Rapid Report: October 1, 2022

The Familial Hypercholesterolaemia Australasia Network (FHAN) brought together a panel of international and Australian experts for a virtual summit in October 2022. Building on the 2021 summit, which focused on detection and implementation strategies, the first day highlighted the complexities and opportunities when managing children with Familial Hypercholesterolaemia (FH), the challenges of managing treatment across all ages, and delved into practical aspects such as genomics and family planning.

Professor Gerald Watts (Winthrop Professor of Cardiometabolic Medicine, University of Western Australia and chair of the FHAN) reminded everyone that 1 in 250 Australians are likely to have FH, but 90% of adults and 98% of children remain undiagnosed. This means that over 112,000 people are unaware that, depending on the severity of their genetic defect, onset of cardiovascular disease for them may be brought forward by 15-30 years. The aim of the FHAN and everyone attending this Summit is to change the trajectory of this CVD through appropriate earlier detection. education and treatment.

Dr Shubha Srivinasan added that, as a paediatrician, she was excited to start the Summit with a focus on paediatrics – especially as FH is a disease which starts at birth.

Dr Amy Peterson, a Pediatric Cardiologist from the University of Wisconsin School of Medicine and Public Health, opened her presentation on newborn screening for familial hypercholesterolaemia by declaring "What's Old is New Again". Principles of screening have been part of medicine for decades, and case reports of infants with elevated cholesterol have been appearing in the literature since 1957. In 1992 the National Cholesterol Education Program (NCEP) released the first guideline covering children, acknowledging that FH is an important issue and suggesting it is a disease that meets the criteria for a widespread screening program.

The implications of FH were again emphasised when in 2011 Integrated Guidelines for CV Health and Risk Reduction in Children and Adolescents were released by the Heart and Lung Institute. Their recommendations were:

- 0-2 years old: no screening
- 2-8 years old: Fasting lipid panel for at-risk children
- 9-11 years old: Non-fasting lipid screen for all children
- 12-17 years old: Fasting lipid panel for at-risk children
- 18-21 years old: Non-fasting lipid screen for all patients

However, actual practice and science are not matching up. Only 3-5% of those aged 9-21, 25% of high-risk children, and 60% of 20-30 year-olds have had their cholesterol measured. Newborn screening happens in 99% of countries, but cholesterol is not included in the panel. Even when FH is diagnosed, only 15% of people are prescribed medication.

Diagnosing FH at birth could mean that over 99% of the population is tested for FH and those with homozygous FH (HoFH) could be diagnosed at that time. This would allow the genetic nature of the disease to be made clear to clinicians and families and the baby's relatives could be diagnosed decades earlier. This may also lead to a heart-healthy lifestyle starting at birth and statin therapy at 8-10 years of age.

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If such an approach were to be taken, regulators and clinicians must avoid misinformation or misunderstanding of FH, particularly if the health care system is not prepared. There may be difficulties with access to life insurance, or discrimination due to a preexisting condition. There may also be consequences within families, perhaps due to non-paternity issues or feelings of anxiety or helplessness from time of diagnosis until initiation of statin therapy.

Refs: Circulation 2018;137:2218-30. J Clin Lipidol 2020;14:224—30. J Pediatr 2017;188:87-90. Clin Cardiol 2014;37:119-25.

Dr Ari Horton, a consultant paediatric cardiologist and geneticist at Monash Health and with clinical expertise in holistic healthcare for children and adolescents with cardiac issues, outlined the recently published FH Model of Care for children and adolescents, which was developed by many clinicians and researchers to address the gap in Australia and New Zealand. Dr Horton emphasized that "Early diagnosis and early treatment changes lives. Morbidity and mortality are reduced with treatment. We must do what we can!"

Given that HoFH leads to an increased cardiovascular disease load around 14 years of age, it is imperative that we make FH easier to diagnose and more consistently treated. He added that genetic testing should be offered, but we need a greater understanding of and a greater exposure to genetic counselling. All health professionals working in the area need to be trained, not just genetic counsellors, increasing the effectiveness and number of conversations across families with FH.

Treatment recommendations have been simplified within the Model of Care. It is important to initiate treatment and support families, emphasizing that medications are safe and effective. Diet and plant sterols can have an impact in early years. Statins should be introduced when families are ready and comfortable – the Model of Care outlines practical advice about starting statins in children. Other therapies are currently being trialed in children.

Children are often the index case in a family and so general practice is vitally important in the diagnostic process. In some current cases there is a shared team, involving an endocrinologist, metabolic physician, paediatric cardiologist, pediatrician, genetics specialist, general practitioner and nurse specialist – FH management may end up entirely with community physicians. Further funding and education is needed.

Dr Horton closed his presentation by inviting any interested to join the Paediatric FH Working Group. Updates and improvements to the National Guidelines and the Medical Benefits Schedule are needed, which gives this Working Group some important areas to address.

Ms Jenny Della-Vedova is currently working with the FH team at The Royal Perth Hospital. As Jenny has FH and was the index case in her family, she was able to bring a real-life consumer perspective to the discussion. There are now 4 generations of her family who have been tested and those needing treatment have been identified – a great outcome following on from her initial consultation with a diligent general practitioner.

Jenny noted that there are many concerns of those diagnosed with FH. For some it's insurance, for others it's potential side effects related to treatment, while others have financial limitations. But as one-third of babies with high cholesterol will have a heart attack before they turn 50, she felt it was important to allay these concerns wherever possible. (PCH/Institute study).

Advocacy is a crucial component of best practice. Jenny is aiming to establish an Australian FH Advocacy Group, with input from the FHAN and the European Familial Hypercholesterolaemia Patient Network. Her aim is for Australia to follow Spain's lead – where all cholesterol-lowering treatment is free to those with an identified genetic variant. Jenny thanked everyone for everything they do in diagnosis and treatment. It's life-saving.

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Professor Tom Brett from the University of Notre Dame, Western Australia and **Professor Karam Kostner** from The Mater Hospital, Brisbane, co-chaired the next session, which focused on the management of patients with FH.

Translation to practice: improving identification and management of FH in primary care was the topic addressed by **Professor Nadeem Qureshi** of the University of Nottingham, United Kingdom, who started by emphasising that it is important to persuade general practitioners (GPs) of the importance of identifying FH – particularly as we all understand that GPs are increasingly time-poor.

Professor Qureshi suggested several approaches which may assist. The first of these, a practice took-kit, asks questions like:

- How many of your patients have had their cholesterol checked?
- If 1:250 people have FH, then how many would this mean for your practice?
    - How many have actually been identified?

He has also developed a new algorithm (FAMCAT) which uses individual patient data, such as the highest ever total/LDL-cholesterol levels, age, triglyceride level, on treatment/not, previous history, family history, and other causes (eg liver disease). Acknowledging that GPs do not have the time to collect all details for all patients, Professor Qureshi suggested that a risk-related strategy may be appropriate. Patients who are in the highest risk category should be encouraged to meet with a health team member (nurse practitioner, GP or specialist) for an in-depth consultation.

Another simple approach would be to identify patients with a total cholesterol >7.5 mmol/L if under 30 years, or over 9 mmol/L if over 30 years; this is also a good indicator for further investigation.

Professor Qureshi added that as important as identifying people with confirmed FH, is the need to make sure they are properly managed in primary care. Firstly, the diagnosis must be noted in their healthcare record. Secondly, each patient must be monitored, including a repeat cholesterol 3 months after initiation of treatment – noting that this should be aiming for a 50% reduction in non-HDL cholesterol. Finally, it is again important to ensure that these follow-up measurements are recorded.

Ms Natalie Raffoul, National Healthcare Programs Manager at the National Heart Foundation of Australia, then presented a compelling case for assessing and addressing non-adherence to medication. In Australia, cardiovascular medication non-adherence rates range between 14 and 43%. Nearly 1 in 4 post-myocardial infarction patients are non-adherent, and lipid-lowering therapy is the least adhered-to cardiovascular medication. Ms Raffoul stressed that we should see non-adherence as a risk factor.

Factors that impact adherence in FH largely relate to the silent nature of FH. Perceived side effects are often a big issue, particularly with younger people. Family history can sometimes aid adherence, but "dad didn't have a heart attack so why should I take medication?" can lead to non-adherence.

Practical and credible resources on medication and lifestyle advice for the patient are needed at the time of diagnosis. Genetic counselling often helps patients to understand the severity of their disorder and this benefits adherence to treatment. It is important to emphasise the effectiveness of medication, rather than instilling fear, and to discuss medication within an FH context, particularly reinforcing the medicine side effects are not common and can be easily managed. It may also be useful to lean on family to support and model treatment adherence.

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In Australia, FH patients are likely to be eligible for a free Home Medicine Review by a pharmacist – although this requires a GP referral. The conversation is often different from that with a GP and is highly recommended.

In summary. Ms Raffoul encouraged all treating physicians to keep things simple, find the time to educate patients and ask about adherence at each and every consultation.

Professor Kausik Ray from Imperial College, London, provided an update on the latest treatments available for FH. As Professor of Public Health, Honorary Consultant Cardiologist, Head of Commercial Trials and Deputy Director of the Imperial Clinical Trials Unit at Imperial College London and President of the European Atherosclerosis Society, he had a wealth of knowledge to share.

Professor Ray opened his presentation with a powerful reminder – time is of the essence for those with FH as the natural progression of coronary atherosclerosis is hastened. Most often, by the time a diagnosis of FH is made – the average age at which FH is diagnosed is 44 – there have been 44 years of exposure to high cholesterol levels and damage to blood vessels. To address this, we need to either target production of atherogenic products (circulating LDL-C and apoB lipoprotein) or improve elimination of them.

The LDL receptor is the most efficient way of removing circulating LDL. Statins make more LDL-receptors in order to remove LDL from the blood stream. PCKS9 inhibitors stop the destruction of LDL-receptors.

Accepted first-line treatment for those with FH is ezetimibe plus a statin – together they can achieve ~75% decrease in LDL-cholesterol.

PCSK9 inhibitors lower PCSK9 levels and therefore LDL, but the liver keeps making more PCSK9. Fairly high levels of the inhibitor are needed, but generally give a sustained 60% lowering of cholesterol.

RNA-based therapies, working either in the cytoplasm or the nucleus, are under investigation and work by reducing the amount of PCSK9 produced by the liver. Positive results in FH patients already on statins plus ezetemide have been reported. The newest oral agent is bempedoic acid which works though the same pathway as statins, although on an enzyme earlier in the pathway, achieving an additional 18-28% reduction in cholesterol depending on level of statin co-treatment.

However, people with HoFH do not have LDL-receptors, or have defective LDL-receptors, and so treatments which work independently of the LDL-receptor are needed for these patients. Two such agents are lomitapide, which stops the assembly of the ApoB-containing lipoprotein, and evinacumab, which influences the catabolism of triglyceride-rich lipoprotein and production of LDL particles.

Early diagnosis and treatment is known to reduce the cardiovascular risk imposed by FH. Combination therapies which work in different ways to reduce lifetime cumulative cardiovascular burden. While most therapies work though increasing LDL-receptor number or activity, in patients with HoFH, alternative therapies that work independently of the LDL-receptor must be considered.

Professor Ray concluded that the challenge is to use medication early enough in our patients to make a real difference.

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Dr Shubha Srinivasan, a consultant specialist in Paediatric Endocrinology at The Children's Hospital at Westmead, outlined the history and management of 8 patients with Homozygous FH (HoFH) who presented to The Children's Hospital at Westmead, Sydney between 1988-2022. These case histories demonstrated the immense burden of FH on families.

The age of presentation ranged from 7 months to 2.5 years old, and total cholesterol from 16 - 35.5 mmol/L. Some children did have family members with known FH or parents with raised cholesterol levels, although they were not always on cholesterol-lowering treatment. Presenting features included xanthomas, corneal "haziness, yellow papules, and fatty skin lesions. In all cases, siblings were also investigated, with several then also having an FH diagnosis.

Each of the children had treatment initiated and several ultimately received liver transplants. However, the opportunity for a liver transplant is limited by the lack of availability of donor organs. While the longest survival post-transplant is over 20 years, immunosuppressives can have adverse effects.

Pharmacological treatments have included statins, statins plus ezetimibe, and more recently evinacumab, which is showing a 50% reduction in LDL-cholesterol levels. Inclisiran, a 6 monthly injection targeting PCSK9, which lowers LDL by 52% has recently been approved by the TGA and FDA for adults with heterozygous FH. Trials in children are continuing.

Gene therapy trials using adenovirus-vector gene replacement are underway in adults. However, there is a limitation of gene addition in a paediatric cohort as the vector DNA will be lost with cellular turnover due to growth of the liver. Developments in this area will need to integrate new genetic material into the host genome at a specific defined locus.

Dr Srinavasan was pleased to conclude that the treatment of severe FH in children is currently changing, with some exciting gene therapy options under development.

Professor Jan Radford, Professor in General Practice at the University of Tasmania and **Dr Dorothy Graham**, a physician at the King Edward Memorial Hospital in Perth then led discussion particularly relevant to the management of those with FH in general practice.

Dr Sibel Saya, a Research Fellow in General Practice at the University of Melbourne presented a primer in genetics, tailored to FH.

Genetic testing of the LDL-receptor, ApoB and PCSK9 genes confirms diagnosis of FH in about 80% of cases. Mutations in these genes are inherited – a mutation in only one copy of the gene is sufficient to cause disease and all first-degree relatives are at 50% risk of also carrying the mutation.

Dr Sibel reminded attendees that is important to consider FH in those with premature atherosclerosis cardiovascular disease (men aged under 55 and women under 60), those with a family history of premature cardiovascular disease, anyone with an LDL-cholesterol >5 mmol/L particularly if they are under 40, and those who present with tendon xanthomas, xantholasmas or corneal arcus.

Once a patient has been diagnosed with FH, treatment should follow the FH guidelines and genetic cascade screening of relatives should be offered. In Australia, testing of relatives is covered by an MBS item in first- and second-degree relatives. Dr Saya recommended sending the request to the same laboratory as the index case, if possible, to enhance reporting. If this is not possible, for example because of geographic challenges, then providing details of the index case laboratory is useful. The pathology request form should state 'cascade gene test' and 'family history of FH', detail the relationship of the patient to the relative with a known genetic

test result (which ideally are attached to the request) and include the patient's cholesterol result to give the genetic pathologist some context.

Drawing a family tree with the patient will help identify the family member the index case should be speaking with.

Insurance is a common worry for those potentially with FH. It is important to know that in Australia there is a moratorium on the use of genetic/genomic test results in life insurance policies until June 30, 2024. Income protection and disability insurance policies are also often raised by families. Consumers are not required to have to disclose the results of any genetic test when applying for insurance and companies should not ask about genetic testing. However, medical and family history is reportable.

Mr Luke Elias, Director, Operations and Engagement, Healthy North Coast brought a unique perspective to the discussion, as a father of two young boys with compound heterozygous FH and having FH himself, and working with about 1,000 general practitioners across northern NSW. He tends to find that general practices and patients ask the same questions:

- How easy is it to find or get access to health information or health care services?
- Which information or service is relevant for the situation I am in right now?
- What decision do I need to make to improve or maintain health?

When discussing the development of care plans within general practice, Mr Elias noted that it is important to assign specific and measurable actions for patients to reach goals. He suggested using memorable time targets, making the "grand final" for each goal matter, for example, by Christmas, lose 5kg or make a decision about treatment. He emphasised that the milestone needs to matter to the patient. Inclusive care plans are those that are understood and co-owned.

In a region with the highest number of "health pathways", the messages can become very confusing and so you need a memorable plan if you want to cut through. Three practical tips were offered:

1. Read the room and set a pace that EVERYONE can keep up with - your patient, you, others involved in care. Things might be happening in a patient's life that affect their ability to commit at that time.
2. Frequent monitoring of the metrics that matter - understanding, experience of care, happiness and a sense of feeling well really matter.
3. Use of care navigation and case management for vulnerable and priority populations – some need extra help to get to the start line (finance, literacy, time, anxiety, other issues), someone to cheer on during the race, more support at the finish line

In these modern times, digital solutions such as "apps" are often seen as the way forward. However, if the patient or the caregiver is not ready for digital, it will fail. Mr Elias encouraged all health practitioners to really work together with each patient to find a set of targets and solutions that work for that individual.

Professor Jeanine Roeters van Lennep, is head of the vascular medicine clinic specializing in hereditary dyslipidemia at Erasmus University in the Netherlands. She combines this research interest with gender-specific medicine, which made her thoughts on family planning and management of pregnancy for those with FH particularly relevant.

Women with FH requiring contraception should be prescribed low-dose oestrogen oral agents, an IUD or other barrier method. When they are thinking about starting a family, it is important to have a discussion and recommend stopping statins 3 months before conception until the end of lactation. Women should be confident that there is no increased risk of maternal pregnancy complications, although FH is associated with atherosclerosis in the placental arteries. Family planning also involves counselling male partners.

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Professor Roeters van Lennep added that as LDL-cholesterol often increases in pregnancy, she and other were keen to follow women with FH and document the life course of pregnancy and statin use. Just over 100 women with FH (70 Norwegian and 32 Dutch) were part of this study. Average age was 39 years, with statins having started when they were 19. On average, they had 2 children.

About one-third of women took statins at some stage in their pregnancy, 87% in the first 12 weeks, and 13% for the duration of their pregnancy. Twenty percent of women had more than 4 years of pregnancy-related off-statin time. And 22% of women reported that they stopped breastfeeding earlier than recommended because of their wish to recommence statin treatment. Importantly, nearly 90% of the participants wanted more information on pregnancy and breastfeeding in relation to FH.

Professor Roeters van Lennep concluded that young women with FH lose years of treatment when discontinuing statins in relation to pregnancy and breastfeeding periods and should be closely followed up to minimize the duration of these off-statin periods. Whether these periods of interrupted treatment increase the cardiovascular risk in FH women needs to be further elucidated. A broader project is planned, and she invited Australian physicians to become part of this study.

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Rapid Report: October 29, 2022

The Familial Hypercholesterolaemia Australasia Network (FHAN) brought together a panel of international and Australian experts for a virtual summit in October 2022. The second day developed the discussion and recommendations from Day 1 and focused on implementation strategies.

Professor Gerald Watts (Winthrop Professor of Cardiometabolic Medicine, University of Western Australia and chair of the FHAN) reminded everyone that it is most important to improve the identification of those with Familial Hypercholesterolaeima (FH), particularly children. The aim of the FHAN and everyone attending this Summit is to change the trajectory of the resultant cardiovascular disease in those with FH through appropriate education and treatment.

In the first presentation **Professor Laney Jones** of the Department of Genomic Health at the Geisinger Research Institute spoke about implementing treatment guidelines – from an implementation science perspective. Simply, implementation science is trying to close the 'know-do' gap by shortening the time from evidence generation to adoption in clinical practice. There are over 100 theories, models and frameworks which describe implementation practices, highlighting the complexity in this seemingly straightforward area.

Implementation strategies are defined as methods or techniques used to enhance the adoption, implementation, and sustainability of a clinical program or practice. Familiar to all clinicians, but perhaps not spoken about and analysed. Professor Jones added thar she and Australia's Dr Mitchell Sarkies developed a framework to help improve guideline translation. The process starts at the guideline development stage and incorporates implementation science methods into the cultivation of clinical practice guidelines by reviewing the evidence and developing implementation recommendations.

The local implementation context is addressed by assessing barriers and enablers to the implementation, tailoring strategies, monitoring, evaluating, and sustaining the implementation. Three key questions incorporated into this framework re:

- What implementation strategies have been used to promote the uptake of statins?
- How completely are the implementation strategies utilized reported in studies designed to promote statin uptake? Most acknowledge who/what/targets but less when, how often, why and measure
- Which implementation strategy, or combination of strategies, is (are) the most effective at promoting the uptake of statins?

Nicely enhancing the earlier presentation by Natalie Raffoul, Professor Jones highlighted the barriers and facilitators to FH treatment.

	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

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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

Multidisciplinary lipid clinics (MDLC) are often recommended as the gold standard for treatment fo complex conditions like FH. This contention was well supported by an evaluation undertaken by Professor Laney and colleagues. In their evaluation of the first year after implementing a new care model, they found there were many individuals within their system with lipid conditions, but only a small number had been referred to the MDLC (0.25%), indicating additional implementation strategies may be needed to improve the reach of the MDLC to improve patient care. They found MDLC improved patient prognosis based on risk stratification, increase in guideline-recommended treatments prescribed, and clinically significant lowering of targeted lipid levels necessary for the prevention of future CVD events. In patients with FH seen through the MDLC, the mean reduction in LDL-C was 79 mg/dL (2.04 mmol/L), which is predicted to reduce CV events by 40%. In addition, through MDLC, the number of patients achieving a target of LDL-C below 100 mg/dL (2.56 mmol/L) increased from 15 to 69%, a more than 4-fold increase.

Professor Jones concluded that implementation science offers methods and tools to expediate the translation of evidence into routine clinical practice. While there are many theories, models, and frameworks exist and are utilized for different purposes, she emphasized that there is not one perfect answer, it is important for each clinical to choose what works for them.

A/Professor Damon Bell, an endocrinologist and clinical pathologist with both private and public hospital affiliations in Western Australia, as well as working for Clinipath Pathology, arm of Sonic Healthcare explained Sonic's role in healthcare and in particular, the Familial hypercholesterolaemia diagnostic service.

Sonic is the largest pathology provider in Australia, Germany and Switzerland and is the largest operator of medical centres across Australia, with more than 2,000 doctors who run their own practices from one or more of 160 clinics.

One of the baseline measures provided by Sonic to clinicians are the automated messages suggesting further investigation, and possible FH, sent in a tailored manner to specialists and general practitioners. These tailored messages have been shown to improve patient treatment and/or referral, but there is still room for further improvement.

Primary care clinicians are central to the detection of FH - and they are very good at using the Dutch Lipid Score (DLS) to identify those with FH. Sonic is planning on further encouraging this this by implementing a General Practice LDL-c Audit (will attract CPD points).

Sonic Healthcare believes that one of their strengths is that all genetic testing is undertaken in Sydney at the one highly experienced laboratory, thereby enhancing cascade screening outcomes. However, numbers are lower than ideal – only 18/570 with LDL >5 mmol/L have genetic testing, and only 10% of these undertake cascade screening.

Genetic counselling is also part of the Sonic FH Program. Pre- and post-test genetic counselling is provided free of charge. Counsellors work with index cases to assist identifying relatives who could be offered the

opportunity to participate in cascade screening of that family. Importantly, individual Sonic practices will work with clinicians to streamline testing in local systems, or across state-wide or countrywide systems.

A/Professor Bell summarised that while FH is common, at least 90,000 people are un-diagnosed in Australia. We have very good tools to diagnose FH with a Medicare rebate available for genetic testing and there are very effective treatments for FH. However, an increased awareness of FH based on an initial high LDL level is required. Sonic Pathology offers a very comprehensive FH diagnostic service including genetic counselling with nationwide links and is committed to improving patient outcomes and supporting healthcare professions in the pursuit of medical excellence.

Professor David Sullivan then introduced **Ms Tiffany Boughtwood**, the Managing Director of Australian Genomics- a real lynchpin in the national genomics scene.

Ms Broughtwood outlined the role of Australian Genomics in supporting health implementation of genomic and genetic technology. Its aims are twofold – firstly to progress a sustainable nationally-coordinated approach to genomic research to improve the efficiency, reach and timeliness of genomic research projects; share genomic research datasets to enhance genomic research in Australia. Secondly, Australian Genomics aims to support Governments in the communication and translation of genomic research outcomes and inform evidence-based policy development and implementation.

Australian Genomics is currently supporting 130 projects and initiatives (over $100 million) as well as some led by Australian Genomics themselves. One of these is PanelApp Australia, an open online platform for gene and virtual gene panel curation. The platform was initially designed by Genomics England for the 100,000 Genomes Project, and a local instance was deployed by Australian Genomics in December 2019 and is currently being used to identify and resolve discrepancies in gene and virtual gene panel curations between participating services, and with Genomics England.

Another project is SHARIANT, which facilitates data sharing across 11 laboratories across 4 states. So far there have been more than 18,000 total shared variant interpretations and 39 discrepant variants identified and 2146 variants submitted to ClinVar. This project is expanding to incorporate additional Australian labs, including private, and some based in New Zealand.

Australian Genomics in collaboration with Patient Support and Advocacy Groups identified and responded to a need for patients and the community to have access to reliable and evidence-based resources on genomics and its implications. This led to genomicsinfo.org.au being launched in March 2019 and it provides information for patients and the public about genomics, such as easy-to-understand genomic materials (factsheets and infographics on topics of genomics, genomic testing, data sharing and insurance), shares additional resources from other research and medical organisations and publishes blog posts on current research and news relating to genomics and genomic testing.

Australian Genomics is also developing guidelines for the involvement of community members in a more meaningful and effective manner within the research program. Three other community programs have also been launched recently in three specific groups: indigenous peoples, ethnic minority ancestry groups and school children (years 5-12).

Linking back to earlier presentations, Ms Broughtwood reminded us that usually when people are found to have gene changes that lead to an increased risk of developing disease, they are told that it's important to

provide and success of information to their blood relatives who might also be at risk. However, it can be difficult for genetics services to ensure that the right information gets to those blood relatives. This has led to a new Genomics Australia project "What are the barriers to clinical genetic services contacting blood relatives of patients directly, with patient consent, to increase the uptake cascade testing?"

Genomics Australia are also involved in workforce education to improve the general levels of understanding of genomics within workforces and to discover what targeted approaches to workforce education would be useful.

Yet another three projects recently implemented ddevelop publicly available indications and guidelines for pharmacogenomic testing in Australia, generate national benchmarks for developing, analysing and reporting Polygenic Score results and a genomic education program for science students from Yr5 to Yr12 and their teacher.

Much has been achieved by Genomics Australia as outlined by Ms Broughtwood, who hoped that the sector and Genomics Australia's place in it would continue to strengthen despite changes in the political scene.

Dr Mitchell Sarkies, of the Schools of Health Sciences at the University of Sydney has been working with the FHAN and the broader FH clinical community for the past few years. He commented that while people are being detected with FH, the use of MBS items particularly for cascade screening is less than optimal. To understand why this might be so and how it might be improved, Dr Sarkies has led meetings in each State/territory to develop a model to improve this both locally and nationally.

The 2021 FH Summit in 2021 identified barriers and enablers to FH detection.

Barriers	Facilitators
• Difficulties in obtaining a good, reliable family history • Difficult family dynamics • Lack of adherence • Competing priorities in clinical practice • Lack of infrastructure • Lack of awareness • Health literacy issues • Prominence of interventional cardiology • Incompatibility of guidelines • Costs and funding not fit for purpose • Lack of urgency	• Enlisting champions • Targeted public health messages • Providing good letters to GPs • Systematise collecting family history • Developing clear care guidelines and pathways • Robust referral pathways • Promoting shared care • Adapting care pathways to local context • Raising awareness of FH • Reduce reliance on craft groups • Upskilling colleagues • Develop GP, nursing and allied health workforce • Integrating tools into practice software and workflow

These were used as a starting point for discussion with the State-based groups. Routine practices and procedures using integrated tools were important part of the conversation.

The project has been termed the 10-P Project, and involves representatives from 10 areas important in the: **p**hysician, **p**aediatrician, **p**recision medicine, **p**athology, nurse **p**ractitioner, **p**harmacy, **p**olicy and public health,

implementation science, pharmaceutical industry and general practitioners. Importantly, there was at least one person representing each of these areas involved in the discussions.

Each meeting framework followed the Expert Recommendations for Implementing Change (ERIC) strategy which has been shown beneficial in other areas requiring intervention. Discussion has centred on the FH Model of Care, index case identification, cascade screening and support.

Several models for FH index case screening have been discussed and suggested either enhancements or improvements in several areas: pathology reports, digital screening of electronic health records, collaboration with coronary care/cardiac rehabilitation units, universal newborn screening, and community pharmacy involvement.

Not all GPs are aware that there is an available system to assist with health record screening: TARB-Ex a validated data extraction tool to help identify patients at high risk of FH in general practice electronic health records is available to all practices using Best Practice Software - and is royalty free!

Options for cascade screening were also canvassed at these meetings. The most pressing need identified was for a dedicated centralised support person to manage the families involved, acknowledging the funding difficulties.

Nine component of implementation strategies have been identified. Tailoring to local contexts, sharing resources between States (no need to reinvent the wheel), state-specific FH websites, patient advocacy, central coordination for cascade testing, involving all stakeholders, GP education and financial strategies all need to be addressed.

Dr Sarkies concluded that once the State and Territory meetings are complete, the data and suggestions collected will be analysed using an implementation science framework and will lead to the development of State and Territory implementation strategies for improving the detection of FH within Australia.

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