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- I would like to acknowledge that we are here today on the land of the Gadigal and Bidjigal peoples.
- I pay my respects to the Elders, community members and the Aboriginal services and organisations across our network who work to improve health outcomes for Aboriginal and Torres Strait Islander peoples in our Network.

Rare Diseases: a team approach

1. Why think about rare diseases
2. Recognising the pattern
3. Genetic testing 101
4. A diagnosis…what next
5. No diagnosis…what next

Elizabeth.palmer@unsw.edu.au

1. Why think about rare diseases?

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What are the challenges around rare diseases?

There are around 1.2 million people in Australia living with a rare disease – 400,000 of which are children.[1] Up to 80% of these rare diseases are genetic or have a genetic etiology.[2,3,4,5]

Clinical Impact of Rare Diseases

50%
Half of rare diseases impact children[5]

30%
30% of children will not survive beyond the age of 5 years[5]

30%
About 30% of NICU admissions are related to a genetic condition[6]

5-7 Years
For many children, the diagnostic odyssey can last 5-7 years[6-8]

8 Physicians
A patient with rare disease may see an average of 8 physicians[8]

2-3 Misdiagnoses
A patient may receive 2-3 misdiagnoses before receiving a correct diagnosis[8]

1. Department of Health, Government of Western Australia.
2. Bick D, Jones M, Taylor SL, et al. Case for genome sequencing in infants and children with rare, undiagnosed or genetic diseases. Journal of Medical Genetics. Published Online First: 25 April 2019. Doi:10.1136/jmedgenet-2019-106111
3. Global Commission. Ending the diagnostic odyssey for children with a rare disease. Published February 19, 2019.
4. Wakap S, Lambert DM, Orly A, et al. Europ J Hum Genet.2019: https://doi.org/10.1038/s41431-019-0508-0. 7. Ferreira CR. The Burden of Rare Diseases. American Journal of Medical Genetics. 2019;179(6):885-892.
5. Eurodis Rare Diseases Europe. About rare diseases. https://www.eurordis.org/content/what-rare-disease. Updated June 14, 2019.

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At Sydney Children's Hospitals Network

- 180,000 children in NSW
- >2,000 children referred to genetics departments annually across SCHN

Rare diseases- where are we going wrong?

- Paucity of rare disease awareness in health care
- Multisystem diseases requiring multispecialist input
- Poorly coordinated care
- No "home" specialty in the health care system
- Lack of effective treatment or lack of access to effective treatment
- Burden of explanation when accessing the health care system
- Medical professionals having less information than them
- Scepticism about rare diseases within health care due to a lack of awareness

Learn.m4rD.org

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Rare diseases- where do we need to be going?

MEDICS 4 RARE DISEASES

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In an ideal world, a patient with a rare disease would have a

- **timely diagnosis**,
- **mental health support**,
- care in a **specialist centre with excellent communication with their local hospital and GP**,
- and be backed up with the support of a **patient advocacy group**
- and the hope provided by **research opportunities**.

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‘No country can claim to have achieved universal healthcare if it has not adequately and equitably met the needs of those with rare diseases.’

Helen Clark, United Nations Development Programme (2009–2017)

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How can we work together to improve outcomes for rare disease patients?

Australian Genomics Health Alliance

RAREDISEASEDAY.ORG

EURORDIS RARE DISEASES EUROPE

rare voices AUSTRALIA®

NORD® National Organization for Rare Disorders

Undiagnosed Diseases Network

2. Early recognition of the rare disease child

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Case example

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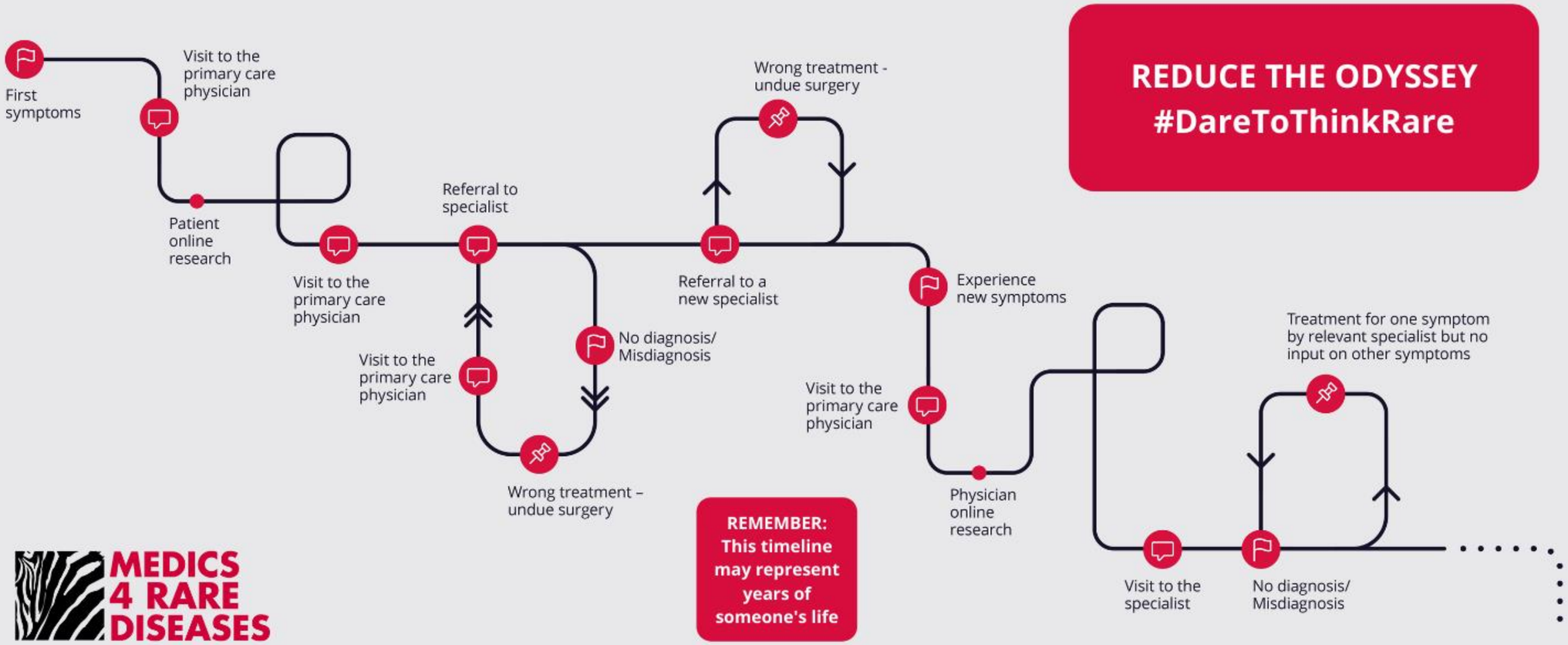
Medical ping pong

The Early Stages of The Diagnostic Odyssey



REDUCE THE ODYSSEY
#DareToThinkRare

REMEMBER: This timeline may represent years of someone's life

MEDICS 4 RARE DISEASES

This image was created, with permission, based on an original image belonging to the Solve-RD project, the EURORDIS-led Community Engagement Task Force (CETF) which sets out the patient journey to diagnosis, published at the European Society of Human Genetics conference. The original infographic demonstrates the diagnostic odyssey many people experience on a daily basis and presents existing resources from CETF member organizations to support patients on this journey.

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Shortening the diagnostic odyssey

Anxiety, frustration

Reproductive implications

Medical 'pilgrimages'

Inappropriate treatments

Loss income

Breakdown of family relationships

- **You cannot learn to recognise over 7000 rare diseases but you can recognise the pattern suggesting that someone might be experiencing the rare disease diagnostic odyssey.**

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Recognise rare disease red flags!

Family (family history)
Group of congenital anomalies
Extreme/ exceptional presentation of common conditions
Neurodevelopmental delay or degeneration
Extreme or exceptional pathology
Surprising laboratory values

- **This can be applied to prompt the question – "Is this a rare disease?"**
- **Family GENES**
- ***Family history: multiple affected siblings or individuals in multiple generations. Remember that lack of a family history does NOT rule out genetic causes.***
- **G: group of congenital anomalies. Common anatomic variations are, well, common; but two or more anomalies are much more likely to indicate the presence of a syndrome with genetic implications.**
- **E: extreme or exceptional presentation of common conditions. Early onset cardiovascular disease, cancer, or renal failure. Unusually severe reaction to infectious or metabolic stress. Recurrent miscarriage. Bilateral primary cancers in paired organs, multiple primary cancers of different tissues.**
- **N: neurodevelopmental delay or degeneration. Developmental delay in the paediatric age group carries a very high risk for genetic disorders. Developmental regression in children or early onset dementia in adults should similarly raise suspicion for genetic etiologies.**
- **E: extreme or exceptional pathology. Unusual tissue histology, such as pheochromoctyoma, acoustic neuroma, medullary thyroid cancer, multiple colon polyps, plexiform neurofibromas, multiple exostoses, most paediatric malignancies.**
- **S: surprising laboratory values. Markedly abnormal pathology results.***

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The Red Flags Working Group of the Genetics in Primary Care (GPC) project created the mnemonic "Family GENES" as a red flag for genetic conditions.

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Baynam, Dr Gareth. (2015). A Diagnostic Odyssey – Red Flags in the Red Sand. AUSTRALIAN MEDICAL ASSOCIATION (WA)..

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Open access tools

Website	Description
Find Zebra	Open access, web-based tool specifically to assist rare disease diagnosis
Online Mendelian Inheritance in Man (OMIM)	The OMIM database is a comprehensive compendium of human genes and genetic phenotypes
Phenomizer	An open access, web-based tool to assist diagnosis. A tutorial is available on YouTube.
Orphanet	Search keywords such as a disease or genes
Google	Please try to use Dr Google only after using these brilliant rare disease tools...

Table adapted from racgo.org.au

FindZebra

Rare Disease Search

cryptorchidism, bleeding problem, short stature, low set ears,

Search

Advanced

Disclaimer: Our website
The search is conducted
FindZebra do not supply
information provided by
direct, incidental, consec
searches, including virus
when you leave the findz

cryptorchidism, bleeding problem, short stature, low set ears,

Search

Diseases (12112) Genes

Advanced

Noonan Syndrome Medlineplus

It is characterized by mildly unusual facial features, short stature, heart defects, bleeding problems, skeletal malformations, and many other signs and symptoms. People with Noonan syndrome have distinctive facial features such as a deep groove in the area between the nose and mouth (philtrum), widely spaced eyes that are usually pale blue or blue-green in color, and low-set ears that are rotated backward. ... Many children with Noonan syndrome have a short neck, and both children and adults may

PTPN11, RAF1, KRAS, SOS1, LZTR1, RIT1, SHOC2,

Related articles

Otofacioosseous-Gonadal Syndrome Omim

The syndrome consisted of sensorineural deafness, short stature, cryptorchidism, inguinal hernia, brachycephaly, prominent forehead, flat face, downslanting palpebral fissures, low nasal root, hypoplastic alae and round tip to the nose, low-set prominent ears, narrow thorax, genu valgum, wormian bones, fusion of carpal bones, delayed bone age, and congenital clubfoot. ... INHERITANCE - Autosomal recessive GROWTH Height - Short stature HEAD & NECK Head - Brachycephaly Face - Prominent forehead - Flat face Ears - Sensorineural deafness - Low set ears - Prominent ears - Posteriorly rotated ears Eyes -

Branchial Arch Syndrome, X-Linked Omim

All 3 showed microcephaly, downslanting palpebral fissures, highly arched palate, apparently low-set, protruding ears, bilateral hearing loss, slightly webbed neck, somewhat short stature, and learning disability. Cryptorchidism was present in 2 and subvalvular pulmonic stenosis and body asymmetry in 1. ... Puri and Phadke (2002) reported a boy with mild mandibulofacial dysostosis, growth retardation with microcephaly, bilateral hearing loss, thoracic deformity with a cardiac valvular lesion, and bilateral cryptorchidism. ... GU - Cryptorchidism Neuro - Learning disability Neck - Slightly webbed neck Inheritance - X-linked

Related articles

Widow's Peak Syndrome Omim

Patients often reported the 'knee giving way' or 'kneecap problems.' Inability to touch the ipsilateral shoulder with the hand was a frequent finding. ... The proband had low-set and posteriorly rotated ears. LaDine et al. (2001) suggested that the patient they described had the same condition as that reported by Kapur et al. (1989). Postnatal onset of short stature, widow's peak, ptosis, posteriorly angulated ears, and limitation of forearm supination was reported in the boy and his mother. ... INHERITANCE - X-linked dominant GROWTH Height - Relative short stature (compared to unaffected males in family) - Short stature,

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AI tools helping you out in the future?

How it works

The MendelScan algorithm captures disease features from electronic health records across a patient population.

Patients are matched to published diagnostic criteria for 100s of rare diseases (and counting).

Mendelian's Clinical Team and Disease Specialists perform an extended medical history review.

Healthcare providers receive a MendelScan report describing the suspected disease, why it's suspected for that patient and the diagnostic pathway.

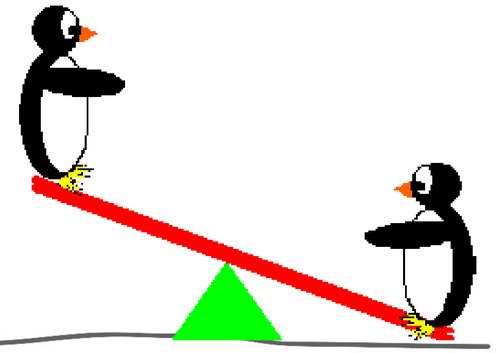
Healthcare providers decide the best way to help each patient by combining their clinical expertise with the novel insights from MendelScan

We've encoded the diagnostic criteria for more than 100 diseases

3. Clinical Genetics 101

- Which test
- When
- How
- HELP!!!!!

Considering.... to test or not to test



Genetic testing can be helpful for:

- **Treatment decisions** – a genetic diagnosis may help the paediatrician or subspecialist select appropriate medication, diet or surgery
- **Clinical trial entry** – certain genetic conditions (will)have access to clinical trials
- **Limit further unnecessary diagnostic testing** – such as neuroimaging or muscle biopsies
- **Accurate genetic counselling** – about the chance of recurrence and options for testing before or around a future pregnancy
- **Patient and family support** – provide an explanation for the child's condition and help connect with other families with the same diagnosis

http://www.pennsw.org.au/clinicians/Genetic-Epilepsy/why-genetic-testing

Chromosomal microarray 101

CHROMOSOMAL MICROARRAY

- This test screens for additional or missing sections of chromosomal material
- It is NOT a screen for all possible genetic conditions
- There are four possible test outcomes:
  - it may provide a **definitive explanation** for the child's epilepsy
  - it may be **non-informative**
  - it may detect a variant in a gene where we are **not sure** if it causes the child's condition
  - it may include an **incidental finding** (that is a genetic variant which causes a condition other than the one we ordered the test for)

RESOURCES

- Centre for Genetics Education
- CMA Testing guide for patient and families
- CMA Factsheets for patients and families
- CMA Review: Chromosome microarray in Australia: A guide for paediatricians (Palmer E, Peters GB. Mowat D; Journal of Paediatrics and Child Health, 48 (2012) E59–E67).
- CMA Review: Chromosome microarray analysis: A soothing guide (Ronan A; Journal of Paediatrics and Child Health, 4 (2018) 599–601).

http://www.pennsw.org.au/clinicians/Genetic-Epilepsy/why-genetic-testing

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The most critical information

What are the possible results of a genomic test?

One or more gene variants are found to explain the condition for which the test was done.

No gene variants are found to explain the condition for which the test was done.

A variant is found in genes associated with the condition but the significance is not known (variants of unknown significance).

A gene variant is found for an unrelated condition (incidental finding).

https://www.genomicsinfo.org.au

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Exome sequencing 101

Journal of Paediatrics and Child Health

doi:10.1111/jpc.15382

VIEWPOINT

Paediatric genomic testing: Navigating medicare rebatable genomic testing

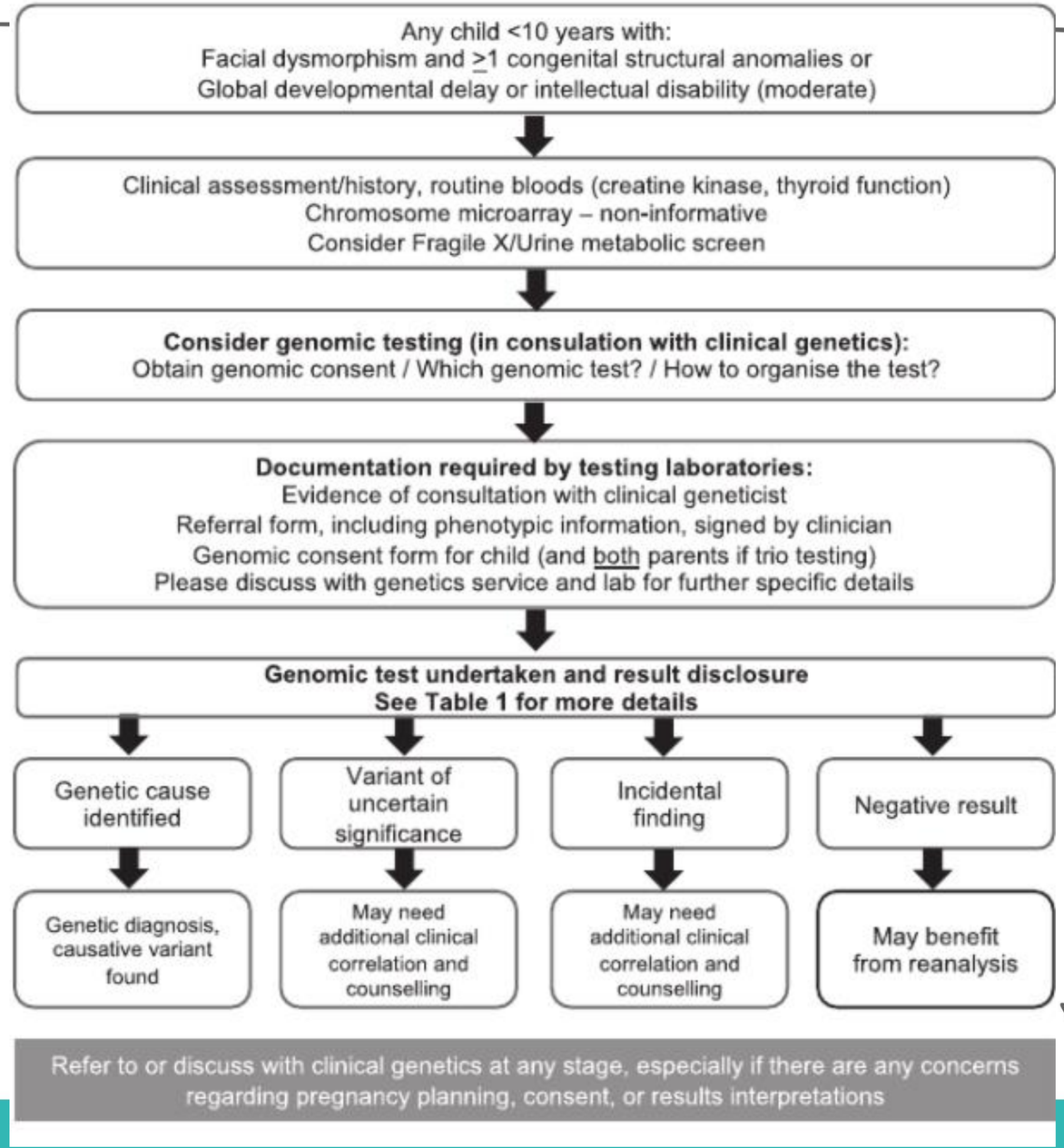
Rani Sachdev,[1,2] Mike Field,[3,4] Gareth S Baynam,[5] John Beilby,[6] Maria Berarducci,[7] Yemima Berman,[8,9] Tiffany Boughtwood,[10,11] Marie B Cusack,[12] Vanessa Fitzgerald,[13] Jeffery Fletcher,[14] Mary-Louise Freckmann,[8] Natalie Grainger,[12] Edwin Kirk,[1,2,15] Ben Lundie,[16] Sebastian Lunke,[17,18] Lesley McGregor,[19] David Mowat,[1,2] Gayathri Parasivam,[12] Vanessa Tyrell,[20] Mathew Wallis,[21,22] Susan M White[17,23] and Alan SL Ma[24,25]

- Medicare funding for exome or genome sequencing **commenced**

  1st May 2020.

- Currently, Medicare funding is limited to 2 circumstances

- It is recommended that the local clinical genetics team be contacted in order to discuss preferred local practice regarding genetics consultation.



Pre-test counselling

Pre-test counselling:

- Genetic testing options are complex **and informed consent is mandatory**.
- It is preferable that counselling is provided by a medical specialist or genetic counsellor.
- Diagnostic laboratories typically require a signed consent form to proceed with testing.
- An **information booklet** should be provided to all families and these important points covered:

Potential outcomes of testing (look familiar?)

The test may:

- it may provide a **definitive explanation** for the child's epilepsy
- it may be **non-informative**
- it may detect a variant in a gene where we are **not sure** if it causes the child's condition
- it may include an **incidental finding** (that is a genetic variant which causes a condition other than the one we ordered the test for)

- **Other:**
- Other issues to be covered in a formal consent process include implications for insurance, sharing results with family members and data and sample sharing.

Resources

- NSW genomic consent document website (on ACI Clinical Genetics Network website)
- NSW genomic supporting document
- National genomic consent document website
- National genomic consent supporting document
- Video from the Centre for Genetics Education - involves a clinician talking through genomic testing and possible outcomes with an adult patient.
- Article JPCH VIEWPOINT Paediatric genomic testing: Navigating medicare rebatablegenomic testing. Sachdev et al., 2021

https://aci.health.nsw.gov.au/networks/clinical_genetics/genetic-and-genomic-testing-consent-forms

4. How to support a family and child after a rare genetic diagnosis?

Prepare
Listen
Signpost
Collaborate

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Advice from rare disease groups

Be Honest

You may not have all the answers, and whilst that is deeply frustrating for healthcare professionals be open and let your patients know when you don't know.

Be Open

Patients and caregivers affected by rare disease often become "Patient Experts" be open to their research and findings. Together you are more effective.

Check In

The early days are a whirlwind and once the dust begins to settle patients will start to have more questions, they will remember things they forgot to ask and they may need more clarification on the things they were told. Scheduling a follow up call in a few days can be a real life line to patients and caregivers during this frightening and confusing time.

Keep your Patients Informed

Silence can be alarming, giving patients regular updates, even to say you are still waiting can go a long way to alleviating anxiety and a sense of being alone felt by most rare disease patients and caregivers.

Think ahead ….Consider 'calling a friend' before a results appointment..

Have your 'go to' quality resources at your fingertips..

Patients and families generally *want to know* as much as possible and can go down confronting and confusing rabbit holes without guidance

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Signposting – knowing where to reach out for help

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Genomic Testing

You are here: Home › Health Professionals › Genomic Testing Consent Resources › The most common ge identified by genomic sequencing

The most common genetic diagnoses identified by ge sequencing

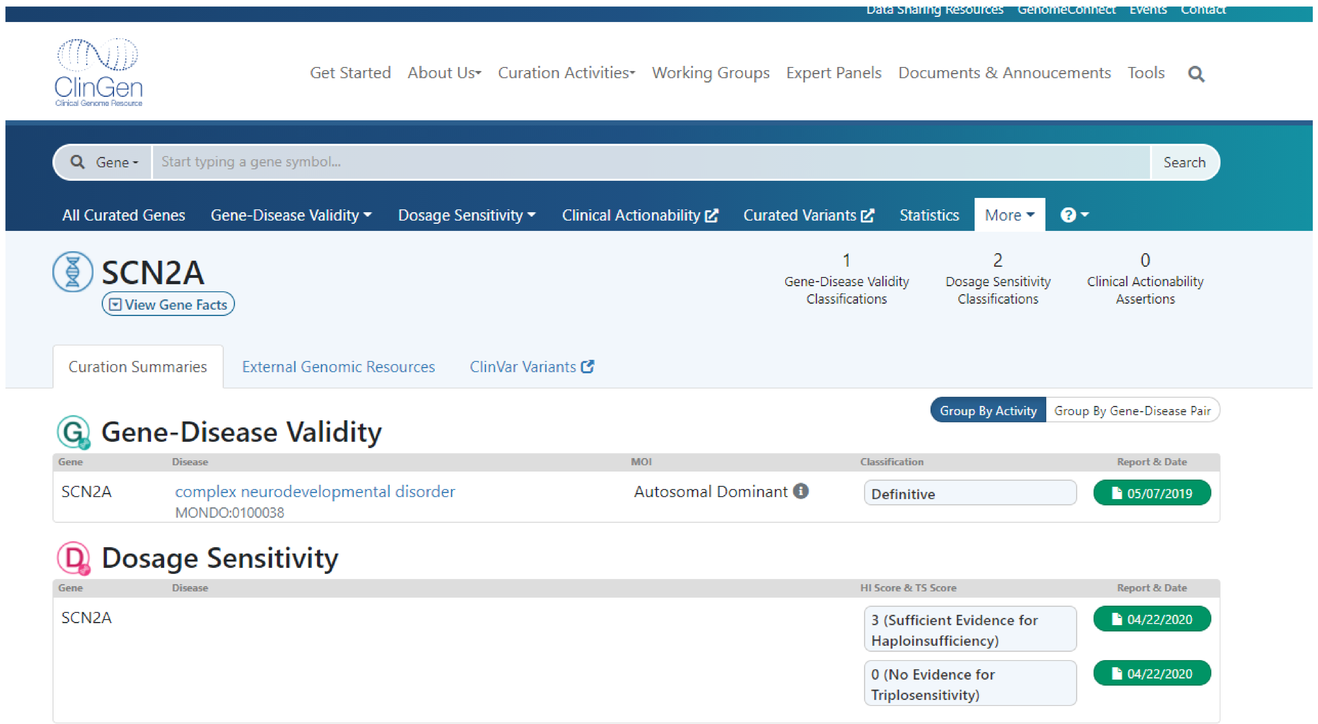
Abbreviations
NORD: National Organization for Rare Disorders
GARD: Genetic and Rare Diseases Information Centre

ALPHABETICAL LISTING

A | B | C | D | E | F | G | H | I | J | K | L | M | N | O | P | Q | R | S | T | U | V | W | X | Y | Z

Gene	Associated condition(s)	General resources	Detailed information
ACTB	Baraitser-Winter syndrome	Unique	GeneReviews
ADNP	ADNP-related multiple congenital anomalies-intellectual disability-autism spectrum disorder	Unique, ADNP Foundation	GeneReviews

NIH: ClinGEN clinicalgenome.org
Type in name of gene .....



How robust is the evidence that this gene is linked to human condition(s)

What types of genetic variants have been proven to be causal?

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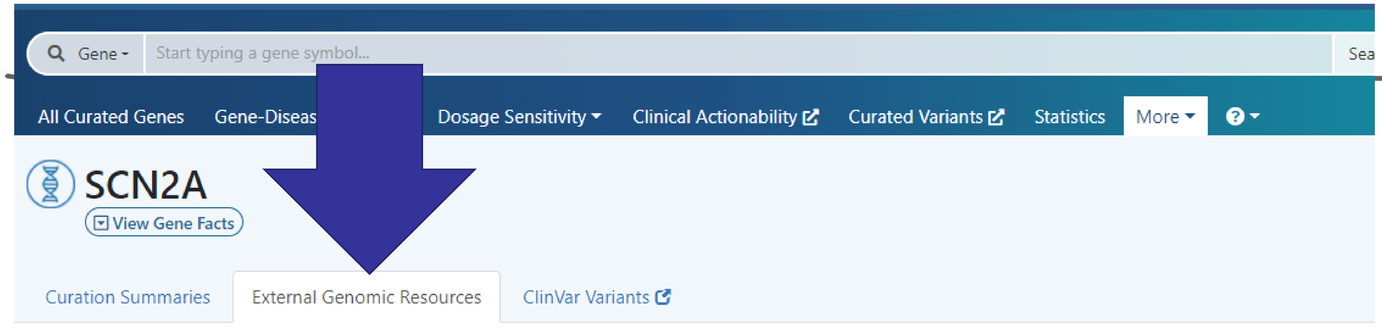

MedGen: Genetics Summary

Organizes information related to human medical genetics, such as attributes of conditions with a genetic contribution.

MedGen: Genetics Summary

Genetic Practice Guidelines: Gene

As guidelines are identified that relate to a disorder, gene, or variation, staff at NCBI connect them to the appropriate records. This page provides an alphabetical list of the professional practice guidelines, position statements, and recommendations that have been identified.

Genetic Practice Guidelines: Gene

Any management guidelines?

GTR: Gene Tests

A voluntary registry of genetic tests and laboratories, with detailed information about the tests such as what is measured and analytic and clinical validity. GTR also is a nexus for information about genetic conditions and provides context-specific links to a variety of resources, including practice guidelines, published literature, and genetic data/information. The scope of GTR includes single gene tests for Mendelian disorders, somatic/cancer tests and pharmacogenetic tests including complex arrays, panels.

GTR: Gene Tests

PharmGKB: Gene

PharmGKB is a comprehensive resource that curates knowledge about the impact of genetic variation on drug response for clinicians and researchers.

PharmGKB: Gene

OMIM: Gene

An Online Catalog of Human Genes and Genetic Disorders.

OMIM: Gene

Gene Reviews

An international point-of-care resource for busy clinicians, provides clinically relevant and medically actionable information for inherited conditions in a standardized journal-style format, covering diagnosis, management, and genetic counseling for patients and their families.

Gene Reviews

Excellent summary for clinicians

ClinVar - Gene

ClinGen and ClinVar are close partners and have established a collaborative working relationship. ClinVar is a critical resource for ClinGen.

NET3524/08

GRIN2A-Related Speech Disorders and Epilepsy

Kenneth A Myers, MD, PhD, FRCPC and Ingrid E Scheffer, FAA, FAHMS, FRACP, MBBS

Author Information

Initial Posting: September 29, 2016.

Estimated reading time: 21 minutes

Summary

Clinical characteristics. *GRIN2A*-related speech disorders and epilepsy are cha
affected individuals and a range of epilepsy syndromes present in about 90%. Se
include dysarthria and speech dyspraxia, and both receptive and expressive lang
affected individuals may display subtly impaired intelligibility of conversational
seizure onset usually between ages three and six years, focal epilepsy with langu
regression, and electroencephalogram (EEG) showing continuous spike-and-wa
centrotemporal discharges. Seizure types include seizures associated with aura c
motor seizures (often evolving to generalized tonic-clonic), and atypical absence seizures. Epilepsy syndromes can include: Landau-Kleffner syndrome (LKS), epileptic encephalopathy with continuous spike-and-wave during sleep

Management

Go to:

Evaluations Following Initial Diagnosis

To establish the extent of disease and needs in an individual diagnosed with a *GRIN2A*-related speech disorder and epilepsy, the following evaluations are recommended:

- Consultation with a speech and language pathologist
- Epilepsy consultation (if not done at the time of initial assessment)
- Sleep-deprived or sleep EEG with monitoring to capture slow-wave sleep (if not done at the time of initial assessment), as this is essential to diagnosing or excluding continuous spike-and-wave in sleep (CSWS).
- Neuropsychological assessment
- Hearing testing
- Consultation with a clinical geneticist and/or genetic counselor

Treatment of Manifestations

Speech/language deficits. Individuals with significant speech/language deficits may benefit from therapy by a speech pathologist. The therapies, which are individualized to the specific speech disorder, often include linguistic approaches and augmentative and alternative communication [Murray et al 2014].

Seizures, if present, should be treated with antiepileptic drugs (AEDs). Many different AEDs may be effective, and no one medication has been demonstrated to be effective specifically for *GRIN2A*-related disorders.

In one individual a good response to refractory epilepsy was achieved with topiramate [Venkateswaran et al 2014].

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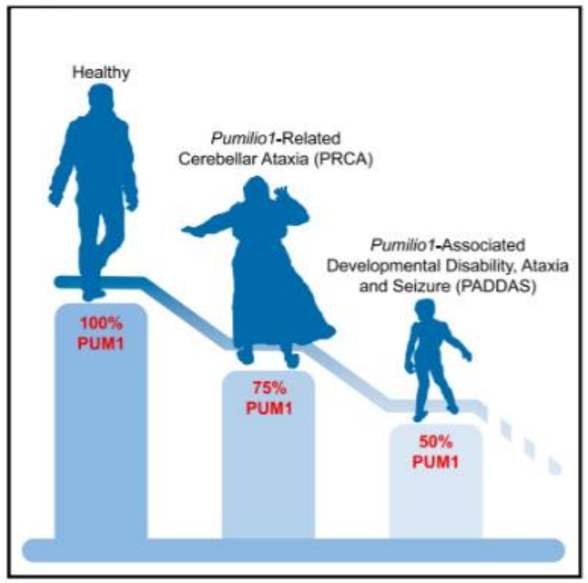
Very new genetic conditions
Human Disease Gene Webseries

Article

Cell

A Mild *PUM1* Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures

Graphical Abstract



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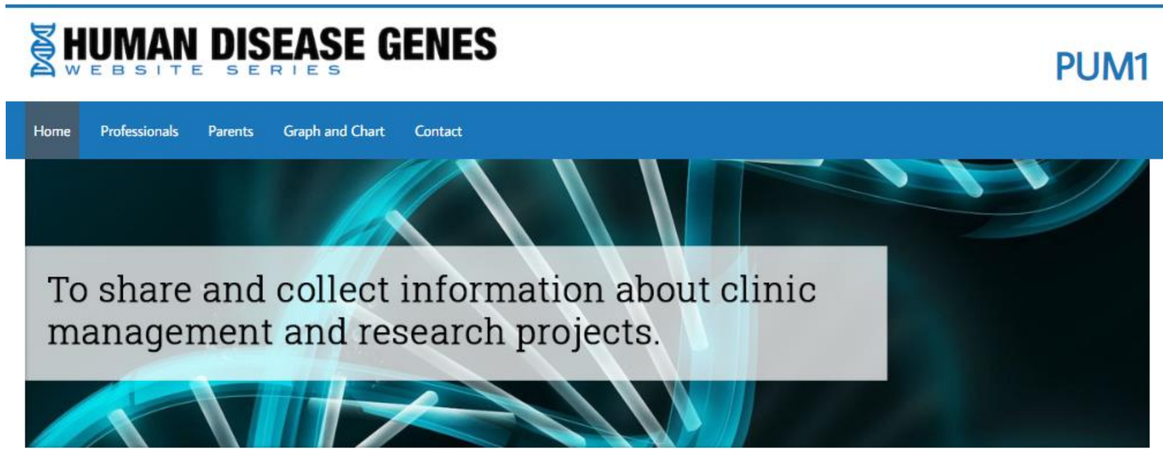
Correspondence

vag2138@cumc.columbia.edu (V.A.G.), hzoghbi@bcm.edu (H.Y.Z.)

In Brief

Different dosages of an RNA-binding protein result in human neurological diseases of corresponding severities.



Welcome

This website provides information on patients with mutations in the PUM1 gene, including clinical data, molecular data, and management and research options.

PUM1 plays an important role in the development and function of neurons (brain cells). When the PUM1 gene does not function properly, due to a change in its DNA sequence (known as a mutation or pathogenic variant), it can lead to a range of neuropsychiatric difficulties, depending on the severity of the mutation.

Currently two distinct PUM1-related disorders are recognised. The more severe disease is an early-onset syndrome called Pumilio1-associated developmental disability, ataxia, and seizures (PADDAS). The features of this disease can vary from one individual to the next.

A milder PUM1 mutation has been found in one family with a slowly progressive, adult-onset ataxia. This disease is called Pumilio1-related cerebellar ataxia, or PRCA.

This website was created to share and collect information about clinical and research projects on PUM1 and to gather more knowledge about patients with mutations in PUM1 with the goal of developing better treatments.

A/Professor Vincenzo Alessandro Gennarino, PhD, Department of Genetics and Development, Columbia University Medical Canter, New York, USA, vag2138@cumc.columbia.edu

Dr (Elizabeth) Emma Palmer (Clinical Geneticist), MD, Genetics of Learning Disability Service, Hunter Genetics, Waratah, NSW 2298, Sydney, Australia, Elizabeth.palmer1@health.nsw.gov.au

A/Professor Jimmy Lloyd Holder, Jr., MD (Pediatric Neurology), Department of Pediatrics, Baylor College of Medicine, Houston, TX 77030, USA, holder@bcm.edu

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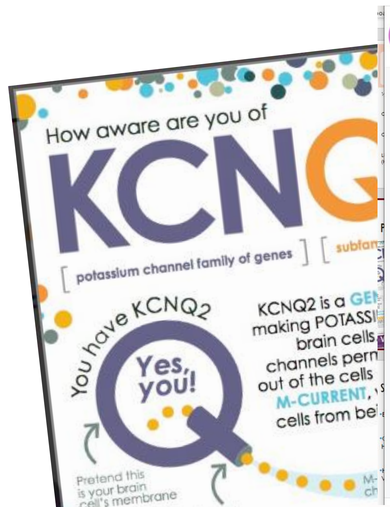
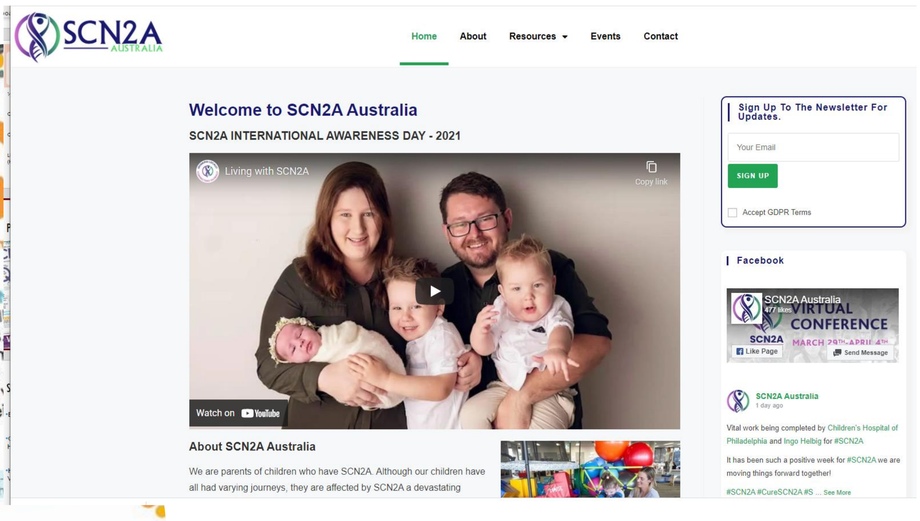
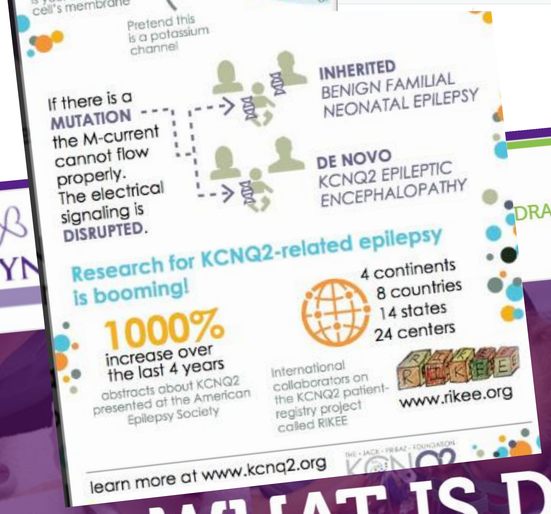
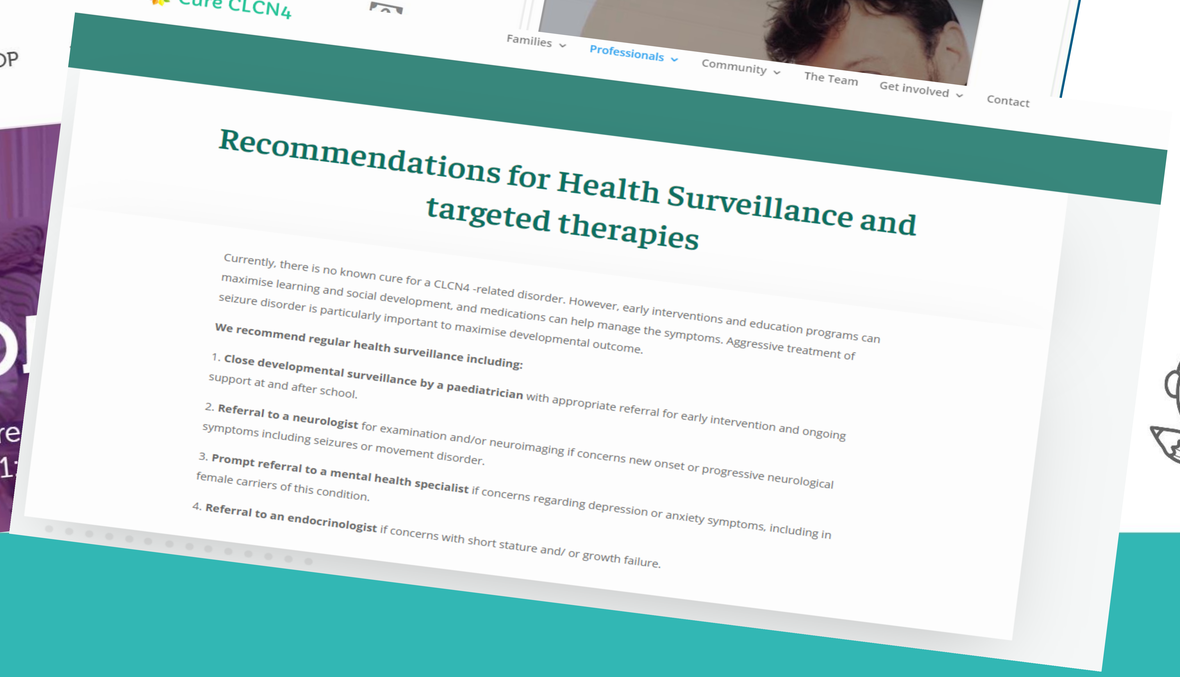
Parent led resources

Google









Due diligence

- Q: Who is on their clinical / scientific advisory board?

Scientific Advisors

EMMA PALMER

Dr Palmer (PhD, MBBS, FRACP, BA (Hons I) Oxon), is a clinician scientist at Sydney Children's Hospital Network & University of New South Wales in Sydney, Australia.
She has extensive experience at the interface of clinical and research genetics leading multidisciplinary teams and establishing international collaborations to discover new genetic conditions. She has led 5 international projects delineating novel genetic conditions (ZSWIM6, ATN1, ARV1, KCNT2, RLIM duplication). She works closely with rare genetic disease advocacy and consumer reference groups and aims to translate genomic discoveries to improved education and management for patients and families. She was the first author on a publication describing the impact of CLCN4 gene changes in 52 individuals, moderates the CLCN4 gene pages on the Human Disease Gene Webseries along with Professor Vera Kalscheuer and is leading the clinical aspects of an international study to better understand the genetic and clinical spectrum of CLCN4 related condition.

Q: Are they a member of a country's rare disease peak body ?

Click each logo to visit their site.

EURORDIS RARE DISEASES EUROPE · GENETIC ALLIANCE UK · Global Genes Allies in Rare Disease · RARE REVOLUTION MAGAZINE · GENETIC DISORDERS UK educate · inspire · empower · SWAN UK · orphanet

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A

Alpha-1 Organisation Australia Inc

Acrodysostosis Support and Research

Acute Necrotising Encephalopathy

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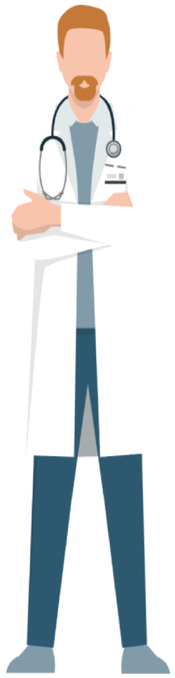
Collaborate

Try to work to coordinate appointments and locations as much as you can.

Advocate for multi-disciplinary meetings so all relevant health care professionals can discuss and plan treatment in a coordinated effort and avoid the need for constant re-telling.



MEDICS 4 RARE DISEASES

Learn.m4rD.org

Integrated Care at SCHN

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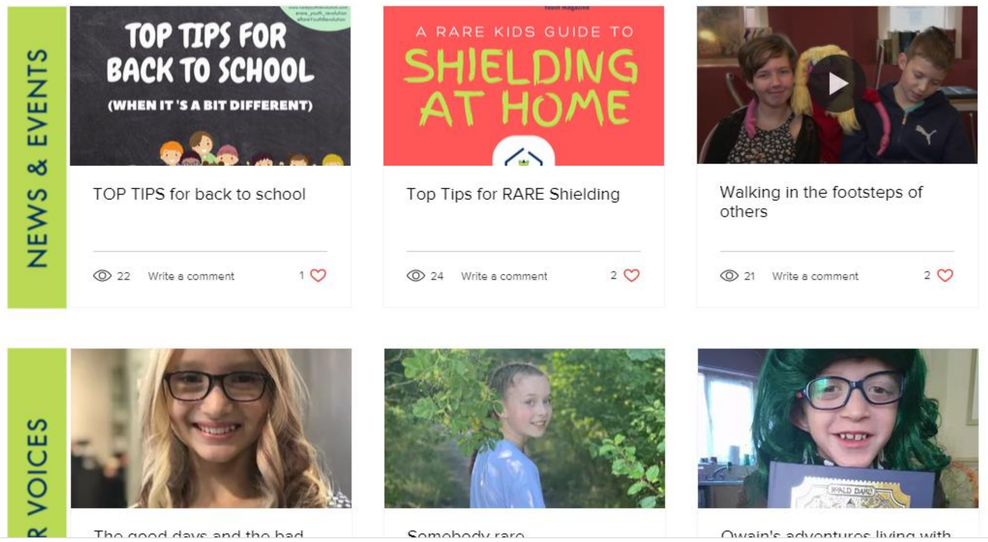
Support for the WHOLE family

https://www.reframingdisability.com.au/

http://siblingsaustralia.org.au/

Powering up the voices of the youth rare disease community



https://www.rarerevolutionmagazine.com/rarevouthproject.html

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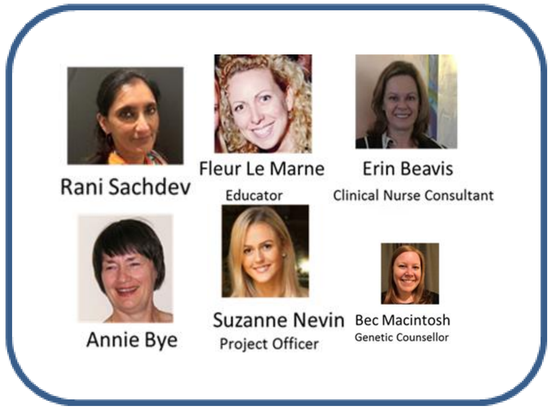
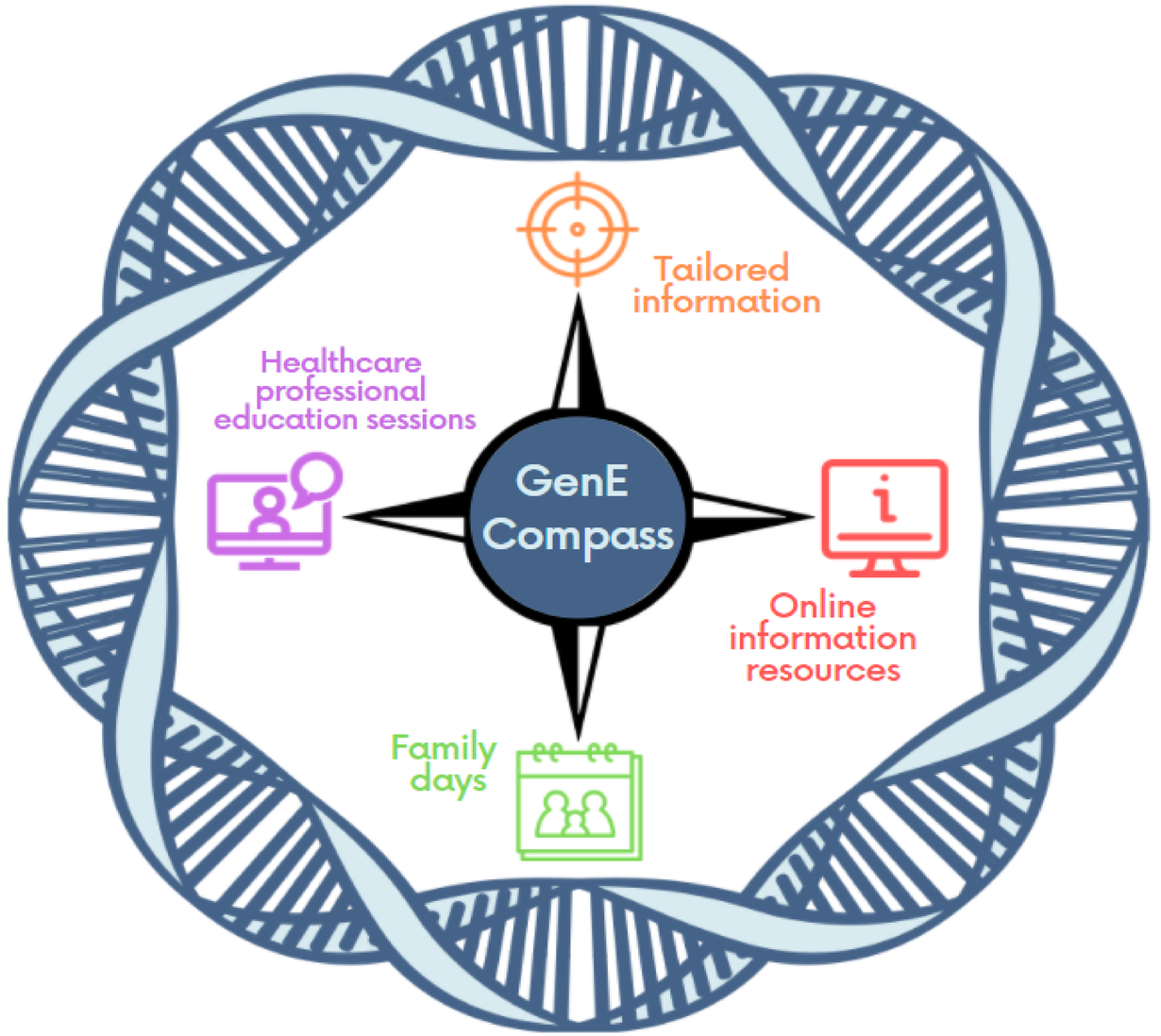
Genetic Epilepsy Pilot





GEM OF THE WEST MANILDRA GROUP 100% AUSTRALIAN OWNED

5. How to support a child and family who remains on the diagnostic odyssey?

NET3524/0816

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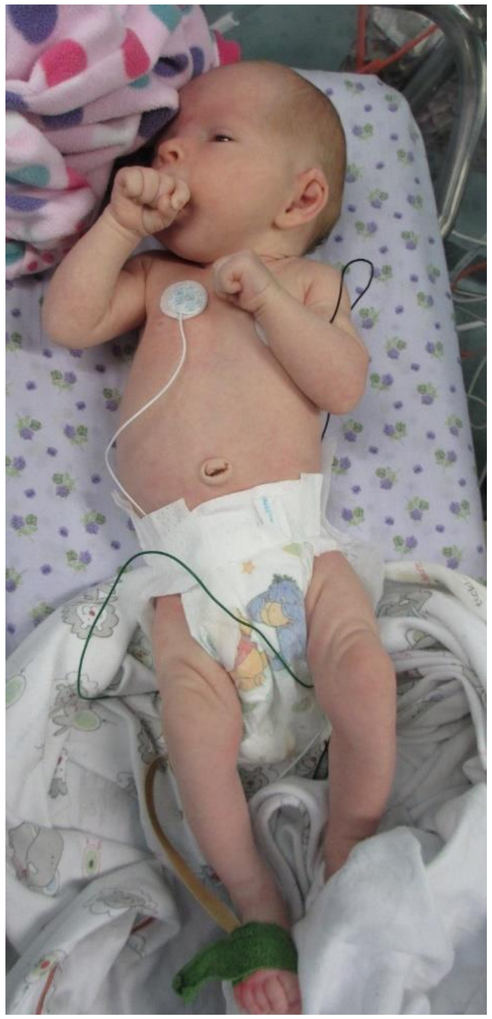
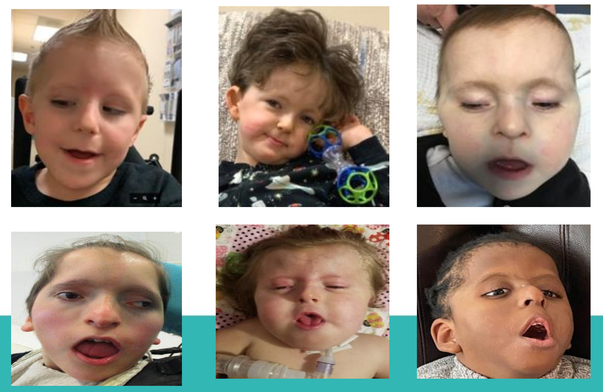
Case study



REPORT

De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome

Elizabeth E. Palmer,[1,2,3,4,25] Seungbeom Hong,[5,25] Fatema Al Zahrani,[6] Mais O. Hashem,[6] Fajr A. Aleisa,[5] Heba M. Jalal Ahmed,[5] Tejaswi Kandula,[1,2] Rebecca Macintosh,[1] Andre E. Minoche,[3] Clare Puttick,[3] Velimir Gayevskiy,[3] Alexander P. Drew,[3] Mark J. Cowley,[3,7] Marcel Dinger,[3,7] Jill A. Rosenfeld,[8] Rui Xiao,[8,9] Megan T. Cho,[10] Suliat F. Yakubu,[5] Lindsay B. Henderson,[10] Maria J. Guillen Sacoto,[10] Amber Begtrup,[10] Muddathir Hamad,[11] Marwan Shinawi,[12] Marisa V. Andrews,[12] Marilyn C. Jones,[13] Kristin Lindstrom,[14] Ruth E. Bristol,[15] Saima Kayani,[16] Molly Snyder,[17] María Mercedes Villanueva,[18] Angeles Schteinschnaider,[18] Laurence Faivre,[19,20] Christel Thauvin,[19] Antonio Vitobello,[19] Tony Roscioli,[1,21,22] Edwin P. Kirk,[1,2,21] Ann Bye,[1,2] Jasmeen Merzaban,[23] Łukasz Jaremko,[5] Mariusz Jaremko,[23] Rani K. Sachdev,[1,2] Fowzan S. Alkuraya,[6,24,25,*] and Stefan T. Arold[5,25,*]



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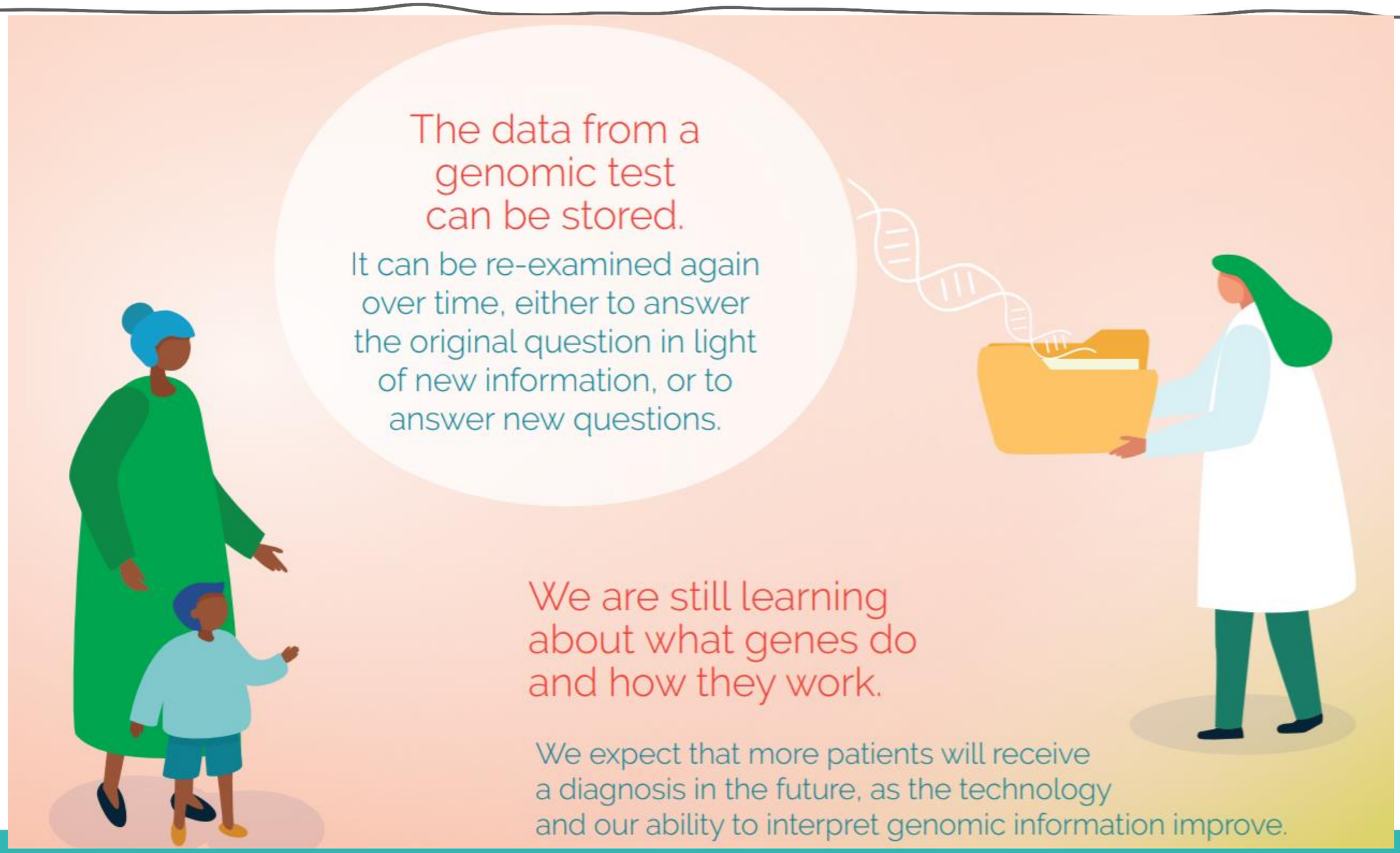
Genetic testing may need to be revisited….



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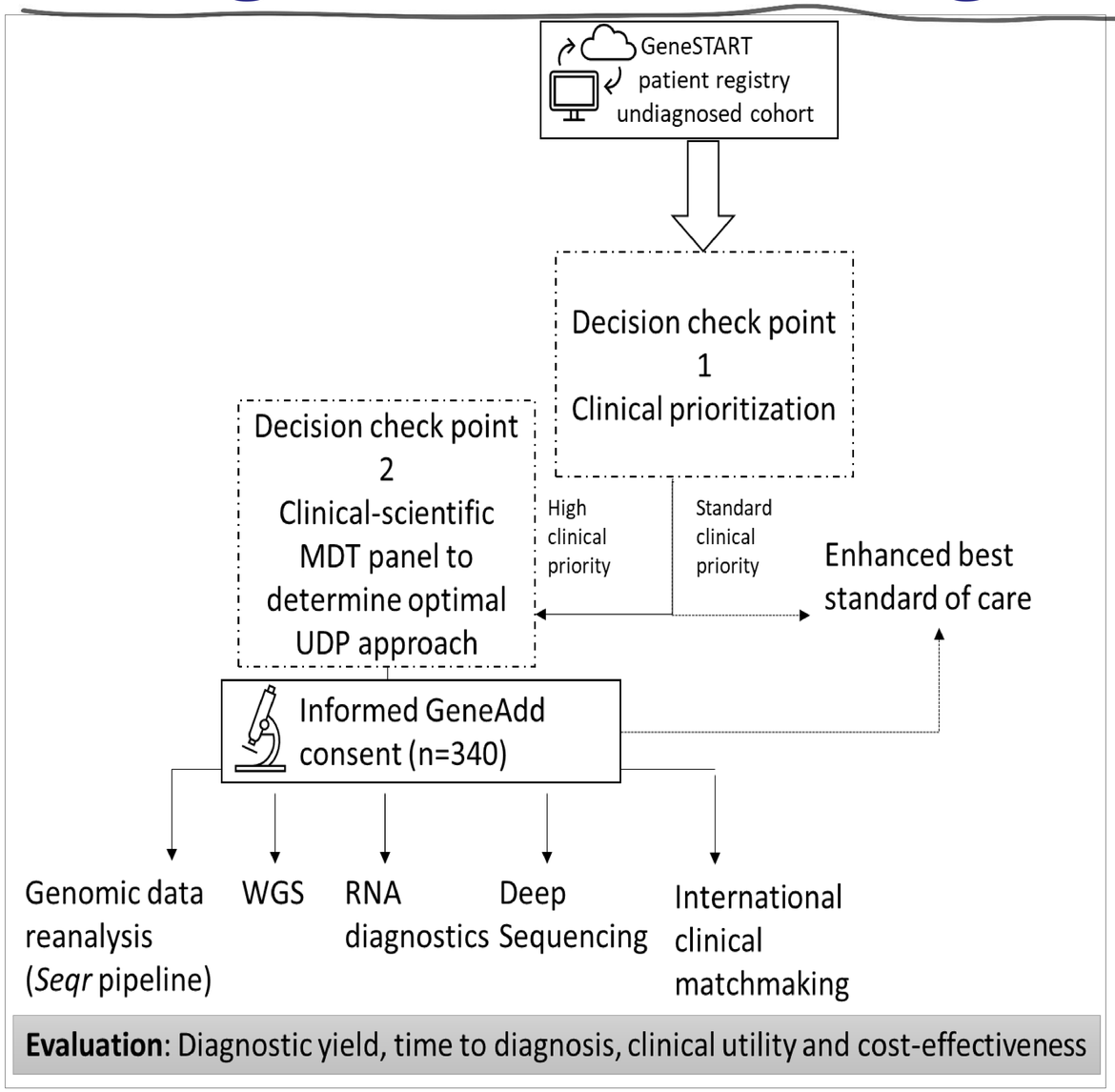
Undiagnosed Diseases Network

Undiagnosed disease programs



Gene2Care incorporating GeneSTART and GeneAdd

- Syndromes without a Name

https://swanaus.org.au/

https://swanaus.org.au/

Summary

- Every clinician can be part of a collaboration to improve the model of care for rare disease children and their families.
- Fabulous resources available through the international website Medics 4 Rare Disease https://learn.m4rd.org/
- Centre for Genetics Education a great first point of call.

#Daretothinkrare

- Listen
- Think (and think, and think again)
- Support diagnosed and undiagnosed families
- Signpost resources
- Work as a team

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UNSW SYDNEY

Short Course

Practical Medical Genomics

The UNSW Medicine and Science faculties have a rich history in building the future of the health industry. Our leading educators, researchers and clinicians have translated discoveries into breakthrough cures, public health strategies, policies and turned high achieving students into leaders of healthcare with globally recognised qualifications.

Who should attend this course?

Clinicians and clinicians-in-training who are interested in learning about clinical genomics and precision medicine. Relevant professions include pediatricians, obstetricians, general physicians, sub-specialists, and general practitioners.

Gain knowledge and skills to integrate genomics into your daily practice

About this course

This course will provide medical practitioners with the specialised knowledge and skills required to integrate genetics and genomics into their daily practice with confidence. This includes the ability to:

- Identify which patients will benefit from genomic testing
- Interpret genomic test result reports
- Develop individual care plans and communicate with patients and families affected by genetic conditions; and
- Identify when to consult with clinical genetics and genomics services.

Course format

Interactive, case- based learning opportunities will be delivered by clinical geneticists and medical specialists who are actively using genomics in their clinical practice. The course will include online learning resources, self-assessment and reflection tasks and live, virtual discussion sessions.

Delivery and assessment

DELIVERY

Fully online via AGSM Virtual Learning Environment.

30 hours of learning, divided between online synchronous teaching, case-based discussions, and presentations (12 hours – split over two weekend days) and asynchronous learning (18 hours) that can be fitted around practitioners' own clinical practice.

The course is delivered in partnership with the faculties of UNSW Medicine and Health, and UNSW Science.

ASSESSMENT

There is an optional assessment associated with this course. All participants who complete this assessment task will be issued with 3 FMECH points upon completion of the course. These points can be credited towards future UNSW postgraduate programs.

Course details

DATES

The course will run over a 6-week period with online course starting 10th August 2021. Live sessions are scheduled for the weekends of 28 August and 11 September.

LOCATION

Online – AGSM Virtual Learning Environment

COST

$2500

POINTS

Faculty of Medicine Executive Certificate in Health (FMECH) points: 3 per course

Contact

Dr Elizabeth Emma Palmer

Dr Emily Oates

clinicalgenetics@unsw.edu.au

> More details

> Register

CRICOS Provider Code 00098G

Elizabeth.palmer@unsw.edu.au