



SPECIAL EDITION FOR RARE DISEASE DAY 2017

Rare Disease Day is celebrated worldwide on the last day of February.

It started 10 years ago on the 29th of February 2008 — a rare day for rare diseases.

People living with a rare disease, patients, politicians, carers, medical professionals, researchers and industry will come together to recognise the importance of rare diseases, to raise awareness and to advocate for a better deal for people with rare diseases.

The 2017 Rare Disease Day theme is **“With Research, Possibilities are Limitless”**. This theme is particularly relevant to all who support and contribute to the APSU. APSU is dedicated to creating new knowledge about rare diseases and using this knowledge to influence policy and practice.



Visit the Rare Disease Day 2017 Australia website to read about Rare Disease Day, local events, patient stories and find out how to get involved www.rarediseasedayaustralia.com.au

Something especially for you on Rare Disease Day!

In our recent survey, many paediatricians told us that they were unsure where to find reliable information about rare diseases and where to refer families affected by rare disease for peer support.

On the next page you will find a brief resource including useful **weblinks to reliable information about rare diseases** including disease summaries, current trials and research.

Please don't forget about psychosocial support for patients and families living with rare disease. Receiving a rare disease diagnosis can be devastating for families. They often feel isolated, under-supported and inadequately informed about their child's rare disease and about accessing appropriate health care and peer support. Some helpful weblinks are listed on the following page.

Rare Disease Resources for Health Professionals and Families

Educational resources for health professionals

Orphanet orpha.net International web portal for rare diseases and orphan drugs. Provides disease summaries, clinical guidelines (if they exist), a support tool for diagnosis, current clinical trials. The portal is searchable by disease name, symptoms, gene etc

OMIM omim.org Online Mendelian Inheritance in Man: enables a comprehensive searchable database of genetic diseases which is searchable by phenotype, clinical features, gene etc

NORD rarediseases.org National Organisation for Rare Disorders: US based site with extensive information, tools and resources for clinicians, researchers and patients/carers

Centre for Genetics Education genetics.edu.au NSW Health web portal for health professionals and patients focussed on genetic conditions; provides fact sheets and guidelines

Australian Genomics Health Alliance australiangenomics.org.au/our-research/rare-disease-flagship research into nationwide implementation of genomic testing

APSU apsu.org.au Australian Paediatric Surveillance Unit: study protocols, case definitions, published research and fact sheets for clinicians and patients/parents and carers



Peer Support Resources

Genetic Alliance Australia geneticalliance.org.au provides peer support and information for individuals and families affected by a rare genetic condition or rare disease; links patients and families with disease specific peer support groups

Rare Voices Australia rarevoices.org.au RVA is Australia's national rare disease alliance supporting all people living with a rare disease, advocating for health policy and healthcare systems

Steve Waugh Foundation stevevaughfoundation.com.au provides support for children and young adults living with the rarest diseases through grants, respite camps, supporting research and raising awareness

RareConnect rareconnect.org/en a world wide platform for rare disease patients and families where they can connect and join or develop online communities across continents and languages

Genetic and Rare Disease Network geneticandrarediseasenetwork.org.au (formerly the Genetic Support Council of WA) works to empower individuals and their families to reach positive health outcomes

ARCAN arcan.org.au Australian Rare Chromosome Awareness Network: an organisation created by parents, for parents and the community; to raise awareness for rare chromosome disorders

SWAN swanaus.com.au Syndromes Without A Name: Information and support for families with a child with an undiagnosed or rare genetic condition

NORD rarediseases.org National Organisation for Rare Disorders: US based site with information, tools and resources for patients/carers, clinicians and researchers

EURORDIS eurordis.org Rare Diseases Europe: Non-governmental world wide patient driven alliance

Australian Paediatric Surveillance Unit

We're on the Web

www.apsu.org.au



Disclaimer

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