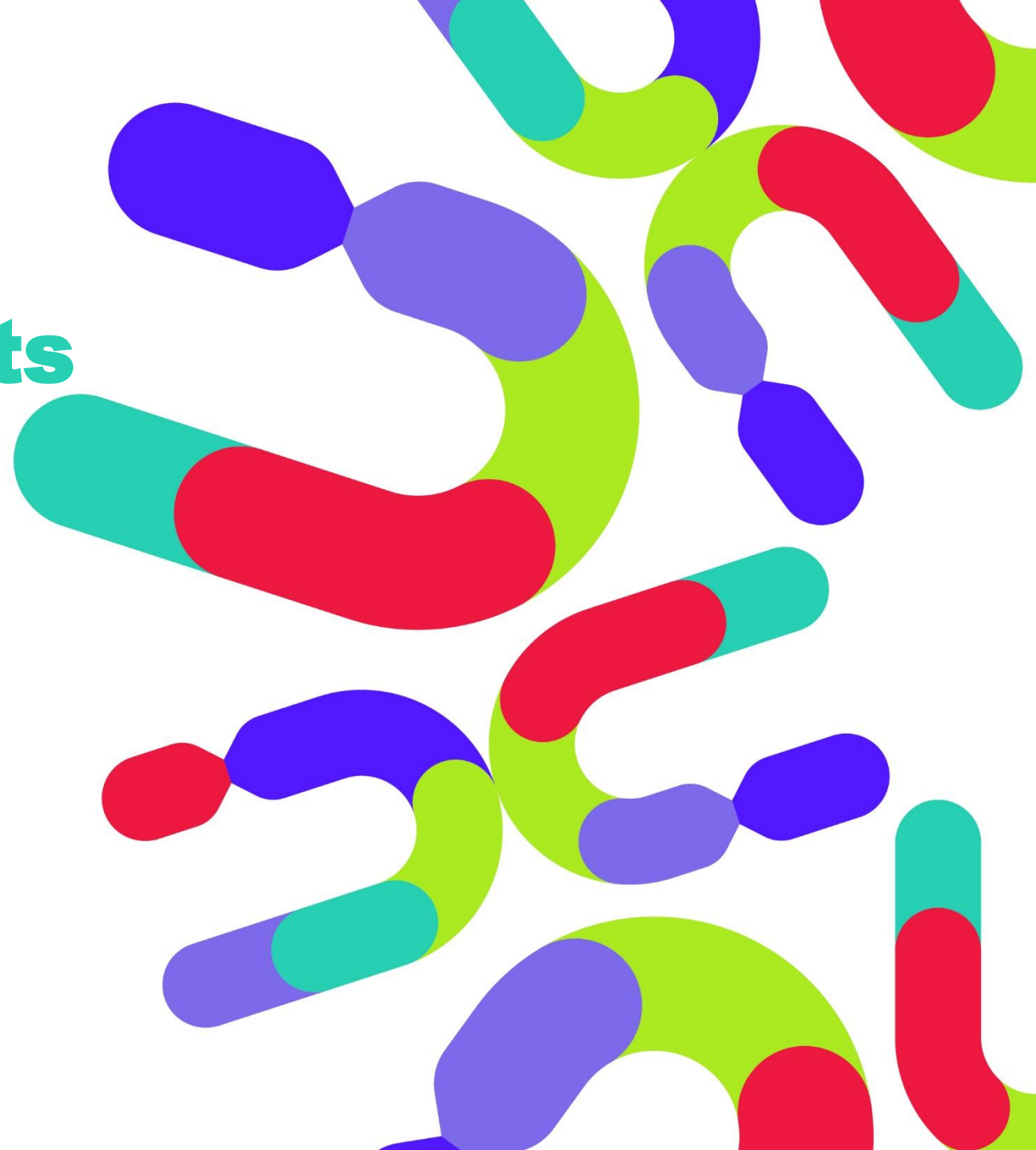


Unique: Resources for Patients and Families

Sarah Wynn, PhD
CEO
sarah@rarechromo.org



Who are Unique?

- All those affected by rare chromosome or gene disorders associated with developmental delay/intellectual disability
- UK-based charity supporting >35,000 families globally
- Since 1984



HELPLINE

Our team offer phone, messaging and email support to families and professionals worldwide.



INFORMATION HUB

We produce information guides for rare genetic conditions as well as practical guides, children's picture booklets and more.



MATCHING SERVICE

We connect families with similar genetic conditions, symptoms or geographic locations. We facilitate connections whilst also providing support.



WEBSITE

All information is free to access on our website. You can also join us, donate and visit our online shop there too.



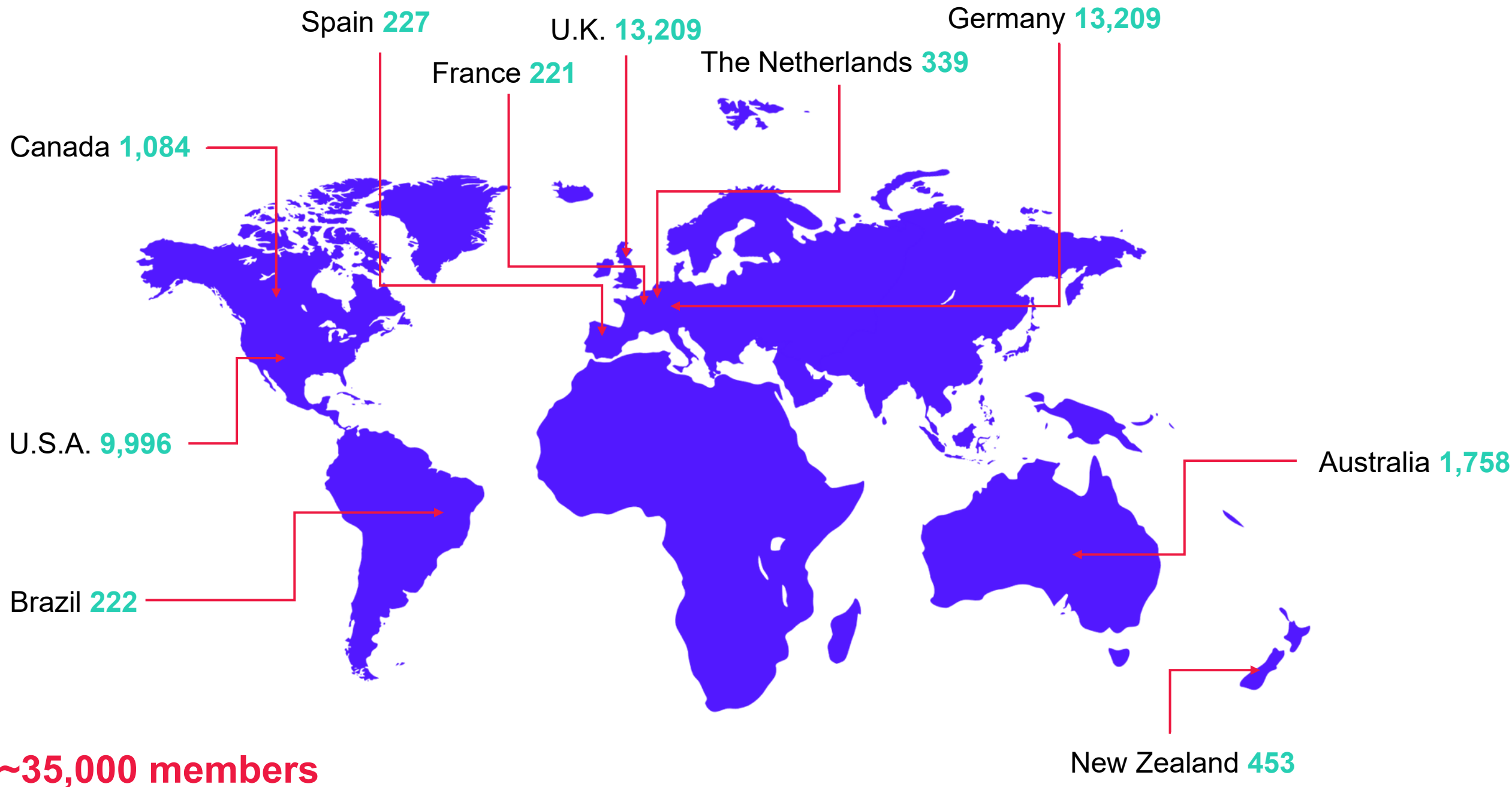
ADVOCACY

We contribute to research, participate in conferences, and collaborate with others in our field.



COMMUNITY BUILDING

We bring people together through events, webinars and our Facebook group.

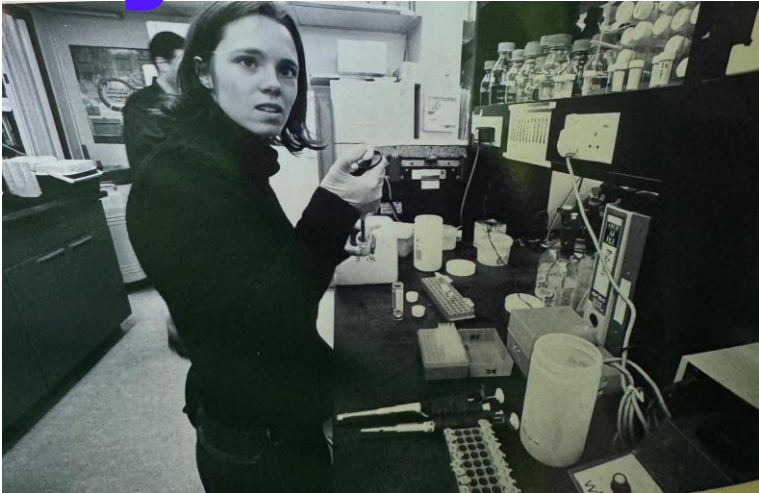


~35,000 members

Who are Unique?



My route to Unique



- Postdoc-ing at HKU, 2003

PNAS RESEARCH ARTICLE DEVELOPMENTAL BIOLOGY



SOX9 and SOX10 control fluid homeostasis in the inner ear for hearing through independent and cooperative mechanisms

Irene Y. Y. Szeto¹, Daniel K. H. Chu^{1,2}, Peikai Chen^{1,2}, Ka Chi Chu^{1,3}, Tiffany Y. K. Au⁴, Keith K. H. Leung⁵, Yong-Heng Huang^{6,7}, Sarah L. Wynn⁸, Angel C. Y. Mak^{4,9}, Ying-Shing Chan⁴, Wood Yee Chan^{4,5}, Ralf Jauch^{4,10}, Bernd Fritzsche⁴, Mai Har Sham^{4,5}, Robin Lovell-Badge⁴, and Kathryn S. E. Cheah^{1,6}

Edited by Stefan Mundlos, Max Planck Institute for Molecular Genetics, Berlin, Germany; received December 23, 2021; accepted September 10, 2022 by Editorial Board Member Denis Duboule

The *in vivo* mechanisms underlying dominant syndromes caused by mutations in SRY-Box Transcription Factor 9 (*SOX9*) and *SOX10* (*SOXE*) transcription factors, when they either are expressed alone or are coexpressed, are ill-defined. We created a mouse model for the campomelic dysplasia *SOX9*^{Y440X} mutation, which truncates the transactivation domain but leaves DNA binding and dimerization intact. Here, we find that *SOX9*^{Y440X} causes deafness via distinct mechanisms in the endolymphatic sac (ES)/duct and cochlea. By contrast, conditional heterozygous *Sox9*-null mice are normal. During the ES development of *Sox9*^{Y440X} heterozygotes, *Sox10* and genes important for ionic homeostasis are downregulated, and there is developmental persistence of progenitors, resulting in fewer mature cells. *Sox10* heterozygous null mutants also display persistence of ES/duct progenitors. By contrast, *SOX10* retains its expression in the early *Sox9*^{Y440X} mutant cochlea. Later, in the postnatal stria vascularis, dominant interference by *SOX9*^{Y440X} is implicated in impairing the normal cooperation of *SOX9* and *SOX10* in repressing the expression of the water channel Aquaporin 3, thereby contributing to endolymphatic hydrops. Our study shows that for a functioning endolymphatic system in the inner ear, *SOX9* regulates *Sox10*, and depending on the cell type and target gene, it works either independently or cooperatively with *SOX10*. *SOX9*^{Y440X} can interfere with the activity of both *SOXE* factors, exerting effects that can be classified as haploinsufficient/hypomorphic or dominant negative depending on the cell/gene context. This model of disruption of transcription factor partnerships may be applicable to congenital deafness, which affects ~0.3% of newborns, and other syndromic disorders.

Significance

Impairment of the endolymphatic system is a common cause of deafness. Our study of a mouse model of campomelic dysplasia, a syndrome characterized in part by deafness of unknown etiology, reveals the roles of *SOX9* and its relative *SOX10* in maintaining a functioning endolymphatic system. Early in its development, these *SOXE* factors are critical for regulating the differentiation of progenitors and the expression of ion transporters and channels essential for ionic homeostasis. Later, they control the expression of *Aqp3*, a water/glycerol channel.

PNAS RESEARCH ARTICLE DEVELOPMENTAL BIOLOGY



Hypomorphic and dominant-negative impact of truncated SOX9 dysregulates Hedgehog-Wnt signaling, causing campomelia

Tiffany Y. K. Au^{1,2}, Raymond K. H. Yip^{1,2,3}, Sarah L. Wynn⁴, Tjong Y. Tan^{4,5}, Alex Fu^{1,6}, Yu Hong Geng^{4,7}, Irene Y. Y. Szeto^{1,8}, Ben Niu^{1,9}, Kevin Y. Yip^{1,10}, Martin C. H. Cheung¹¹, Robin Lovell-Badge¹², and Kathryn S. E. Cheah^{1,13}

Edited by Clifford Tabin, Harvard Medical School, Boston, MA; received June 10, 2022; accepted November 2, 2022

Haploinsufficiency for *SOX9*, the master chondrogenesis transcription factor, can underlie campomelic dysplasia (CD), an autosomal dominant skeletal malformation syndrome, because heterozygous *Sox9* null mice recapitulate the bent limb (campomelia) and some other phenotypes associated with CD. However, *in vitro* cell assays suggest haploinsufficiency may not apply for certain mutations, notably those that truncate the protein, but in these cases *in vivo* evidence is lacking and underlying mechanisms are unknown. Here, using conditional mouse mutants, we compared the impact of a heterozygous *Sox9* null mutation (*Sox9*^{Y440X}) with the *Sox9*^{Y440X} CD mutation that truncates the C-terminal transactivation domain but spares the DNA-binding domain. While some *Sox9*^{Y440X} mice survived, all *Sox9*^{Y440X} mice died perinatally. However, the skeletal defects were more severe and HH signaling in developing limb cartilage was significantly enhanced in *Sox9*^{Y440X} compared with *Sox9*^{Y440X}. Activating *Sox9*^{Y440X} specifically in the chondrocyte-osteoblast lineage caused milder campomelia, and revealed cell- and noncell-autonomous mechanisms acting on chondrocyte differentiation and osteogenesis in the perichondrium. Transcriptome analyses of developing *Sox9*^{Y440X} limbs revealed dysregulated expression of genes for the extracellular matrix, as well as changes consistent with aberrant WNT and HH signaling. *SOX9*^{Y440X} failed to interact with β -catenin and was unable to suppress transactivation of *Ihh* in cell-based assays. We propose enhanced HH signaling in the adjacent perichondrium induces asymmetrically localized excessive perichondrial osteogenesis resulting in campomelia. Our study implicates combined haploinsufficiency/hypomorphic and dominant-negative actions of *SOX9*^{Y440X}; cell-autonomous and noncell-autonomous mechanisms, and dysregulated WNT and HH signaling, as the cause of human campomelia.

Significance

SOX9 heterozygous mutations cause skeletal malformation and bent bones of Campomelia Dysplasia. The cause of bent bones and whether similar molecular mechanisms apply *in vivo* for all heterozygous mutations in *SOX9* are unclear. In this study, we generated a mouse equivalent of the human CD *SOX9*^{Y440X} mutation, which truncates the C-terminal transactivation domain. We discovered, unlike the null mutation, heterozygosity for *Sox9*^{Y440X} exerts cell-autonomous and noncell-autonomous dominant-negative effects on

SOX9 | campomelic dysplasia | osteoblast differentiation | skeletal disorders |

From the Unique Balanced Translocation guide:

46,XY,t(5;7)(q33;p22)

Comment

A male karyotype with an apparently BALANCED RECIPROCAL TRANSLOCATION, between the long arm of one chromosome 5 at breakpoint q33 and the short arm of one chromosome 7 at breakpoint p22.]



The father has a balanced translocation. This family had three miscarriages before having three children: two with the balanced translocation (daughter, left, and son) and one with ordinary chromosomes (daughter, right).

The *Unique* Challenge

Can you match the face to the diagnosis?

Each set of pictures shows individuals with a recently delineated microdeletion syndrome, a well-known chromosomal syndrome or a fairly common chromosome deletion or duplication.

1



2



3



4



5



Please fill in your answers overleaf

info@rarechromo.org



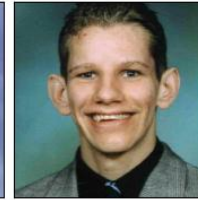
www.rarechromo.org

The *Unique* Challenge

Can you match the face to the diagnosis?

Each set of pictures shows individuals with a recently delineated microdeletion syndrome, a well-known chromosomal syndrome or a fairly common chromosome deletion or duplication.

6



7



8



5 1p36 deletion syndrome 7 9q34.3 deletion

1 3q29 deletion 2 17q21.31 microdeletion

4 Trisomy 8 mosaicism 3 22q13 deletion syndrome

6 9p duplication 8 Ring 22

The families and individuals shown here have given consent for their picture to be published and fully support *Unique's* aims to raise awareness of rare chromosome disorders and to enhance the life prospects of everyone affected by them.

info@rarechromo.org



www.rarechromo.org

Name:.....
Institution:.....
Address:.....
Email:.....

You can find more information and images of children and adults with rare chromosome disorders in *Unique's* disorder-specific leaflet series.

Unique's records of over 6000 affected people show the natural history, lifetime effects and real consequences of living with a rare chromosome disorder. Relating the story from the family's perspective, these records confirm, refine and expand what is known about rare chromosome disorders and can challenge assumptions

Rare Conditions



After diagnosis: Living with a rare genetic condition

1 in 17

people are affected by a rare condition at some point in their lifetime



A rare condition affects fewer than

1 in 2000

There are over

7000

rare conditions, with new conditions regularly identified through scientific progress



7 in 10

rare conditions affect children. And more than

3 in 10

children with a rare condition die before their fifth birthday



Diagnosis with a rare condition comes with specific challenges

- Lack of prognostic information
- Lack of a pathway to follow
- Struggle for recognition
- Lack of knowledge – public and professionals
- Challenges finding support
- Lack of research
- Lack of evidence-based treatments

Confused. A bit bewildered and just left really

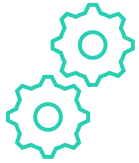
What Unique can Offer Families



Information & signposting



Information guides



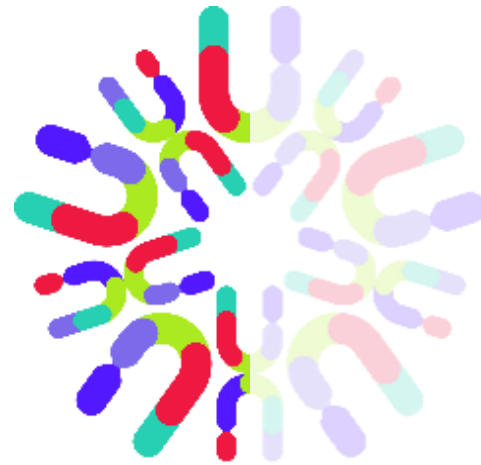
Practical guides



Unique database



Regular magazines



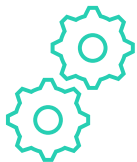
unique

UNDERSTANDING GENES
& CHROMOSOMES

Information & signposting



Information guides



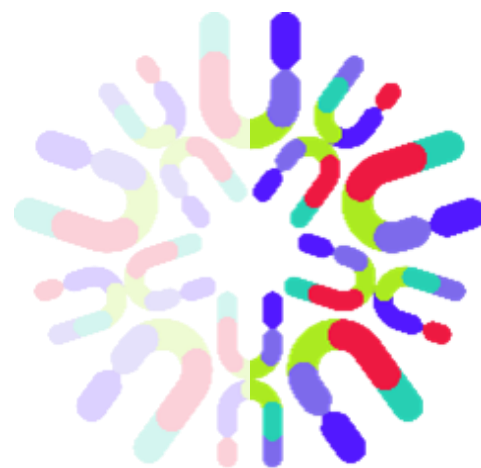
Practical guides



Unique database



Regular magazines



unique

UNDERSTANDING GENES
& CHROMOSOMES

Emotional support

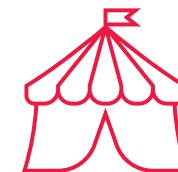
Family matching service



Helpline



Family days & events



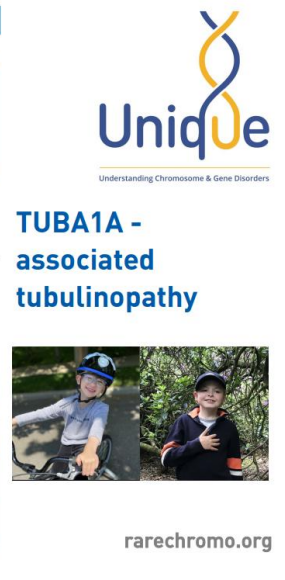
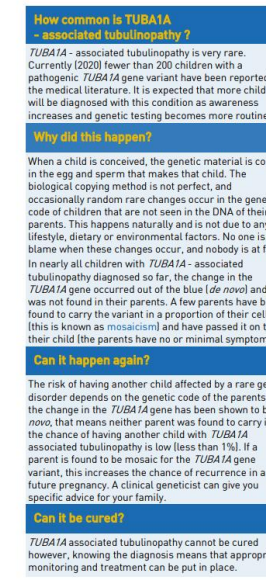
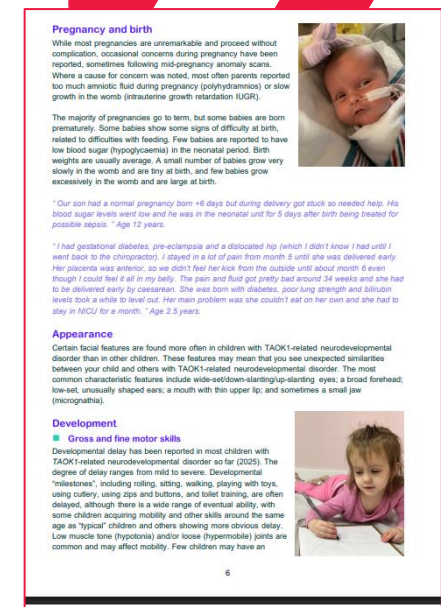
Community & belonging

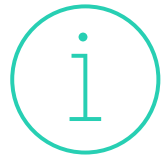


Unique Guides for Families and Professionals

- Information guide project launched in 2004. Information often focused on the “worst case scenario” or was written in complex, medical terms that weren’t easily accessible to families.
- Over 300 guides to genetic conditions including over 80 guides to single gene conditions
- Developed in collaboration with clinical and academic experts
- Over 550 translations across 22 languages

<https://rarechromo.org/disorder-guides/>





Information guides



Clear overview of what might happen over time



Practical suggestions and tips based on real experiences



Reassurance that they are not alone



Validation



Useful tool when working with professionals

The Challenge

With a small team, how can we keep up information guide production?

- ~300 guides but ~800 conditions
- Could AI be a solution?



Guides in the pilot project

Published

- TUBB3- related tubulinopathy
- QRICH1 (Ververi Brady syndrome)
- BUB1B (MVA) Mosaic Variegated Aneuploidy
- ERCC6 (Cockayne syndrome)
- ZNF699 (DEGCAGS)
- PPP2R5D, PPP2R1A, PPP2CA, PPP2R5C (Houge Janssens syndrome)
- KCNK9 - Birk Barel syndrome
- ODC1 (Bachmann Bupp syndrome)

In progress

- HERC1
- BCL11A (Dias Logan syndrome)
- KPTN-related disorder
- PACS2-related syndrome

[The guide] is so incredibly informative and lovely. This looks wonderful. We truly appreciate your hard work on making this such a comprehensive guide. Thank you for all of your work on putting this together!! What a gift this will be for our community!



**Bachmann-Bupp
syndrome (BABS)**

rarechromo.org

Thank you so much for sharing the completed guide. I was truly excited to see [photos of my son] on the cover and in other sections. I'm so happy that I was able to contribute something to help complete the guide for other families and healthcare professionals. Thank you for giving me the opportunity to be part of this project. We are always happy to help in any way

The guide looks amazing. Thank you for all your hard work!



**KCNK9 imprinting
syndrome (KIS)**
(Birk-Barel syndrome (BIBARS))

rarechromo.org

[The] guide looks absolutely fabulous. Thank you for putting this together. It is going to be an incredibly valuable resource for the community



**BCL11A intellectual
developmental disorder
(BCL11A-IDD)**
(Dias-Logan syndrome)

rarechromo.org

*Wow, this looks wonderful!!
I'm so glad so many families
contributed!*

*Once again, a huge thank
you for this remarkable
piece of work. We are truly
delighted to have been able
to contribute to this project,
and we would like to
congratulate you on the
quality of the guide. We are
sure it will be an incredibly
valuable resource for
families and professionals
affected by Dias-Logan
syndrome.*

Chromosome	Syndrome/condition name	Gene	Members (updated end of Sept 2026)
18q21.2	Pitt Hopkins syndrome	TCF4	14
2q23.1	MBD5-associated neurodevelopmental disorder (MAND).	MBD5	13
14q12	FOXG1 syndrome	FOXG1	11
Xp11.23	WDR45-related disorders	WDR45	8
13q34	Gould Syndrome	COL4A1	8
2q24.3	Dravet Syndrome	SCN1A	8
5q35.3	Sotos syndrome	NSD1	8
1p36.33	GNB1 Encephalopathy	GNB1	8
2p23.3	Tatton-Brown-Rahman syndrome or Heyn-Sproul-Jackson syndrome	DNMT3A	7
11p15.1	Epilepsy, progressive myoclonic 7	KCNC1	6
1q42.11-q42.12	Skraban-Deardorff syndrome	WDR26	6
1p35.3	Xia-Gibbs syndrome	AHDC1	6
Xp22.12	Coffin-Lowry	RPS6KA3	6
1p36.23	RERE-Related Disorders	RERE	5
6p21.33	CSNK2B-related NDD or Poirier-Bienvenu neurodevelopmental syndrome (POBINDS)	CSNK2B	5
22q12.1	CEBALID syndrome (MN1 C-terminal truncation (MCTT) syndrome)	MN1	4
18q21.31	NARS1-related neurologic disorders	NARS1	4
3q13.31	Primrose syndrome	ZBTB20	4
9p24.3	Nicolaides-Baraitser syndrome	SMARCA2	4
17p11.2	Smith-Magenis syndrome (SMS)	RAI1	3
3q26.33	SOX2 anophthalmia syndrome	SOX2	3
Xq13.2	Allan-Herndon-Dudley syndrome	SLC16A2 (MCT8)	3

Chromosome	Syndrome/condition name	Gene	Members (updated end of Sept 2026)
15q11.2	Schaaf-Yang syndrome	MAGEL2	3
4p16.3	Muenke syndrome	FGFR3	3
Xq22.2	PLP1-related disorders	PLP1	3
12p13.31	ATN1-related NDD	ATN1	3
8q24.22	Seizures, benign neonatal, 2	KCNQ3	3
2p13.2	Beck-Fahrner syndrome	TET3	2
16q21	Vissers-Bodmer syndrome	CNOT1	2
14q32.2	Gabriele-de Vries syndrome	YY1	2
1q42.12	Bryant-Li-Bhoj syndrome	H3-3A / H3F3A	2
3q21.1	ADCY5-related movement disorder	ADCY5	2
2q34	UNC80 deficiency	UNC80	2
Xp22.2	Opitz GBBB syndrome	MID1	2
9q22.33	Loeys-Dietz syndrome 1	TGFBR1	2
3p14.3	The FLNB-related disorders	FLNB	2
Xq23	DCX-related disorders	DCX	2
Xq26.3	Christianson syndrome	SLC9A6	2
17q25.1	TSEN54 pontocerebellar hypoplasia	TSEN54	2
18q21.33	Fryns syndrome	PIGN	2
Xq28	FLNA deficiency and FLNA-related otopalatodigital (FLNA-OPD) spectrum disorders	FLNA	2
7q36.1	EZH2-related overgrowth	EZH2	2
2q14.2	RNU4atac-opathy	RNU4ATAC	2

Would you like to get involved?

- Please get in touch!
- sarah@rarechromo.org



<https://www.smartsurvey.co.uk/s/G7ZCCS/>

Building the evidence: Sharing stories and experiences



Unique's Little Red Book

<https://rarechromo.org/little-red-book/>



Laith

15q25.2 deletion
UK

Laith has suffered with GERD (gastroesophageal reflux disease) from 9 weeks old and by 2 years old was diagnosed with failure to thrive as a result. With 3 specialists across 3 hospitals the decision was taken to conduct genetic testing. We were told that in all likelihood nothing would show.

In April 2024 we received the confirmation that he has a 2Mb deletion on 15q25.2 and as quickly as we felt relief at the diagnosis, we felt lonely and isolated at the lack of knowledge held by any of his professionals.

The inevitable Google searching that we all do led us to the Unique website and that was when things changed and we, as a family, began to feel informed and empowered.

THE SUPPORT ON UNDERSTANDING GENETICS IN LAYMAN'S TERMS MEANT WE COULD GO TO APPOINTMENTS AND REALLY FEEL PART OF THE CONVERSATION

The guides provided us with an overview of the deletion that all of a sudden made everything about Laith make sense. The support on understanding genetics in layman's terms meant we could go to appointments and really feel part of the conversation, comfortable to question and challenge for the first time.

We are now in a position where Laith is supported in mainstream school. He is now fed through a G-Tube. He is developmentally delayed and short in stature, however he tackles every challenge and every day without hesitation and is a source of joy to those around him.

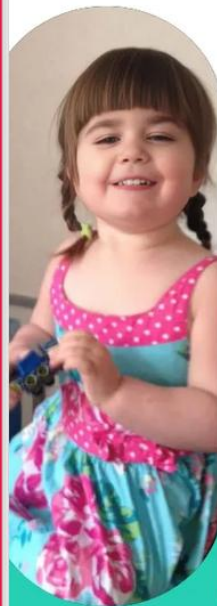
We will be forever grateful to the Unique team and community for giving us the information, support and power to be able to help everyone involved in Laith's life and care to support him to be the best version of himself!

Unique connections

21

Michelle's daughter

NTRK2 associated syndrome
UK



I have 3 children, I suffered multiple miscarriages, and my youngest daughter was my last pregnancy (number 10). Cassie was born in a planned C-section which had complications and delayed her birth. She showed a multitude of delays including failing newborn hearing, having visual maturation delay, delay in every milestone and didn't speak until she was 3 and a half years old. Her knee made a very loud clicking and clunking noise each time she used it with weight bearing, but this was dismissed by paediatricians. She was unable to crawl and walking was the only thing that wasn't significantly delayed.

SHE WAS UNABLE TO CRAWL AND WALKING WAS THE ONLY THING THAT WASN'T SIGNIFICANTLY DELAYED

They suspected she was autistic at 2 years old but this wasn't diagnosed until she was 8 years old. She had suffered with severe asthma. She had fine motor and gross motor delays, which ended up being symptomatic of dyspraxia/DCD. She had signs of learning disability with problems storing things she learnt and she would have to over learn (be taught several times before it went in).

I battled with school to get any form of input for her, having to do my own Education Health and Care Plan (EHCP) with help from other parents and my own knowledge. During this time a physiotherapist finally listened to me about my daughter's knee and referred her for surgical opinion. She was diagnosed with a congenital abnormality in her knee. When we next

Journey to diagnosis

saw the respiratory doctor, he tested for basic genetic conditions especially as her BMI was increasing. These being negative prompted a referral to genetics and she was finally diagnosed after whole genome sequencing was able to be performed (it was newly available). This showed 2 mutations of unknown significance, the first being dismissed due to not enough evidence, but the second was looked into thoroughly with a family history taken and other family members being tested.

It was discovered that not only did she have it but that it was present in her father, aunt and grandfather. All of whom had a degree of learning problems, hyperphagia and ASD. She also has sleep apnoea due to other issues.

DESPITE HIS STRUGGLES HE IS THRIVING

Louise's son

16p11.2 distal microdeletion
UK

My son has been diagnosed with 16p11.2 distal microdeletion. He had many delays through his milestones. He is now 4 years of age and, despite his struggles, he is thriving and such a happy boy.



OLLIE WAS TESTED FOR GENE DISORDERS DUE TO HIS COMPLICATIONS

Ollie

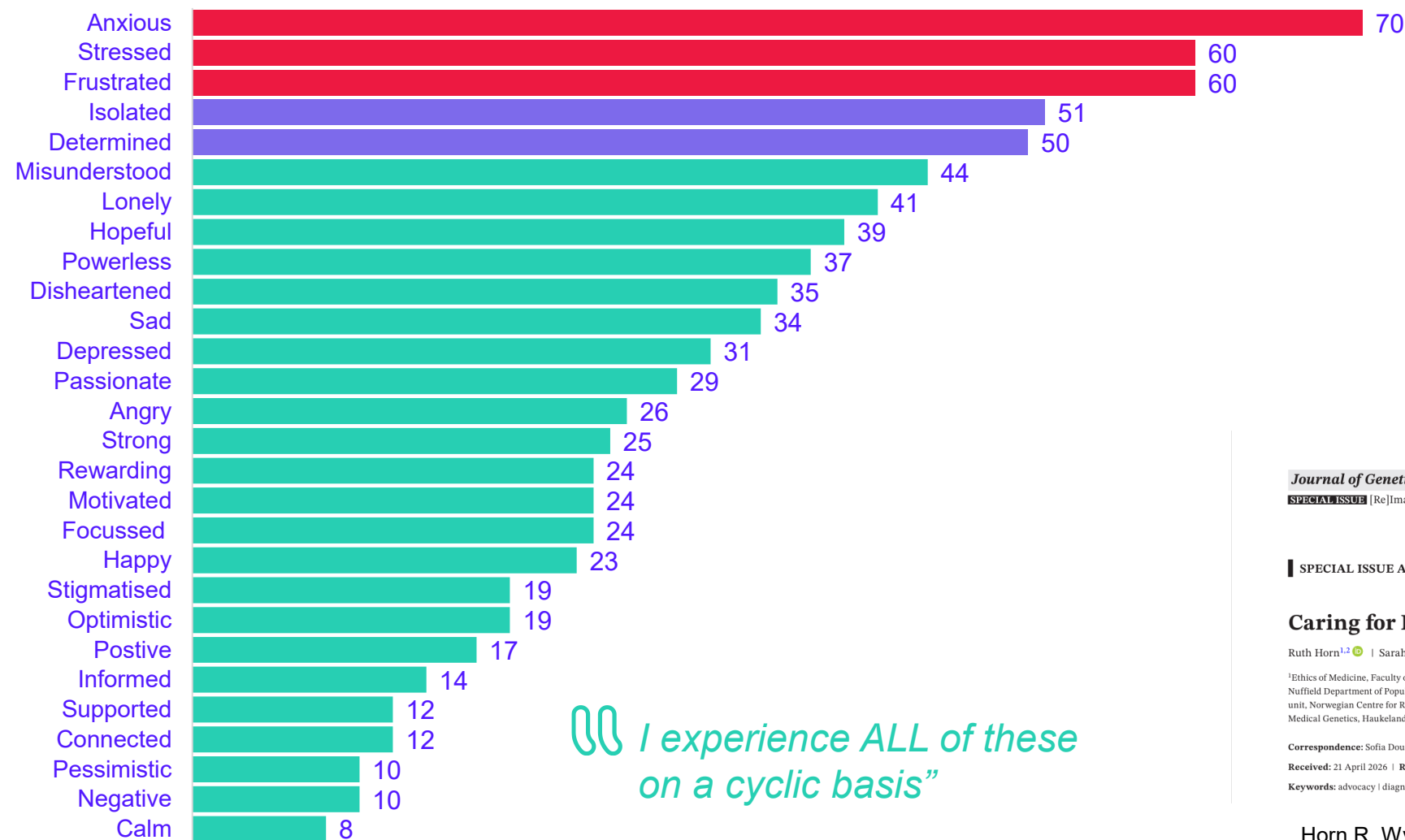
16p11.2 microduplication
UK

I, I'm mum to 7 children. My youngest, Ollie, was tested for gene disorders due to his complications, feeding, choking, global developmental delay (GDD), gastroesophageal reflux disease (GERD), silent reflux, not meeting his milestones, speech delay, always poorly and respiratory problems too.



The emotional impact of a rare diagnosis

“From the list of words below, which best describe your experiences of having, or caring for someone with, a rare chromosome or gene disorder?”



*I experience ALL of these
on a cyclic basis”*

Top responses

- 70% Anxious
- 60% Stressed
- 60% Frustrated
- 51% Isolated
- 50% Determined

Journal of Genetic Counseling

SPECIAL ISSUE [Re]Imagining the Future of Genetic Counseling

WILEY | National Society of Genetic Counselors

SPECIAL ISSUE ARTICLE OPEN ACCESS

Caring for Rare Genetic Disease: A Vision for the Future

Ruth Horn^{1,2} | Sarah L. Wynn³ | Sofia Douzgou Houge^{4,5,6}

¹Ethics of Medicine, Faculty of Medicine, Institute for Ethics and History of Health in Society, University of Augsburg, Augsburg, Germany | ²Ethox Centre, Nuffield Department of Population Health, University of Oxford, Oxford, UK | ³Unique, Rare Chromosome Disorder Support Group, Oxted, UK | ⁴Frambu unit, Norwegian Centre for Rare Diseases, Siggerud, Norway | ⁵Brain disorders unit, Norwegian Centre for Rare Diseases, Oslo, Norway | ⁶Department of Medical Genetics, Haukeland University Hospital, Bergen, Norway

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Received: 21 April 2026 | Revised: 11 August 2026 | Accepted: 24 August 2026

Keywords: advocacy | diagnosis | education | engagement | genomic medicine | healthcare | people living with a rare disease

Unique's Survey Report

- 1) Lack of awareness and information about rare chromosome and gene disorders creates significant stress.
- 2) Rare chromosome and gene disorders affect every aspect of daily life.
- 3) Individuals and families feel isolated.
- 4) Advocacy is not always valued or a choice.



Representing the patient voice



- Raising awareness with professionals and the public
- Sitting on committees and research advisory boards, e.g.
 - NHSE Genomics Clinical Reference Group
 - Co-chair for the Diagnosis Working Group, Future for Rare (Genetic Alliance UK)
 - SAB for DECIPHER, Manchester BRC, Manchester Rare Conditions Centre, MRC CoRE in Therapeutic Genomics
 - GenROC project
 - MHRA Rare Diseases Framework
- Responding to government consultations (e.g. unpaid carers, SEND reform)
- Education and training
 - Students, special schools, social workers etc



Research & Data Sharing

Participating in national & international research

RESEARCH OPPORTUNITIES

Brain and Behaviour in Neurodevelopmental Disorders of Genetic Origin (BINGO)

Who can get involved?
UK-based individuals over the age of 10 with a genetic diagnosis from the following list:
ACTB, ACTL6B, ARID1A, ARID1B, ARID2, ATRX, BICRA, DPF2, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1

BINGO Brain and Behaviour in Neurodevelopmental Disorders of Genetic Origin

RESEARCH ON CHROMATIN-RELATED CONDITIONS

WHAT IS THE PROJECT AIM?
This project aims to understand how differences in chromatin-related genes influence brain development, cognitive processes, and everyday experiences.

WHAT IS INVOLVED?
We have two research activities available:
(1) Online behavioural questionnaires and interviews
(2) Brain scans at the MRC CIB in Cambridge

WHO CAN TAKE PART?
UK-based individuals over the age of 10 with a genetic diagnosis from the following list:
ACTB, ACTL6B, ARID1A, ARID1B, ARID2, ATRX, BICRA, DPF2, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1

HOW TO GET INVOLVED
To express interest, visit our website: <https://bingo.mrc.ac.uk>
Have any questions? Contact us via: bingo@mcg.ac.uk
Telephone: 01223 855588

Attitudes towards sleep measures for children

Who can get involved?
Parents of a child diagnosed with a condition caused by a de novo (new) genetic change through the National Health Service (NHS) in England and Wales

Attitudes towards sleep measures for children

Does your child have a DE NOVO genetic variant?
PARENT VIEWS needed for a research study.

WHO CAN GET INVOLVED?
We are looking for parents who have a child with a condition caused by a de novo (new) genetic change. The chance of this happening again in future pregnancies is extremely small (1 in 10,000). We would like to hear your views about your child's condition, how you feel about it, and what you think would be the best way to manage it. There are no right or wrong answers.

PARTICIPATION WILL CONSIST OF AN ONLINE INTERVIEW WITH A RESEARCHER.
This will take approximately 30 minutes. You will be asked what you think you would like to know about your child's condition, how you feel about it, and what you think would be the best way to manage it. You will also be asked what you think would be the best way to manage it.

WE INVITE BOTH THE PARENT, INCLUDING FATHERS.
There is no obligation to participate. You will be asked what you think you would like to know about your child's condition, how you feel about it, and what you think would be the best way to manage it.

IF YOU WOULD LIKE MORE INFORMATION, PLEASE CONTACT RESEARCHER ALISON KAY
Email: alison.kay@nhs.uk
Phone: 01223 855588

FaceMatch
INTERNATIONAL RESEARCH INITIATIVE

Help another family achieve an earlier diagnosis

We are seeking facial photographs of children with genetic conditions or syndromes associated with intellectual disability. Children with the same genetic condition often share similar facial features. FaceMatch is a facial recognition technology that matches people with similar facial features to help families identify their child's condition.

You can find out more here: <https://www.facematch.org.uk>

NSW Health Hunter New England Local Health District

Helping families achieve an earlier diagnosis through facial matching

Who can get involved?
Families of children with a diagnosed intellectual condition or syndrome

Are you or your child affected by MCOPS12?
Join our natural history study!

RARB-related disorder, also known as MCOPS12, is a rare genetic condition found worldwide in mostly children. Mutations in the RARB gene were only recently discovered to cause this condition. Since it is so rare, the understanding of this condition as it presents over time is poorly understood. The fact that it is unclear how the condition changes, or how symptoms could appear or disappear, make it difficult to develop treatments.

You can help by participating in our study!

By participating, you can:

- Contribute to research
- Understand the condition
- Benefit from the knowledge generated by the research team

Interested?
Contact the study coordinator at: mcops12@mcg.ac.uk

How does RARB-related disorder affect you over time?

Who can get involved?
People affected by RARB-related disorder (also known as MCOPS12)

DO YOU OR YOUR CHILD CARRY A VARIANT IN NRXN1 / 2P16.3?

The ASPE Program is recruiting all individuals with a NRXN1 variant or 2p16.3 copy number variant (e.g. deletion) and their families. This includes those with and without an autism diagnosis, and individuals both with and without intellectual disability.

PARTICIPATION INCLUDES...

- Completing a series of online questionnaires
- Returning a free saliva kit for DNA analysis
- Option to wear a research-grade (Fitbit-like) device to track activity and sleep
- All completed in the comfort of your own home!

Scan to join ASPE today!

Visit our website at www.aspe.med.upenn.edu

Research in NRXN1 / 2p16.3

Who can get involved?
People with, or who have children with an NRXN1 gene variant of 2p16.3 deletion

The SAFEED Study
Join our natural history study!

FOCUS GROUPS OR INTERVIEWS

Caregivers of individuals with intellectual disability - have you used a safe sleeping space for your child in the last two years?

Please share your experiences with us!
Contact Bethan Corrick
(b.corrick@aston.ac.uk, 0121 204 5016) or scan the QR code to find out more about taking part in a 2 hour focus group with other caregivers or a 1 hour one-to-one interview, online or at Aston University

ASTON UNIVERSITY

REC ID: 53900 WPD Recruitment Paper v2.0 11.04.2025

Understand the perspectives of caregivers who use enclosed beds

Who can get involved?
Caregivers of people with intellectual disability

Share your views on new tests and treatments for genetic conditions during pregnancy

We need diverse input from people with lived experience of genetic conditions, including parents, carers, men, women and people from a range of backgrounds

In-person workshop, London
Date: 14 October 2025
Time: 10:00 - 15:30

Join online
Date: 22 October 2025
Time: 12:00-14:00

Scan to join ASPE today!

Views on new tests and treatments for genetic conditions during pregnancy

Who can get involved?
People with, and parents/carers of children with, genetic conditions

Decipher/EMBL-EBI, Genetic Alliance UK and Unique video on data sharing



Peer reviewed research and comment pieces

Health E

Received: 26 September 2023 | Revised: 5 January 2024 | Accepted: 10 January 2024

DOI: 10.1002/pd.6537

ORIGINAL ARTICLE

ORIGINAL

Can It
Explor
Pregn

Alison Kay^{1,2}
Shwetha Ram

¹MRC Weatherall

'Something tha
parents offered
clinical care in

Hannah McInnes-Dea
Holly Walton⁴ | Em

Journal of Community Genetics (2026) 17:82
<https://doi.org/10.1007/s12687-026-00911-w>

CONFERENCE PROCEEDING

Rare disease genomics and just
Fondation Brocher, 22–24 Janu

Angus Clarke¹ · Ruth Horn² · Elena Avram³ · Sid
Mike Drummond² · Roseline Favresse⁸ · Angelir
Meghan Halley¹² · Sasha Henriques¹³ · Hadewy
Tamar Nov Klaiman² · Teodora Lalova-Spinks^{19,2}
Elena Nicod²⁵ · Kelly E. Ormond²⁶ · Barbara Prai
Francesca Forzano³⁰

JMIR PEDIATRICS AND PARENTING

Original Paper

Parental Use of Social Media and the Internet in the
of Their Child's Genetic Neurodevelopmental Disor
Methods Study Nested in the GenROC Cohort Stud

Karen Jaqueline Low^{1,2}, MBChB, MRCPH, BSc; Georgia Treneman-Evans¹, MSc; Sarah L Wynn³, PhD; Jenny
Ingram¹, PhD; John Powell⁴, PhD

¹Centre for Academic Child Health, Bristol Medical School, University of Bristol, Bristol, United Kingdom

²Department of Clinical Genetics and Genomics, University Hospitals Bristol and Weston NHS Trust, Bristol, United Kingdom

³Unique, Oxted, United Kingdom

⁴Nuffield Department of Primary Care Health Sciences, University of Oxford, Oxford, United Kingdom

Karen Jaqueline Low ^{1,2}, Amy Watford,² Peter Blair,¹ Ian Nabney,³ John P
Sarah L Wynn,⁵ Julia Foreman,⁶ Helen Firth,^{7,8} Jenny Ingram ⁹

Keywords: advocacy | diagnosis | education | engagement | genomic medicine | healthcare | people living with a rare disease

Comment

<https://doi.org/10.1038/s41591-026-04542-z>

Reducing the diagnostic odyssey in rare
disease: why screening is not the only answer

Caroline F. Wright, Amicia Phillips, Sarah L. Wynn, Nick Meade, Emma L. Baple,
Anneke M. Lucassen & Leigh Jackson

 Check for updates

The timely detection of rare diseases is crucial,
and using a range of approaches will be key to
reducing the diagnostic odyssey.

Newborn screening (NBS) is one of the most successful public health
programs globally. NBS using a heel prick bloodspot test is now
offered in most countries and has substantially reduced the mor-
bidity and mortality associated with a small number of rare genetic
conditions. Whether, when and how to add further conditions to NBS
depends not only on technological and therapeutic developments,
but also on evidence that earlier detection leads to better outcomes,
as well as resource allocation. Many places are now exploring using
whole-genome sequencing (WGS) to augment NBS with hundreds of
additional rare genetic conditions. Here, we consider the potential
effect of genomic newborn screening (gnBS) through the lens of the
'diagnostic odyssey' in rare disease.

n", Caroline Lafarge², Kerr
ker ¹⁰

ir(s) 2024

sequencing tests are being
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; the implementation of p
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the ethical issues raised. Ty
laboratory analytical processes, awa
to private healthcare), and (2) Timel
termination of pregnancy, decision-making in the absence of prenatal sequencing results, and the importance of prenatal
sequencing results). Recognising both the practical and systemic ethical issues that arise out of delivering a national prenatal
sequencing service is crucial. Although specific to the English context, many of the issues we identified are applicable to prenatal
sequencing services more broadly. Education of health professionals and parents will help to mitigate some of these ethical issues.

European Journal of Human Genetics (2025) 33:360–367; <https://doi.org/10.1038/s41431-024-01700-0>

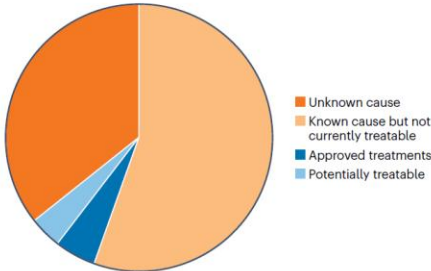


Fig. 1 | Rare conditions by genetic basis and treatability. Breakdown of rare
conditions by whether a genetic cause is known and, if so, whether the condition
is currently treatable. Data modelled from OMIM¹ and Bick et al.².

neurodevelopmental disorders: A Qualitative Study

Karen J. Low^{1,2}  | GenROC Consortium | Georgia Treneman-Evans¹ | Sarah L. Wynn³ | Jenny Ingram¹ 

¹Centre for Academic Child Health, Bristol Medical School, University of Bristol, Bristol, UK | ²Department of Clinical Genetics, UHBW NHS trust, Bristol,
UK | ³Unique, Rare Chromosome Disorder Support Group, Oxted, UK

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Keywords: distress | genomics | parent | rare

WILEY

Annals of
human genetics

as for Patients and

Information
; With Genetic
Information

on for Clinical Genomic

Low-key Kind campaign





Everyone notices.

Not everyone stares.

Low-key kind is out there.

In partnership with the JCDecaux
Community Channel, it's in **223 places**
across the UK.

Noticed one near you? Send us a photo.
We'll add it to the map and share some
of the sightings too.

**So one little sighting can help low-
key kind travel a bit further than the
street it's on.**

Send a photo



Noticed 14 Oct by Sarah

"Hello Manchester!"

Noticed.

**Market Street, outside 52-56,
Adidas**

Manchester · Street screen

On screen until 18 Oct

[Seen it too? Send a photo](#)



Take Home Messages

- Unique is here for you as well as patients and families
- Please use our information guides and resources
- Please refer patients and families to Unique (or the appropriate support group)
- Get involved in writing, reviewing or translating an information guide
- If you have any questions, please get in touch: sarah@rarechromo.org