
**Report: FH Summit** October 27, 2024

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The Familial Hypercholesterolaemia Australasia Network (FHAN) brought together a panel of Australian experts for a face-to-face Summit in October 2024. Building on the previous Familial Hypercholesterolaemia (FH) Summits and the State-based workshops, this Summit provided updates in several areas of the management of FH, thereby updating others on State-based activities and the development of a risk-reduction pathway for children.

**A/Professor David Sullivan** acknowledged the traditional owners, thanked the education partners and welcomed everyone to the FH Summit, both those in Melbourne and online. He commented that the FH Summit is facilitating progress and a highlight of the AAS's Atherosclerosis Education Program.

**Dr Ari Horton** (paediatric and genetic cardiologist and clinical geneticist at Monash Children's Hospital and Victorian Heart Hospital; co-chair Paediatric Working Group FHAN) addressed precision population screening strategies and outcomes. In Australia there are around 90,000 people over 15 and 22,000 under 15 years of age with undetected FH. Multiple strategies can and should be used to improve detection. However, he stressed that there must also be an implementation program developed as it is unlikely that if all patients were identified, the Australian healthcare system would be able to manage everyone within current structures. By employing population screening, the benefit comes from not only identifying the single individual at risk but also cascade testing and managing all their at-risk family members. Dr Horton recommended that the focus be on raising awareness of FH in younger people, identifying and treating earlier than traditionally through the use of new screening strategies. He concluded by reminding the audience that we owe it to our patients to identify and treat children with familial hypercholesterolaemia early so that they can lead a long and healthy life. Preventative health care is more cost-efficient and saves lives.

**Associate Professor Damon Bell** (physician and pathologist) spoke about the role of the Clinipath's trial FH detection program. In the four years since the Medicare item has been available, only ~2500 index cases and 418 cascade tests have been performed. Clinipath (Sonic Healthcare, Pathology) are trying to reduce the barriers by increasing awareness through direct messages to clinicians accompanying laboratory results, clinical education and clinical audit activities for GPs focusing on FH, and a new diagnostic service for primary care in which patients will be flagged, and on referral from GPs, patients may be contacted directly by Sonic pathologists to augment FH genetic testing. Genetic counselling is a key part of the national service offered by Sonic. Around 94% of those identified with FH by the Sonic team on a recent audit from the Sonic Genetic counsellors discuss the diagnosis with family members and 64% undergo both pre- and post-test counselling. The index cases are not surprised (32%), feel relief (38%) and sometimes disappointment, fear or sadness (30%) when given the molecular confirmation of FH. Seventy percent of patients prefer a letter from the doctor to assist informing family members, who are often interested to know more despite some concern for their wellbeing.

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**Dr Jane Tiller** (Senior Research Fellow, School of Public Health and Preventive Medicine, Monash University) addressed ways to resolve genetic discrimination. Soon to be published data exploring the reasons why some people decided not to participate in DNA Screen, a national research program for detection of FH and cancer predisposition gene variants, was discussed. The recent proposed changes to legislation governing insurance and genetic data, which will ban the use of genetic tests in determining premiums or cover for life insurance, were highlighted.

**Professor Charlotte Hespe** (Head of General Practice and Primary Care Research, University of Notre Dame Australia in NSW) discussed new approaches to screening in general practice. Noting that if GPs did everything they "should" do, it would mean 36 hours of work per consultation!

The CHECK (Cholesterol Hereditary Evaluation and Cardiovascular Risk) program has been designed to assist in identifying people who may be at higher risk of having Familial Hypercholesterolaemia by gathering personalised information before appointments with their regular GP. Patients are sent an SMS text message via the "Better Consults" IT platform linked to the electronic medical record. They are asked a series of questions about cholesterol and a family history of high cholesterol, aligned with the validated Dutch Lipid Score. If the patient is identified as being at risk of FH, they are requested to complete an FH screening questionnaire. The results of this questionnaire are then made available to the GP at the time of the scheduled appointment, along with evidence-based recommendations regarding suitable investigation and management options.

This system allows the GP and patient to prioritise their issues for discussion – and could be used as a targeted screening tool for FH in the primary care setting. The program has a high level of acceptability, especially with younger patients. The results of an initial trial of the pre-appointment FH screening program are currently in press with AJGP (publication date 20 December 2024).

**Dr Andrew Martin** (paediatrician, Perth Children's Hospital) explained the successful FH in Kids Program. Three children with FH will be born every day in Australia. It is important to remember that this is a treatable paediatric disorder. The likely adverse cardiovascular outcomes can be prevented or minimised. The FH in Kids program aims to identify 25% of all children with FH in WA over 3 years through effective cascade testing. Working with primary care and nurse practitioners is critical, being potentially more effective than conventional than hospital-based programs. All potential opportunities for detection of FH, such as newborn screening, child-parent screening at immunization, teenagers, DNA screening and coronary care unit screening should ideally be integrated.

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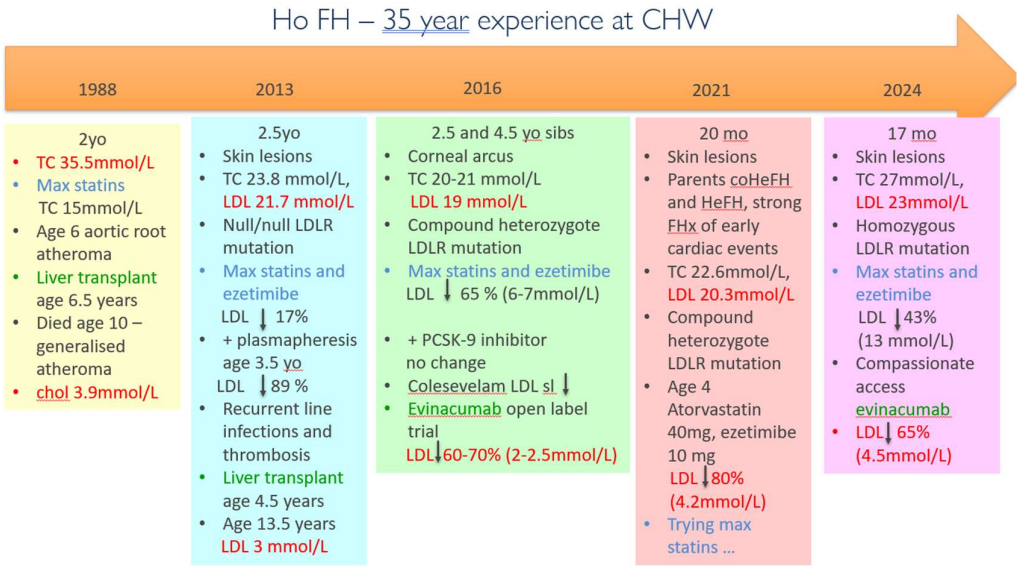
**Clinical Associate Professor Shubha Srinivasan** (paediatric endocrinologist, The Children's Hospital at Westmead; co-chair Paediatric FH Network Group) discussed new therapies for children with FH given the importance of addressing the lifetime LDL burden. Most patients are started on statins, which work in children and are safe. The Children's Hospital at Westmead now has 35-years' experience of homozygous FH, which most probably affects 1 in 300,000 people. The lifelong exposure to extremely elevated LDL-C leads to an exceedingly high risk of developing premature atherosclerosis and cardiovascular events at a very early age. The table below shows how treatment has changed over 35 years.




Treatment options for children with FH continue to evolve. If the patient has some residual LDL receptors, PCSK9 inhibitors or gene silencing with small, interfering RNA molecules (siRNA) or monoclonal antibodies may be useful. Lipoprotein apheresis for paediatric homozygous FH patients can result in 60-80% LDL reductions but it is a very time-intensive and expensive option. Liver transplants have been performed since the early 1980s – the longest reported survival is nearly 30 years. Angiopoetin-like 3 (ANGPTL3) regulates LDL-C through an LDL receptor independent mechanism and monoclonal antibodies such as evinacumab which inhibit this process have been shown reduce LDL by up to 50% on top of other therapies and regardless of LDL receptor function. Cost is the limiting factor with evinacumab at present. Lomitapide inhibits microsomal triglyceride transfer protein (MTP), which prevents the formation of apolipoprotein B, VLDL and chylomicrons. It is an oral, once-a-day formulation which also appears to reduce LDL by up to 50%. Gene therapy and gene editing for homozygous FH are still being investigated, with safety and feasibility remaining critical issues for children.

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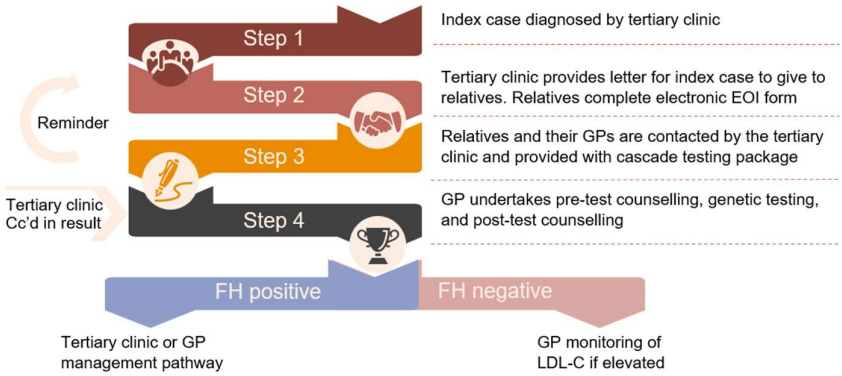
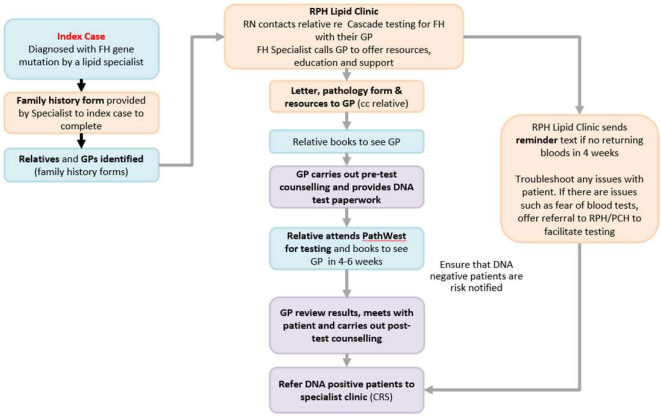
**Dr Karen Birkenhead** (Post Doctoral Research Fellow, University of Sydney) addressed the implementation of a primary-tertiary shared care model to improve the detection of FH which was implemented at the Royal Prince Alfred Hospital and NSW Health Pathology as described below. The involvement of the genetic counsellor has been important and the various packages (cascade screening, positive/negative results) found useful. Preliminary results were presented, showing an increasing in uptake over time, with the study still in progress.




**Dr Jane Purdie** (General Practitioner, WA) described the EDICIE Program (Enhanced Detection of Familial Hypercholesterolaemia in Primary Care), which aims to to enable patient-centric, cascade testing via GPs, to upskill GPs in undertaking genetic testing and care of FH and to increase number of FH relatives tested in the community.




Around three-quarters of all relatives have participated in cascade testing, allowing treatment initiation or in those found to have FH. Children with FH are referred to Perth Children's Hospital for management. Barriers to cascade genetic testing among relatives include family dynamics, needle phobia, other life priorities, lack of motivation and psychiatric/psychological issues. Patient reported outcome measures and GP experience of testing need further evaluation. Dr Purdie concluded that development and implementation of effective risk reduction pathways are needed in GP, especially for the care of children, review and assessment of these pathways is ongoing.

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**Ms Jenny Della-Vedova**, chair of the newly formed FH Australia and a leading patient advocate from Western Australia updated the audience. FH Australia is a health promotion charity and peak body that supports, educates and advocates for individuals and families with FH. FH Australia Ltd was founded to advance health by improving Australia-wide awareness, understanding and access to diagnosis and treatments for individuals and families with FH. Incorporated in November 2023, FH Australia is now a registered charity with deductible gift recipient status. The FH Australia Board is based in Western Australia but has commenced reaching out across Australia to ensure that it is a truly Australia-wide organisation. The Professional Advisory Boady consists of expert clinicians and academics. Several resources have been developed and are available via the website (https://fhaustralia.org.au/). Physical resources such as posters and banners for clinics are available. All attendees were invited to contribute content and announcements for the website and social media posts. FH Awareness Day was marked at 3 hospitals in Western Australia which included heart health checks, FH Education and point of care testing and will be more extensive in 2025.

FH Australia
VOICE FOR INHERITED HIGH CHOLESTEROL

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## State Updates

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| NSW | Shubha Srinivasan reminded the audience that the only tertiary Paediatric FH in NSW is run monthly at the Children's Hospital Westmead. A Western Sydney Health Network combined paediatric-adult health pathway has been developed. This model includes referral to general paediatricians, cost allowing, to reduce the clinic load and as many GPs are not confident to start statins in young children. This pathway is available for other services to use as a template.<br>A lipid clinic dietitian sees patients first via telehealth and ideally genetic studies completed before being seen by the specialist. |
| VIC | Rebecca Quin commented that the Royal Children's Hospital clinic has about 60 patients and is lucky to have the support of a dietician – who following the suggestion from NSW will now see the patient before their physician appointment which is anticipated to have positive benefits. There is no health pathway in Victoria – one would be helpful. Ari Horton added that the Victorian Heart Hospital also has a paediatric clinic and together with Steve Nicholls they are contributing both child and adult data to the FH Registry. A dietician is not part of their service, but they do participate in several clinical trials. Edward Janus commented that there are also adult clinics at St Vincent's, the Alfred and Austin Hospitals. There are no resources such as genetic counsellors, dieticians, data entry to assist with cascade screening or Registry input. |
| SA | Kathryn Waddell-Smith reported that there is one tertiary paediatric clinic which runs monthly and has a very long waiting list. The demand-supply misbalance continues into the adult world as there are only 3 public adult lipid clinics. There are some benefits of the statewide electronic medical record which can result in earlier diagnosis by accessing parent data during a consultation. Ian Hamilton-Craig commented that the SA lipid group is concerned about the lack of understanding of FH in the community, with this being exacerbated by misreporting in the media. Quoting Prof Roger Williams "never too early, never too late" he encouraged all of us to ensure positive stories are reported in the media. |
| WA | Andrew Martin commented that the paediatric service has a nurse practitioner associated with their clinic and this has been a real bonus, allowing the clinic to manage numbers and see patients more quickly. Increased collaboration with Royal Perth, Fiona Stanley and the Sir Charles Gairdner Hospitals has also improved cascade-testing but this area will remain a focus for 2025. Pleasingly, most children recently diagnosed have now started on statins, with an average of 35% reduction in LDL. In 2025 a major focus will be investigating ways to improve adherence to treatment. Other projects will be looking at digital solutions to cascade testing and FH management (similar to current diabetes apps) as well as a universal screening program for FH in childhood. Gerald Watts added details of two projects:<br>• EDIFICE: showed an increase in the annual rate of diagnosis and referral of FH in children since 2008 and an associated 41% reduction in LDL in these patients.<br>• DYNAMITE: looked at young, non-adherent patients and discovered that postcode and education were significant factors in adherence to medication/lifestyle advice. Individuals will be invited back to discuss options which might suit them. |
| ACT | Tony Lafferty advised that with an ANU Student, Rachael Liu and others, they are aiming to demonstrate that add on lipid screening to patients having blood taken for other purposes will improve FH diagnoses. Patients will be provided with Opt in/Opt out information. GPs and parents will be given educational materials including a link to consumer website. The trial will run for 6-7 months and hopes to screen nearly 2300 patient specimens. They anticipate 9-10 positive and additional people from reverse cascade screening. GP resources include Health Pathways and link to key papers such as Watts *et al*, Horton *et al*. They hope to utilise general paediatrics and interested clinicians/healthcare workers such as dieticians to assist with FH Registry data entry. |


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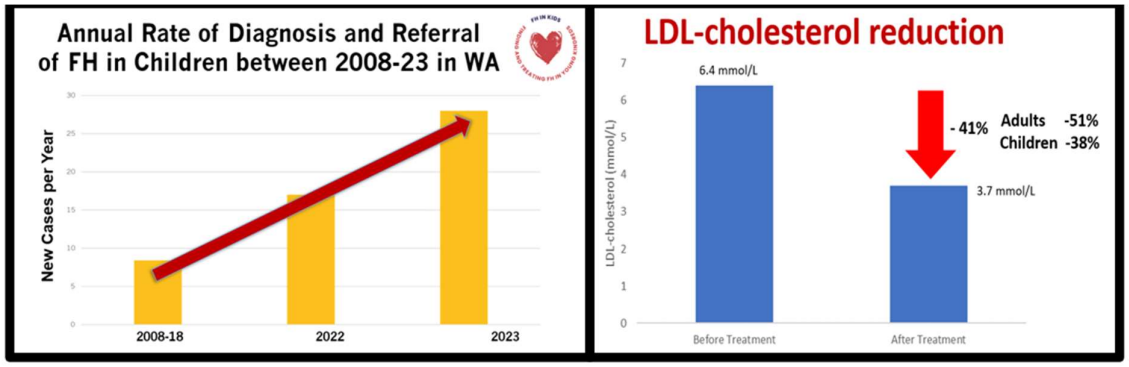
| | Tony congratulated the FH Australia team commenting that their website is excellent and will be a great resource. |
| NZ | Nikki Reid commented that NZ clinicians are working with parliament re genetics legislation. The Christchurch clinic is run as a family clinic, rather than separate adult and child clinics. Funding and resources remain an issue. Campbell Kyle commented that the rest of NZ does not offer the same level of service/support as in Christchurch. There is no national FH registry. Statins are the mainstay of treatment – newer medications are not available. Callum Wilson heads the national paediatric metabolic service which have about 50 children with FH across the country. |

**Professor Gerald Watts** (Winthrop Professor of Cardiometabolic Medicine, University of Western Australia and chair of the FHAN) advised that the FH Registry Board has determined to move the registry to another platform – REDCAP. The infrastructure will be managed through the University of Western Australia which will allow the registry to continue and at a reasonable cost. All sites will require new ethics approval. Further funding will be required to provide limited support for data entry and registry management. With nearly 3,000 eligible entries, there are opportunities for PhD projects.

**A/Professor Natalie Taylor** (Director Implementation to Impact, Director of Research, School of Population Health, University of New South Wales) reminded all that Newborn Bloodspot Screening (NBS) tests for ~30 genetic conditions selected based on whether a childhood intervention is available and recommended with aim to avoid death or disability. While whole genome sequencing determines the entirety, or nearly the entirety, of the DNA sequence of a person's genome, targeted genomic sequencing examines a select set of genes for genetic variations for early detection of genetic conditions and may be more affordable and appropriate. Two projects which will address the effectiveness and implementation strategies for targeted adapted gene sequencing for NBS were described. Investigating the suitability for including FH into NBS has been complex and controversial. Several changes to the current screening process have been recommended and a FH Pathway developed. All involved in these projects agree that it is important to identify people with FH at the earliest possible stage. This is clear cut for HoFH but less so HeFH. NBS for FH appears to be feasible and it will be explored in this Queensland-based project.

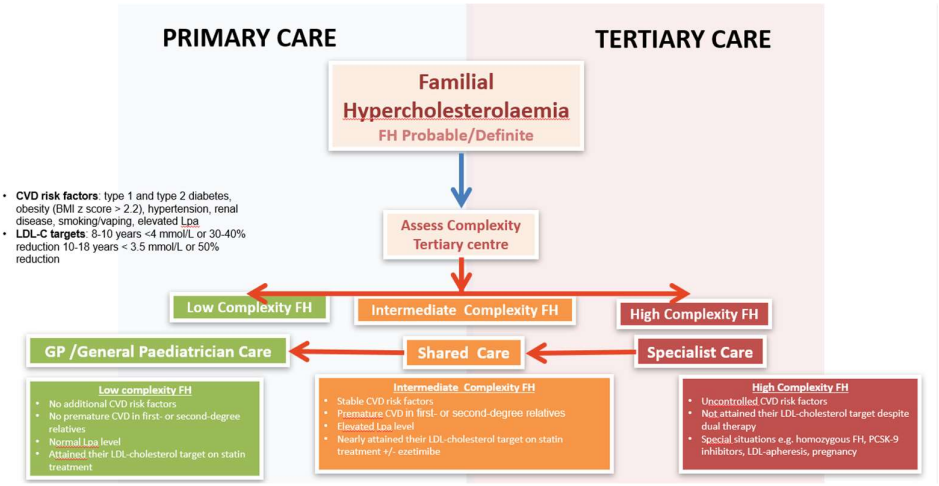

## THE WORKSHOP

**Dr Michell Sarkies** (Senior Lecturer, NHMRC Emerging Leadership Fellow and Sydney Horizon Fellow at the University of Sydney), described the development and implementation of risk-reduction pathways for children and adolescents with FH. The first phase of this project was a meeting with all stakeholders in Western Australia and input from those present was invited to map barriers and solutions to implementing such a pathway. The FH in Kids program has shown the benefit of involving GPs in screening and diagnosis of FH.




Most GPs preferred to refer a child to a specialist or at least discuss treatment initiation before medications are prescribed. Guidance for treatment of children and adolescents is available.

The workshop held aimed to consider the balance between general practice, shared care and specialist care, with the following model as a starting point.

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The FH Summit workshop focused on developing and implementing a risk reduction pathway for familial hypercholesterolaemia (FH) in children, emphasizing a shared care model. It was suggested that this model could involve initiating statin therapy in a tertiary clinic, with ongoing management by general practitioners (GPs) who have access to FH specialist support when needed. Although there is no MBS Item number for phone consultations between medical doctors, case-conferencing could represent an option for support. Shared care models like those used in shared pregnancy care, when applied to FH, may allow early management by GPs unless the patient is high-risk.

To enhance access to FH specialists, creative solutions such as virtual consultations were proposed. Timely and specific communication regarding patient management responsibilities between providers is crucial. The implementation of this model will require funding for expert advisory lines and pathways, as well as upskilling for all involved, particularly in treating children with FH. Addressing any knowledge gaps in FH is essential, with an emphasis on different treatment targets for FH patients compared to the general population. Educating and empowering families to advocate for optimal care might support better treatment outcomes.

Many families prefer continued management by their family GP, but geographical relocation can complicate care. Technology, such as an FH App, could support families and children, similar to those used in type 1 diabetes. Stakeholder involvement such as involving nurse practitioners where available is vital for reframing current models. An audit process should be part of exploring new management processes for FH, incorporating education, upskilling, audit and continuous professional development will be attractive to GPs.

FH is a chronic condition affecting families and communities. A care plan involving allied health, with regular reviews, can attract financial compensation for GPs and applies to both children and adults. The model must also cater to rural and remote populations, potentially through Telehealth. Activity-based funding options vary across states, and support for clinicians and financial strategies need consideration in both private and public sectors. FH Australia could assist patients in navigating their risk-reduction pathway.

The attendees at the FH Summit then discussed the ideal approach when a patient is identified with FH. The advice given in extensive discussion, facilitated by Mitch, was collated and will be analyzed accounting for outcome of initial WA workshop; further focused workshops with states that were not present (Tasmania and Northern Territory) are planned.

Mitch thanked everyone for their contribution, adding that they will be developing a pathway with local implementation into routine care.

Gerald Watts closed the FH Summit thanking all for participating. He also thanked the AAS and Education Partners for their support.

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