



**Country:** Australia

**Organization:** Genetic Services of Western Australia

**Program/Project:** Genetic Services of Western Australia (GSWA) established the “Rare and Undiagnosed Diseases Diagnostic Service” (RUDDS) in 2013 as a clinically integrated state-wide genomic diagnostic platform to address unmet need for those with undiagnosed rare conditions. It operates in partnership with other public health system managers and service providers, including but not limited to the Office of Population Health Genomics, Diagnostic Genomics (PathWest Laboratories) and with executive level support from the Western Australian Department of Health. It is operating under the WA Rare Diseases Strategic Framework, 2015-2018. A complementary initiative that dovetails with the RUDDS is the Undiagnosed Diseases Program of Western Australia (UDP-WA). This cross-disciplinary approach is directed through GSWA at King Edward Memorial Hospital and it is enabled by the cohesive and combined efforts of multiple paediatric specialties at the Perth Children’s Hospital. Its purpose is to find answers for children with long-standing, very complex, usually multi-system disorders that are undiagnosed despite intensive efforts. Other key partners include the Western Australian Register of Developmental Anomalies; the Telethon Kids Institute, including through its Centre for Precision Medicine in Children; and the Garvan Institute of Medical Research, including Genome.One, in New South Wales. The critical input and support for initiatives to address undiagnosed diseases of Syndromes Without A Name, Australia; the Genetic and Rare Diseases Network, WA; Tea Lake and the Rare Disease Foundation; Rare Voices Australia and others is highly valued. An Australia-New Zealand UDP Executive has been established to further promulgate the UDP in Australia and New Zealand.

The Western Australian Department of Health is an IRDiRC Member Institution

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**Website of GSWA:** [kemh.health.wa.gov.au/services/genetics/](http://kemh.health.wa.gov.au/services/genetics/)