepath targeted gene sequencing methodology for newborn screening was developed nearly 10 years ago and has since been validated in several diseases. Most recently three genes which cause FH have been investigated as part of the screening panel and the process has been found to be accurate, cost-effective and feasible. Clinical validation of the assay is underway. It is hoped that Genepath data currently being generated will support an application for the inclusion of FH newborn screening.

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Associate Professor Paul Lacaze, Head, Public Health Genomics Program at Monash University opened by saying that several approaches will be needed to identify all those with FH, including newborn and population genetic screening. His team have recently explored the cost-effectiveness of adult genomic screening, and have identified the appropriate price-point, particularly when combined with screening for several diseases. This research is being expanded to explore the public acceptability of adult screening for hereditary disease, including FH. The project was launched on TV and through social media. Within the first 24 hours, 10,000 people expressed an interest in participating and this grew to 25,000 over a week. High-risk patients are being identified across the country and referred for appropriate management. It is hoped that this pilot study will evolve into an appropriate additional method of adult screening for FH across Australia.

Professor Zanfina Ademi, Professor of Health Economics, Health Economics and Policy Evaluation Research (HEPER) Group, Faculty of Pharmacy and Pharmaceutical Sciences at Monash University explored the Economics of Detection Strategies. She reminded the audience that chronic disease is responsible for 83% of premature deaths and 66% of the burden of disease, but only attracts 1.34% of all health spending on primary prevention, or $89 per person.

Australia was one of the first countries to introduce the fourth hurdle cost-effectiveness assessments when deciding which treatments are supported by taxpayer funding. An unpublished research project has reviewed all of the published cost-effectiveness screening strategies for FH, most of which were cascade-screening studies relying on genetic diagnoses. Most of the screening strategies (cascade, opportunistic, systematic and population screening) assessed showed robust cost-effective results and that FH detection could be implemented with positive results.

Current health economic models significantly underestimate the benefits of existing and newer therapeutics by neglecting well-known disease biology. Given the short-term follow-up of trials, interventions that may have more long-term benefits could systematically underestimate the benefit of interventions. Professor Ademi outlined the Australian and Dutch health economic evaluation of screening and treating children with FH, which importantly and uniquely includes the number of years of high cholesterol in the economic calculation. The findings of this study suggest that the early detection and treatment program for FH in children may offer a good value for investment, being both health and cost-saving. The return on investment for the detection and treatment program for FH in children was €8.37 ($9.12) from a Dutch perspective. These results are important as atherosclerosis in FH starts at birth, and childhood is an important period for screening and prevention – gaining health and saving costs over a lifetime.

Discussion: these cost-savings may not transfer to newborn screening as cascade screening already selects for high-risk patients. Adding in "reverse" cascade-screening of patients this may improve the cost-effectiveness of newborn screening. Healthcare should ideally be considered an investment rather than a burdensome cost to society.

Ref: Ademi Z, Norman R, Pang J, Liew D, Zoungas S, Sijbrands E, Ference BA, Wiegman A, Watts GF. Health economic evaluation of screening and treating children with familial hypercholesterolemia early in life: Many happy returns on investment? Atherosclerosis. 2020 Jul;304:1-8. doi: 10.1016/j.atherosclerosis.2020.05.007. Epub 2020 May 20. PMID: 32526542.

Ademi Z, Norman R, Pang J, Sijbrands E, Watts GF, Hutten BA, Wiegman A. Cost-effectiveness and Return on Investment of a Nationwide Case-Finding Program for Familial Hypercholesterolemia in Children in the Netherlands. JAMA Pediatr. 2023 Jun 1;177(6):625-632. doi: 10.1001/jamapediatrics.2023.0763. PMID: 37126315; PMCID: PMC10152372.

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Ms Jenny Della-Vedova, a leading patient advocate from Western Australia updated the audience on the newly forming FH Australia Foundation. Patient advocacy provides a voice for patients to governments, researchers and other stakeholders to ensure patients receive the best possible care and outcomes. It supports patients and their families in their healthcare journey, guiding the patient and family through the healthcare system, through screening, diagnosis, treatment, and follow-up. A patient advocate helps patients communicate with their healthcare providers so they get the information they need to make decisions about their health care. This process empowers patients, improves communication and provides support.

Three generations of Jenny's family have been cascade tested for FH by a well-coordinated process of shared care between the specialist who first made her genetic diagnosis and the family GP who has now tested her great niece and nephew and provides ongoing care to the extended family. This personal experience, along with the statistics (1 in 250 people have FH, 3 babies are born in Australian with FH every day and 90,000 Australians walking around not knowing they have FH) has led Jenny to become the lead person in founding Familial Hypercholesterolaemia (FH) Australia, whose purpose is to advance health by improving Australia-wide awareness, support and understanding, access to diagnosis and treatments for individuals and families with FH. The organisation will be an Australian not for profit charity, providing advocacy, education, awareness of FH along with support for FH patients and their families. Funding will be obtained from industry partners, donors, grants and perhaps research projects. A Board and a Scientific Advisory Committee will be established, along with State branches and advocacy groups. Ideally a national help-line will be established.

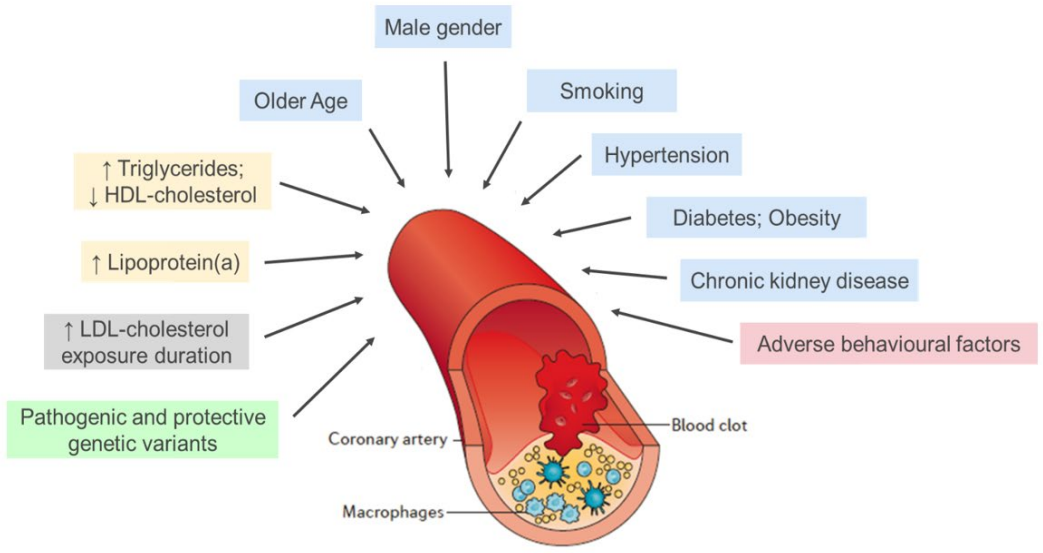
Jenny concluded by inviting all to reach out to FH Australia if:

- they have patients/families interested in forming a state/territories branch
- they have patients that want to join us as members (FREE!)
- they need consumers involved in research/grants
- they are interested in the development of resources (or have resources to share!)
- they have FH matters that need advocacy
- they would like to partner with us on specific projects
- they want to provide a central resource for patient and family information

By working together, healthcare providers, patients, and advocates can increase awareness, improve access to care, and develop new treatments and guidelines for FH.

THE WORKSHOP

Professor Watts commented that the afternoon focus is management – how do we implement the guidelines we have all contributed to, remembering that there are many other risk modifiers in familial hypercholesterolaemia plus the disease itself, which means that risk stratification is at the root of management whatever the age of the patient.

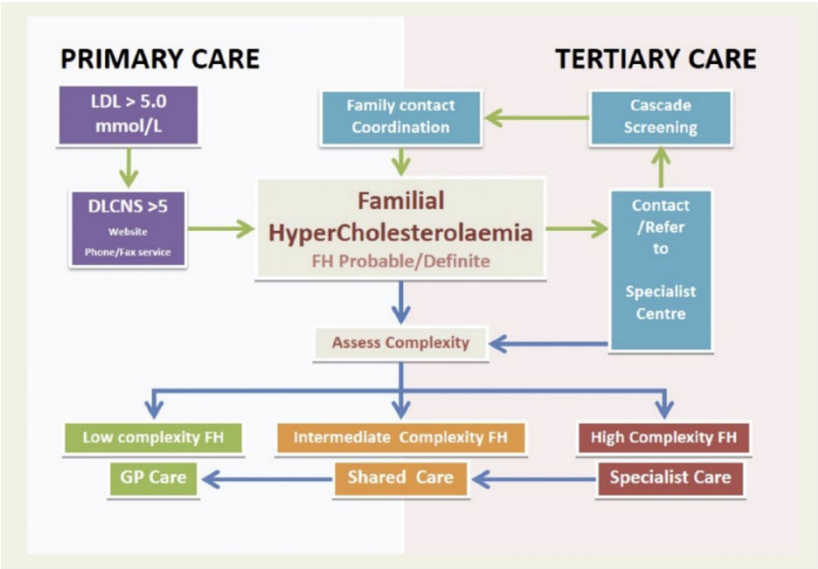


HDL, high-density lipoprotein; Lp(a), lipoprotein (a); TG, triglyceride

Khera AV, Kathiresan S. *Nat Rev Genet*. 2017;18:331-344
Perez de Isla L *et al Circulation* 2017;135:2133–44.

Shubha Srinivasan commented that there are probably 20,000 children in Australia with FH – only 250 are captured in the FH registry. A major complication of coordinated FH management is the wide variety of specialists who may be treating a paediatric FH patient. Cascade screening, or reverse cascade screening will increase the detection of FH cases and lead to measurable health benefits but will have real workforce implications.

Adherence is a major factor in treating patients with FH – about 30% of whom are likely to be non-adherent. If patient's disease is low complex, they can be managed well in general practice, a highly-complex case should be referred to a tertiary service,
and a shared-care model may work best for the remaining patients.



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FH Summit 2023 was presented by the Familial Hypercholesterolaemia Australasia Network and the Australian Atherosclerosis Society. With thanks to the 2023 Education Partners: Amgen and Novartis.

Managing children and adults with heterozygous FH

Improved awareness is a major need amongst GPs, specialists and patients when treating patients with heterozygous FH. It is important for both the detection of FH or simply to ensure that a high LDL is treated, and for the understanding of the importance of adherence to therapy. Keeping patients motivated to maintain treatment in general practice is not easy. As cholesterol is asymptomatic, motivating patients is often problematic while post-myocardial infarction patients are easier to motivate. Changes in the treatment of acute coronary events, with reduced hospitalization has also meant a reduction in the dissemination of cholesterol-related information to the extended family. Discharge summaries/handover documentation for primary care is often sub-standard and should be improved. Use of quality website information with clear patient and GP guidelines is helpful (see athero.org.au/FH), making sure they are up to date and consistent with published guidelines.

The general awareness of the risks of high cholesterol is very low amongst patients. Presenting information in a way which is appropriate for each patient is important. Providing a patient with a target LDL level may be helpful.

Paediatricians concentrate on primary prevention – the challenges of treating a 10-year-old and reducing the risk of them having a coronary event in 20 or 30 years' time are immense. Many paediatricians are reluctant to commence statins in children. Children are often hyper-responsive so using a very low dose is very effective and most often without side effects. Adherence in children will most likely be increased with injectable treatments, which are also likely to be very well accepted by both parents and children.

Patients need to be advised of the benefits of intervention, as well as the generally low level of side effects.

Family planning and the management of pregnant women/young mothers with FH is also a fraught area for management of FH. Contraceptive treatments and appropriateness of investigations such as CT angiograms have generally not been covered in FH guidelines. However, teenage treatment can also be a motivating factor – get cholesterol levels under control before you need stop treatment for pregnancy.

The severest form of FH – homozygous FH

Both paediatric and adult cases should be managed in the tertiary setting as they are very complex and very high risk. Access to the newer, and hence more expensive, treatments is also an issue. Lipoprotein apheresis appears to be underutilised, although it is difficult in children. Liver transplantation is appropriate in some patients, including children. Special referral centres within each State are needed.

An increased awareness of high cholesterol and FH, particularly if newborn screening is implemented, will create enormous workforce issues which also need to be addressed.

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