Office of Population Health Genomics  
October 2017 Newsletter

2017 has been another successful year for The Office of Population Health Genomics (OPHG), and the team feel very proud to present their major milestones and achievements in this edition of the OPHG newsletter. OPHG’s notable work in 2017 to date includes progressing the key initiatives of the WA Rare Diseases Strategic Framework 2015-2018 at a local level and contributing to the development of a policy framework about the role of genomics in health at a national level.

Along the way, OPHG developed new partnerships, received a unique opportunity to showcase their work to an international audience and was also met with a couple of line up changes within the team.

Progressing well into the second half of 2017, OPHG is excited about what other opportunities the year will bring as the team continue their work through key projects and collaborations to improve the WA health system and outcomes for people living with rare and genetic diseases.

We’re making progress!

At the end of June, OPHG hit the half way point for the implementation period of the WA Rare Diseases Strategic Framework 2015-2018. At this key milestone, OPHG would like to reflect on key achievements to date in supporting the needs of people living with rare and genetic diseases.

Milestones achieved:

- Contributed to the development, implementation and evaluation of the Undiagnosed Diseases Program WA (UDP-WA)
- Established the WA Genomics Health Network Executive Advisory Group (WAGHN EAG)
- Led the development of the Newborn Bloodspot Screening National Policy Framework
- Drafted and consulted on a definition of ‘rare disease’ for use by the WA health system
- Conducted focus groups with consumers on their experiences of care coordination
- Contributed to the National Health Genomics Policy Framework

- Partnered on the Better Indigenous Genomics health services project
- Conducted a literature review on self-management for rare diseases
- Mapped existing services for the referral and diagnosis of rare diseases
- Evaluated current referral pathways for rare diseases
- Implemented Patient Archive into the WA public health system
- Led a survey on healthcare experiences of adults living with rare diseases
- Investigated the impact of rare diseases on the WA public health system
- Drafted guidelines for the development of rare disease patient registries

Now heading into the second half of the implementation period for the framework, OPHG is looking forward to progressing the remaining initiatives. Going forward, key priority areas will include investigating best practice delivery of healthcare for rare diseases and the role that a Centre of Expertise in rare diseases might play within the WA health system.
Contributing to a national policy framework on genomics in health

A draft National Health Genomics Policy Framework has been developed to help integrate genomics appropriately into Australia’s health system. When finalised, the framework will guide all levels of government and relevant stakeholders towards a collaborative and coordinated approach in leveraging the potential for genomics to improve the way healthcare is delivered.

The framework was drafted by a working group comprised of representatives from the Commonwealth and all states and territories. Prof Hugh Dawkins, Director of OPHG, was the WA Department of Health representative in this group.

Five strategic priorities have been identified in the framework, where action is required to harness the opportunities genomics can provide in enabling Australians to lead better and longer lives. These relate to a person-centred approach, the workforce, financing, services and data.

Currently, the framework is working its way through the relevant government committees, and it is expected to be publicly released and implemented from 2018 to 2021.

Bridging gaps for rare disease patients

The Undiagnosed Diseases Program WA (UDP-WA) was formed in 2016. It aims to provide a definitive diagnosis for people with complex and long-standing medical conditions that remain undiagnosed despite exhaustive efforts by clinicians.

Conducted from a single paediatric hospital site, the program utilises the interdisciplinary knowledge and diagnostic skills of nearly 40 specialist clinicians to review between one and two cases each month to find a diagnosis for ‘undiagnosable’ patients.

Updates from the UDP-WA

The UDP-WA is currently tailored towards obtaining a diagnosis for children. However, when children with undiagnosed diseases reach the age of 16 years and begin to transition from paediatric to adult care, they are no longer eligible for the program. This creates the need for a UDP within the adult care sector to meet the diagnostic requirements of youths aged 16 to 24 years.

OPHG, in collaboration with the WA Register of Developmental Anomalies and Genetic Services of WA, have engaged Linear Clinical Research to scope the clinical and non-clinical support required for a UDP targeting the transitional patient cohort.

Uncovering the secrets to success

OPHG has also commenced work with a team of organisational psychologists and researchers at the Centre for Transformative Work Design, who are leading a study called Solving the unsolvable: uncovering the active ingredients in the UDP-WA. The overall aim of this project is to uncover the individual, team, organisational and national-level factors that contribute to the success and future sustainability of the UDP-WA.

A key outcome of this collaborative project will be the development of a comprehensive, multi-level framework that provides new insights into the key functions of the UDP-WA. This framework will inform the future development of the program, as well as other initiatives that seek to pioneer new methods of clinical collaboration.

Progressing the WAGHN EAG

Last year, the WAGHN EAG was established to provide strategic leadership for the implementation of the WA Rare Diseases Strategic Framework to help improve the WA health system and outcomes for people living with rare and genetic diseases.

The WAGHN EAG have had two quarterly meetings to date, where the group provided critical insights into a range of key areas including:

(1) reviewing the draft National Health Genomics Policy Framework,
(2) exploring a proposed standard definition of ‘rare disease’ for the WA health system and
(3) evaluating the current referral pathways and diagnostic processes for the diagnosis of rare diseases in WA.
Highlighting the results of an Australian survey on rare diseases

In May, OPHG released a short report summarising the results of the Survey of healthcare experiences of Australian adults living with rare diseases. The survey was led by OPHG together with Genetic Support Network Victoria, Rare Voices Australia, Genetic and Rare Diseases Network and Genetic Alliance Australia.

The report highlights the common experiences that are shared by Australians living with a rare disease, which were identified by survey respondents.

For many of the respondents, obtaining a diagnosis for their disease took a long time (more than five years), and most had to see three or more doctors to receive a diagnosis. Almost half of the respondents had experienced an incorrect diagnosis. The report also includes a checklist of questions that consumers can use to help guide conversations with healthcare professionals when they are first diagnosed with a rare disease.

A little update from OPHG

Earlier in the year, OPHG bid farewell to Karla, who has done a fabulous job developing stellar policy, supporting WA screening programs and leading the Screening Policy Section since its inception five years ago. Thank you and good luck at your new job, Karla!

In July, OPHG recruited Kristen Nowak to lead the Screening Policy Section. The team extend their warmest welcome to Kristen and look forward to working with and learning from her expertise in genetic testing, rare disease research and molecular genetics.

In June, Caroline Walker was seconded to the Environmental Health Directorate to provide additional scientific support. Caroline worked in a small team of experts from the Water Unit, led by Chief Health Officer (CHO) Tarun Weeramanthri, to investigate and resolve issues in the potable water system at Perth Children’s Hospital. The findings of this project are detailed in the CHO’s report.

OPHG is also looking forward to welcoming Trinity back to the team. Trinity is currently on maternity leave and will be re-joining the team later in the year.

More about us

OPHG is a small team that sits in the Public Health Division of the WA health system. Its goal is to develop system-wide and service-specific public policy that optimise the benefits of genomics for the people of WA. To learn more about the team and what they do, please feel free to check out OPHG’s Yearbook or email genomics@health.wa.gov.au.
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