GENERAL GENETICS AND RARE DISEASE RESOURCE LIST

ORGANISATION	WEBSITE	TYPE OF RESOURCE- INFORMATION
Centre for Genetics Education P, C	www.genetics.edu.au	Plain English fact sheets for families about genetics and a selection of common genetic disorders (e.g. NF1, CF) Information for health professionals Information about genetic services Publications & resources (including multilingual)
Unique- The rare chromosome disorder support group	www.rarechromo.org	International group, supporting, informing and networking with anyone affected by a rare chromosome disorder and health professionals Plain English fact sheets about chromosome disorders including information directly from families with an affected child
MedGen	http://www.ncbi.nlm.nih.gov/medgen	NCBI's portal to information about human disorders and other phenotypes having a genetic component. MedGen is structured to serve health care professionals, the medical genetics community, and other interested parties by providing centralized access to diverse types of content.
Genetic Home Reference P,C	https://ghr.nlm.nih.gov/	National Library of Medicine Web site for consumer information about genetic conditions and the genes or chromosomes associated with those conditions. Sections include summaries about genetic conditions, genes (including location, function and health implications of variations in the gene), chromosomes and mitochondrial DNA (mtDNA). General information about genetics also available.
GeneReviews C	http://www.ncbi.nlm.nih.gov/books/NBK1116	An international point-of-care resource for clinicians, providing clinically relevant and medically actionable information for inherited conditions in a standardized journal-style format, covering diagnosis, management, and genetic counselling for patients and their families
Find Zebra C	http://www.findzebra.com/	A tool for helping diagnosis of rare diseases, using freely available, high quality curated information on rare diseases and open source information retrieval software tailored to the problem
Genetic and Rare Diseases Information Center (GARD) P,C	rarediseases.info.nih.gov/GARD	Created by the NIH Search by disorder and/or access experienced Information Specialists who provide current and accurate information about genetic and rare diseases in both English and Spanish
Orphanet C	http://www.orpha.net/	Orphanet is the reference portal for information on rare diseases and orphan drugs, for all audiences. English and French

		Rare disease inventory, classification of diseases, encyclopaedia of rare diseases, inventory of orphan drugs, directory of expert resources, assistance to diagnosis tool, and guidelines for emergency medical care and anaesthesia
Online Mendelian Inheritance in Man (OMIM) C	http://www.ncbi.nlm.nih.gov/omim	Comprehensive compendium of human genes and genetic phenotypes that is freely available and updated daily
Undiagnosed Diseases Network C	https://undiagnosed.hms.harvard.edu/	Undiagnosed Diseases Program clinical sites in the US Advocacy organisations Find matching patients Genetic information Research information Parent blog
Personal website of a parent just FYI	http://matt.might.net/articles/rare-disease-internet-matchmaking/	Find matching patients- describes how to use the internet to find a second case for a previously unknown genetic disorder.
Genetic and Rare Disease Network (GaRDN) A	www.geneticandrarediseasenetwork.org.au	Genetic and rare disease support group base in WA See brochure for extent of services: http://www.geneticandrarediseasenetwork.org.au/resources/Updated-GaRDN-Introduction-brochure.pdf Resource handbook at:
Genetic Alliance A	www.geneticalliance.org.au International group: www.geneticalliance.org	Based in NSW but part of a wider international group Facilitates contact between families/individuals affected by the same or similar condition, and/or provide information about relevant support groups both nationally and internationally. Enquiries about services and facilitates ongoing support for individuals, families, and health professionals Extensive rare disease database representing 1400 conditions and over 3500 individuals and families affected by genetic and rare conditions.
Syndromes Without a Name (SWAN) P,A	www.swanaus.com.au	Based in VIC Support, information and advocacy for families with a child who has an undiagnosed condition
Children's Craniofacial Association	http://www.ccakids.com/	Based in the US. Group for individuals and families affected by facial difference- medical, financial, psychosocial, emotional, and educational concerns

P = patient C = clinician A = advocacy

EURORDIS (European Organisation for Rare Diseases)	http://www.eurordis.org/	Advocacy at the European level Patient advocacy, health policy and healthcare services, medicines and therapies, research policy and action, patient empowerment and training, information and networking. Multilingual (European languages)
RareConnect P	www.rareconnect.org	Created by EURORDIS A safe, easy to use platform where rare disease patients, families and patient organizations can develop online communities and conversations across continents and languages. RareConnect partners with the world's leading rare disease patient groups to offer global online communities allowing people to connect around issues which affect them while living with a rare disease.
NORD (National Organisation for Rare Disorders) P	http://rarediseases.org/	US based Patient advocacy, patient and professional education, patient assistance program, mentorship for patient organisations, research support, international partnerships
Rare Voices Australia A	https://www.rarevoices.org.au/	National alliance Leadership, advocacy, education, collaboration and raising awareness at the national level. Works with governments, researchers, clinicians and industry to promote research, diagnosis, treatment and services for all rare diseases in Australia.
NIH/GARD P	https://rarediseases.info.nih.gov/guides/pages/24/tips-for-the-undiagnosed	Guide for parents in seeking information about support, advocacy, clinical trials, research and financial concerns associated with an undiagnosed condition

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