Government of **Western Australia**
Department of **Health**

What an Undiagnosed Diseases Program would do for WA

Rare diseases and the diagnostic odyssey

Rare diseases (RD) are a health priority. They are estimated to affect up to 6–8% of the population which in WA is up to 190,000 people, including more than 60,000 children. Many RD have their onset in childhood, continue for life, and are disabling and burdensome to individuals, families and the healthcare system.

An accurate diagnosis is the bedrock of best practice medical care. For RD achieving a diagnosis is particularly challenging. There are 5,000–8,000 known RD and most are complex with multisystem dysfunction.

Many patients experience a diagnostic odyssey. In a European study, 25% of individuals waited 5-30 years for a diagnosis and in 40% of instances the initial diagnosis was wrong (2). A recent WA lead study showed similar findings (3).

What is an undiagnosed disease?

An undiagnosed disease is a long-standing medical condition for which the health system has been unable to provide a diagnosis.

An Undiagnosed Disease Program for Western Australia

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The health system could say...

- We have more comprehensively addressed the needs of individuals and families living with undiagnosed diseases.
- We can further partner with patients in the development of new management approaches.
- We can benefit from health savings.
- We can be lead partners in global health networks.
- We can further support clinical training and clinical translational research.

Undiagnosed Diseases Network

The Undiagnosed Diseases Program and Network

The Undiagnosed Diseases Program was established within the USA National Institutes of Health (NIH) in 2008. It has become a global network of clinical genetics centres, using multidisciplinary teams, to provide diagnoses for patients with severe undiagnosed diseases.

Many UDN patients have previously visited multiple specialists, have had many hospital admissions and a myriad of investigations.

Diagnosis for those who had none

As a consequence of having no diagnosis, patients, and their families experience anxiety, uncertainty and sometimes inappropriate management of their condition.

In these most diagnostically intractable cases 25% have received a definitive diagnosis. This figure is higher when in children.

Health System Savings

For adult patients, direct costs accrued within the health system prior to assessment by the UDN was estimated to be a minimum of US$ 36,000 (AU$ 49,000) per patient.

With an early, accurate diagnosis much of this cost would have been averted. Future savings will also accrue along with an individuals life span

Preliminary assessments by the UDN suggest that the cost per patient diagnosed is less than a single admission in a tertiary hospital.

Paediatric costings are begin finalised and are anticipated to reveal similar high pre-existing costs and savings opportunities.

Families could say...

- We have closure.
- We are less isolated.
- We better understand what the future might (or might not) hold.
- We have avenues for better treatment, disorder specific medicines or best practice medical care.
- We have improved engagement with the health system.
- We can make financial savings.
- We have improved emotional well being.

Those not receiving a definitive diagnosis could say...

- We have closure for our family.
- The avenues to pursue a diagnosis have been further and more cohesively explored.
- We are less isolated, through connection with the community of undiagnosed individuals e.g. through UDP-related resources and relevant organisations such as Syndromes Without A Name (SWAN) and the Genetic and Rare Diseases Network WA (GaRDN).
- We have improved medical care by integration with relevant services and/ or specialists.
- We can give insight into better management and contribute to the development of new therapies.
- We have improved engagement with the health system.

References

1. Department of Health Western Australia, *WA Rare Diseases Strategic Framework 2015-2018*, 2015.
2. EURORDIS, *Survey of the delay in diagnosis for 8 rare diseases in Europe*, EurordisCare2, 2007.
3. Molster et al., *Health care experiences of adults living with a rare disease in Australia*, In preparation, 2015.
4. Lacbawan et al., Costing the Diagnostic Odyssey: The UDP-NIH Experience