17q12 Foundation NEWSLETTER

A RESOURCE FOR CHROMOSOME 17Q12 DELETION AND DUPLICATION SYNDROMES

July 2026

LETTER FROM THE BOARD

To our 17q12 community,

We are proud to celebrate our eighth annual 17q12 Awareness Day with all of you! The 17q12 Foundation now includes members from 22 countries and counting; a testament to the strength and growth of our global community. As we expand, our mission remains clear: to connect and support individuals, parents, and caregivers affected by chromosome 17q12 syndromes.

Last year, we were honored to hear from dedicated researchers and clinicians at the 17q12 Virtual Forum hosted by PRISMA, and we were proudly represented by our president, Allaina Wellman, at NORD's Rare Disease Summit, deepening our connection to the broader rare disease community.
We also marked an important milestone: one full year since launching the 17q12 Patient Registry, which now includes nearly 100 participants. This registry is a vital step toward advancing research and understanding.

Looking ahead, our next major goal is to expand our volunteer network to better support the 17q12 community. We remain committed to advocacy, research, connection, and support for every member of the 17q12 community. Thank you for being part of this journey and for supporting the 17q12 Foundation.

17q12 foundation

ALLAINA WELLMAN
PRESIDENT

LIZ FOURIE
VICE PRESIDENT

SHERIE SCOTT
SECRETARY

MARK DEMPSEY
TREASURER

MARGO CASADOS
BOARD MEMBER

Celebrating ACHIEVEMENTS

Hudson, Age 6 USA (DUP)

Hudson just finished kindergarten and he had a wonderful year! His speech has grown so much this year. He absolutely loves school and can't wait to go back.

Kyle, Age 15 UK (DUP)

Kyle is smashing every hurdle in his way. He is one of only a few who will be put forward for his exams next year. He recently became an uncle and wants to be a doctor who specialises in infectious diseases when he's older.

Alina, Age 6 USA (DUP)

Alina completed kindergarten and is learning to read! This is huge because last year she struggled to remember her letters and this year it just clicked for her.

Eden, Age 2, UK (DEL)

Eden has been doing so well with her speech and language therapy; naming all farm animals, counting to 10 and has learnt half of the alphabet. She is also learning how to communicate her needs by using Makaton to ask for help or more and she uses her manners by signing please and thank you!
Her parents are so proud of how Eden is developing in her own way in her own time

Daisy, Age 7 USA(DUP)

Daisy just finished reading her first chapter book, Magic Tree House #3

Joyann, Age 11, UK (DUP)

Joyann received a gold certificate because she did so well in maths.

Oliver, Age 9 US (DUP)

Oliver was nonspeaking until 4.5 years old. He is now thriving in a general education class at school and able to communicate fully!

Research Anouncements

Junghee Jenny Shin, M.D., Ph.D.
Assistant Professor of Medicine
Yale University School of Medicine

17q12 Immunological Study

We have been investigating T cell immune responses in patients with 17q12 deletion syndrome (17q12DS). We recruited 37 patients with 17q12DS and collected their medical histories, including allergies, infections, and autoimmunity. As expected, the majority of patients were presented with well-known clinical phenotypes of 17q12 DS, including renal diseases and neuropsychiatric disorders. Of interest, around 60% of patients were presented with recurrent respiratory infections. Therefore, we investigated alteration of T-cell characteristics and functions in 17q12 DS using bulk RNA sequencing, flow cytometric analysis and multiplex cytokine array.

Here we found that individuals with 17q12DS had a substantially decreased frequency of CD4+ T cells producing the T helper (Th) 1 cytokine IFN-g, a major cytokine involved in defense against viral and bacterial infection, compared to age-matched healthy controls (HCs). RNA seq analysis of CD4+ T cells revealed decreased levels of TBX21, encoding the Th1 transcription factor T-bet, IFNG, and other Th1 chemokine encoding genes in subjects with 17q12DS compared to HCs. These findings were validated at the protein level using flow cytometry and a multiplex assay. Our study is the first to demonstrate immune alterations in 17q12DS characterized by decreased T-bet and its downstream molecules such as IFN-γ. This work was published in Lancet eBioMedicine. We plan to explore the possibility of evaluating. recombinant IFN-γ as a potential therapeutic. option for individuals with 17q12DS who have recurrent sinopulmonary infections. We also plan to study the association of genetic and epigenetic changes and T cell alterations in 17q12DS.

LEARN MORE

Research Anouncements

Dr. Pengfei Liu's Study

There is an IRB-approved study at Baylor College of Medicine led by Dr. Pengfei Liu that focuses on providing a comprehensive and precise diagnosis for the genetic condition of 17q12 in you or your family member. Participating involves collecting blood samples from you and your family member including your medical information. Genome sequencing will be performed on the samples collected. Analysis of the sequence data will be done to understand the cause of your genetic condition. If you are interested in study participation, please contact us at precisiongenome@bcm.edu . Please provide your full name, phone number, e-mail address, and good day and time for contact.

Enrollment requirements:

- Individuals with the 17q12 deletion.
- Individuals with the 17q12 duplication who have either renal, endocrine, or reproductive system issues.

Pengfei Liu, Ph.d.
Molecular and Human Genetics
Baylor College of Medicine

Daniel Moreno De Luca MD MSc
CASA Research Chair
Associate Professor & Principal Investigator
Precision Medicine in Autism (PRISMA) Group
Child and Adolescent, and Adult Psychiatry

PRISMA Study

The PRISMA group continues to connect with the global community through research, clinical care, education, and community engagement. Stay up-to-date at https://link.edgepilot.com/s/f61a3318/7RXPjIry9ECfWeS__JU4hw?u=http://www.precisionmedicineinautism.org/

Our research study "A Genomic Approach to Precision Medicine for Autism and Neurodevelopmental Conditions" is enrolling children and adults with a 17q12 deletions or duplications to understand how genetics can impact their healthcare. Participation is open to those who have this genetic change, whether or not they have autism, epilepsy, kidney disease, or other neurodevelopmental conditions. The study will take approximately 6 hours and involves collecting health information and completing questionnaires and assessments online.

LEARN MORE

FUNDRAISE FOR 17Q12

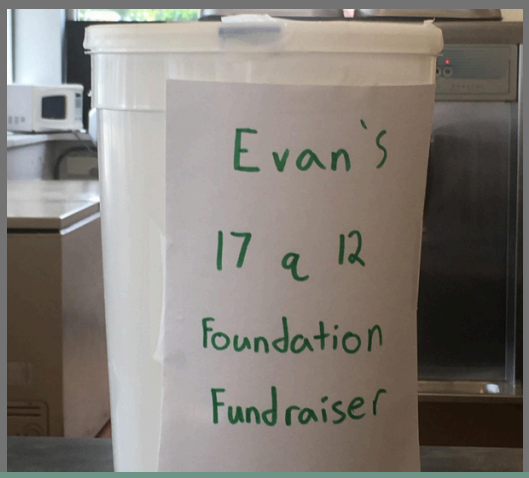
Our aim for fundraising this year is to finance a crucial family conference in the future. By joining forces, we can gather enough funds to achieve this target.

Do you have an idea for a fundraiser? No fundraiser is too small. The 17q12 Foundation is a registered 501(c)(3) organization and relies heavily on fundraising and community support. Any fundraising big or small makes an impact.

FUNDRAISING IDEAS

- Birthday Fundraisers on Facebook
- Run your own Bonfire shirt campaign
- Plan a silent auction
- Organize a bake sale
- Ask a local restaurant about Dine & Donate
- Hold a car wash or garage sale

Please email us at info@chromo17q12.org if you would like to organize a fundraiser. We can provide our tax ID number, advertising, and support your efforts!



CALL FOR VOLUNTEERS

Join Us in Making an Impact!

The 17q12 Foundation is growing, and we need your help!
We're a volunteer-run organization dedicated to supporting individuals and families impacted by chromosome 17q12 syndromes. As we continue to expand, we're forming new committees to help us strengthen our impact. We're looking for passionate, committed volunteers that are verified members of the 17q12 Foundation to get involved.
We're currently seeking volunteers to help launch two key committees:

- Fundraising Committee – Help us raise essential support for research, programs, and outreach.
- Program Committee – Contribute to developing meaningful programs for the 17q12 community.

Whether you are an individual with 17q12, a parent, or caregiver, your skills and time can help shape the future of the 17q12 Foundation.

Together, we can build a stronger, more connected community.

Interested in learning more?
Email us at info@chromo17q12.org

17q12 Terms

Check out Unique's glossary of genetic terms to learn more: https://rarechromo.org/glossary/

Deletion

When a piece of DNA is missing. Deletions can vary in size from one base pair to tens, hundreds, thousands (kb) or millions (Mb) of base pairs. Those too small to be seen using a microscope are known as microdeletions.

CMA chromosome microarray analysis

CMA is a high resolution genetic test that analyses a person's entire genome using microchip based technology. There are different types of CMA, e.g. Array CGH and SNP array.

Duplication

when a piece of DNA has been copied and is present as an extra piece (or multiple extra pieces). Duplications can vary in size from one base pair (bp) to tens, hundreds, thousands (kb) or millions (Mb) of base pairs. Those too small to be seen using a microscope are known as microduplications.

De Novo

latin terminology meaning 'a new event'

q (in 17q12)

long arm of chromosome 17

Pathogenic

this word means 'disease causing'. When it's used to describe a genetic change it means that features or symptoms have been observed in association with the change.

CNV Copy Number Variant

This is a variation in the number of copies of a specific piece of DNA in a chromosome. For example, we normally have two copies of each autosomal chromosome (chromosomes 1-22); if a piece of DNA is missing from one chromosome but not the other, the new copy number for that section of chromosome is 1. If a piece of DNA is duplicated on one chromosome but not the other, the new copy number is 3. It is possible to have copy numbers of 0 or multiples of more than 3.

COMMUNITY SPOTLIGHT

Teodora, Age 6, Switzerland (DUP)

At 9 months old, Teodora was hypotonic and unable to sit up, but doctors advised waiting years before pursuing genetic testing. With no support and growing concern, her family—originally from Serbia—traveled back there for answers. After months apart and many uncertainties, they finally received a diagnosis: a de novo 17q12 duplication. Despite the emotional toll and sleepless nights filled with worry, a dedicated physiotherapist named Jelena helped Teodora make incredible progress—from not being able to roll over to walking at 16 months and later speaking her first words. Today, aside from slight ataxia, Teodora is thriving. Although there is always some fear about what the future might bring. Her older sister, Nikolina, also played a key role by encouraging and modeling milestones alongside her. Teodora's mother encourages us all to trust your instincts, seek support where you can, and believe in your child while not giving up-because these children are true fighters. —The Milosevic Family

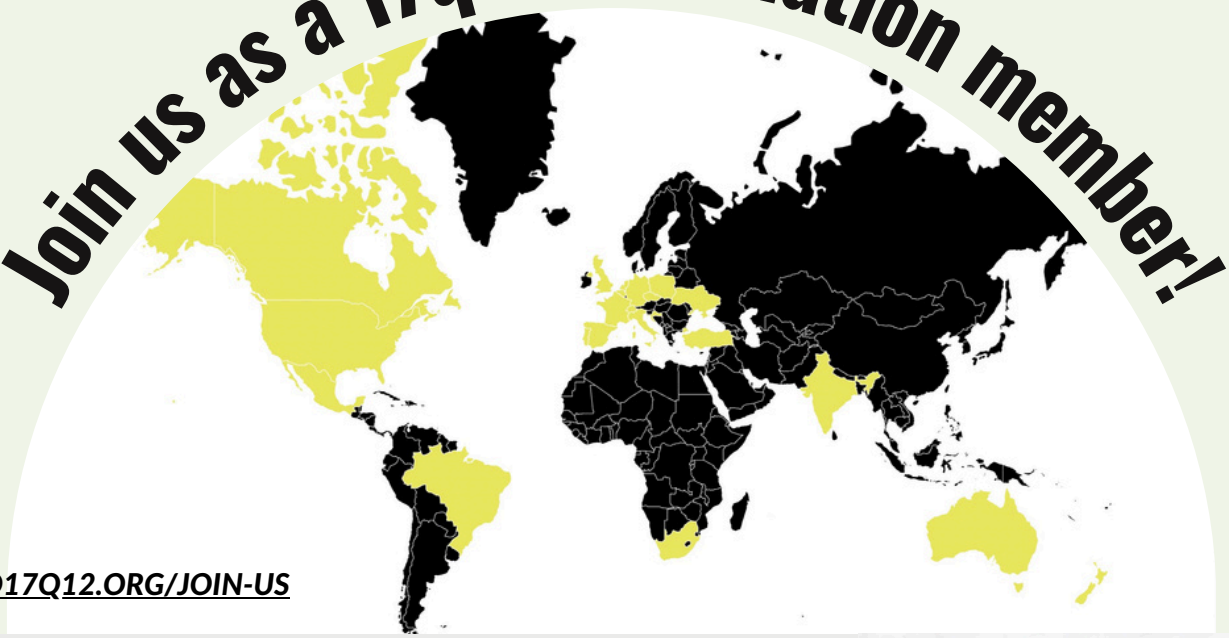
Mason, Age 10, USA (DEL)

Mason was diagnosed at 8 months old following a visit with a genetics specialist. As much as we try not to let it define Mason, 17q12 has been part of his journey since birth. He's experienced delays in all developmental milestones. He began early intervention at 10 months old and started private PT, OT, speech, and feeding therapy at age 1. By age 3, he began preschool where therapies were incorporated into his daily routine. Over the past 9 years, Mason has faced and overcome countless challenges—from frequent doctor visits and blood draws to multiple surgeries. Mason will be entering 4th grade this year. He spends most of his day in the general education classroom, with support from special education services as well. He has an IEP in place and works closely with a 1:1 associate who helps support his learning and daily success at school.
Mason is a true social butterfly—his smile, joy, and energy naturally draw people to him. He makes friends wherever he goes and feels emotions deeply, wearing his big heart on his sleeve. He's incredibly passionate about sports and thrives on competition. He is also intensely loyal to his older sister and younger brother—he adores them and is always their biggest cheerleader and protector.

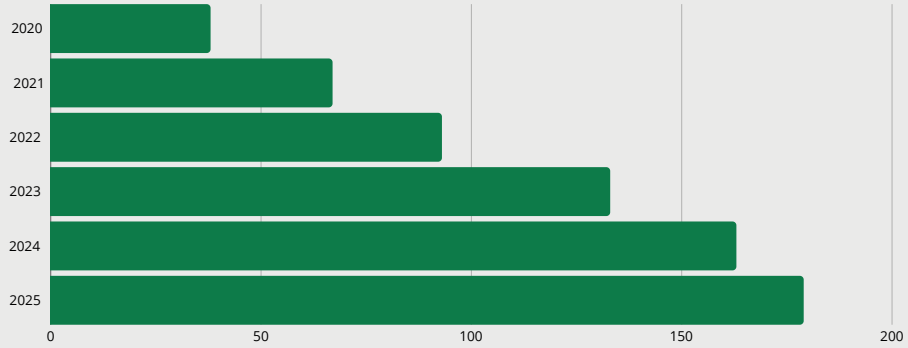
Wesson, Age 3, USA (DUP)

Wesson was diagnosed at 9 months old and he has Autism level 2, hypotonia, feeding difficulties, Gerd, eczema, behaviors, SPD, restless leg syndrome, sleep apnea, tracholmacia , abnormal head MRI, etc. He is very determined, loves to help do things, always on the go, very smart, loving and full of life.

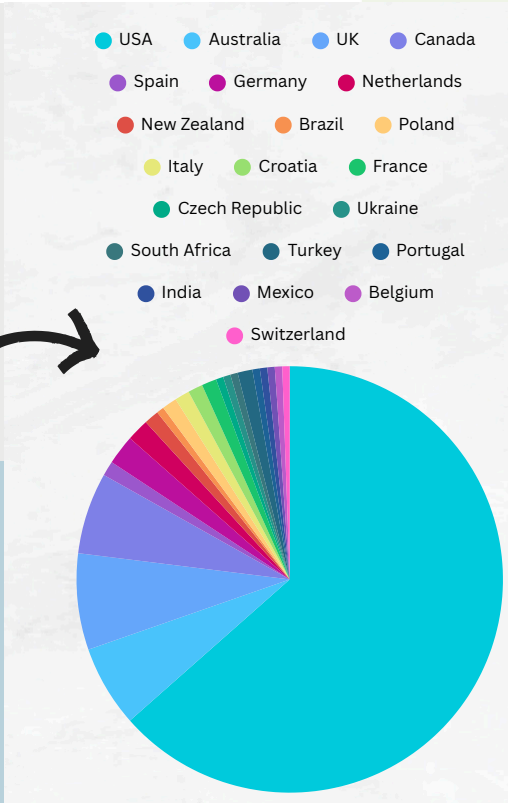
Join us as a 17q12 Foundation member!



CHROMO17Q12.ORG/JOIN-US



VERIFIED MEMBERS 2020-2025



WHO ARE WE?

We currently have 179 verified members/families from the following countries:

76 verified members have 17q12 **duplication**.
103 verified members have 17q12 **deletion**.

There are 296 verified members in our private support group on Facebook for parents, caregivers, and individuals directly affected by a 17q12 syndrome.

408 Membership Registration forms have been submitted.

As of July 2025, the 17q12 Foundation proudly includes confirmed members from 22 countries around the world! By joining the 17q12 Foundation, individuals become part of a growing global community focused on connection, support, and collaboration. Members receive exclusive updates on research opportunities, upcoming conferences, and virtual events. Verified members also gain access to our private Facebook support group, created specifically for those directly impacted by chromosome 17q12 syndromes. In addition, members can opt into 17q12 Near You, an initiative designed to connect families and individuals within their local regions for added support. If you or your child has been diagnosed with a chromosome 17q12 deletion or duplication, we warmly invite you to join us at: chromo17q12.org/join-us

Let's build this community—together.

MAKE A MARK ON THE FUTURE OF RESEARCH FOR CHROMOSOME 17Q12 SYNDROMES

chromo17q12.org/registry

17Q12 PATIENT REGISTRY FAQS

How is the data collected?

Data is collected through a secure web-based application (that can be accessed by computer, tablet or phone) developed by the National Organization for Rare Disorders, Inc. (NORD®), Study participants respond to questions grouped within a series of surveys developed per study standards and in collaboration with syndrome specific experts.

What types of data will be collected in the 17q12 Patient Registry?

The data collected includes but is not limited to:

- Socio-demographics
- Medical and diagnostics
- Treatment and syndrome progression
- Management of care
- Quality of life

Who can join the study?

This study is open to anyone who has a chromosome 17q12 deletion or duplication syndrome diagnosis and meets the study inclusion criteria for participation.

Is there a cost to participate?

There is no cost to the patient to join this study.

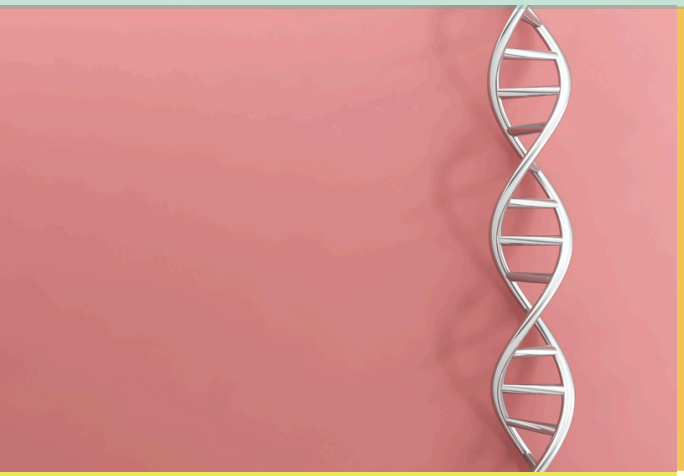
Can data be collected worldwide?

The registry uses an online platform which allows participants to contribute data from anywhere in the world. Individuals from other countries who enter data into the registry should be aware that data and privacy laws are different in the U.S. from other countries. This U.S. based registry will protect data and privacy according to U.S. requirements.

Is the data safe?

The registry follows strict government guidelines to ensure patient information is protected. The platform is served over HTTPS, which means that the data is encrypted when being sent from the user's browser to the NORD servers. The data is also kept encrypted in the NORD database. Communications between the registry platform application server and the database are also encrypted. As with any information you provide electronically, there is a very rare chance that your privacy could be compromised. However, the registry and the security measures minimize the chance of this occurring.

Learn more at 17q12registry.iamrare.org/faq/



17q12 Deletion Overview

Source: GeneReviews®: 17q12 Recurrent Deletion Syndrome (Updated August 2025), National Center for Biotechnology Information (NCBI).

MOST COMMON FEATURES (>50%)

- Kidney structural or functional defects
- Neurodevelopmental/ neuropsychiatric disorders
- Mild dysmorphic features

COMMON FEATURES (25-50%)

- Maturity-onset diabetes of the young type 5
- Male or female reproductive tract differences
- Structural & functional liver abnormalities
- Hyperparathyroidism
- Eye abnormalities
- Structural & exocrine abnormalities of the pancreas
- Prematurity
- Nonspecific structural brain findings

LESS COMMON FEATURES (<25%)

- Congenital cardiac anomalies
- Musculoskeletal features
- Other gastrointestinal features
- Seizures

What is 17q12 Deletion Syndrome?

17q12 deletion syndrome is a rare genetic condition caused by a missing piece of chromosome 17. Because a copy of ~15 genes are deleted (including HNF1B), the syndrome can affect multiple organs and body systems.

Every person with 17q12 deletion syndrome is unique. Some individuals have only mild symptoms, while others have more complex medical and developmental needs. Even members of the same family may be affected very differently.

IMPORTANT TO REMEMBER

- Because 17q12 deletion syndrome can affect many body systems, care is often provided by a multidisciplinary team.
- Not everyone develops every feature.
- Symptoms may appear at different ages, from before birth through adulthood.
- Many individuals lead healthy, productive lives with appropriate medical care and early intervention.
- Regular follow-up with healthcare providers helps identify and manage new symptoms as they arise.

17q12 Duplication Overview

Source: GeneReviews®: 17q12 Recurrent Duplication (Updated January 2022), National Center for Biotechnology Information (NCBI).

MOST COMMON FEATURES (>50%)

- Other neurologic abnormalities
- Hypotonia (low muscle tone)
- Intellectual disability
- Speech delay
- Behavioral abnormalities

COMMON FEATURES (20-50%)

- Gross motor delay
- Microcephaly (head circumference <5th percentile)
- Seizures/Epilepsy
- Ophthalmologic (eye) abnormalities
- Endocrine abnormalities
- Short stature (height <5th percentile)
- Low weight (weight <5th percentile)
- Renal (kidney) abnormalities

LESS COMMON FEATURES (<20%)

- Cardiac abnormalities
- High weight (weight >95th percentile)
- Tall stature (height >95th percentile)

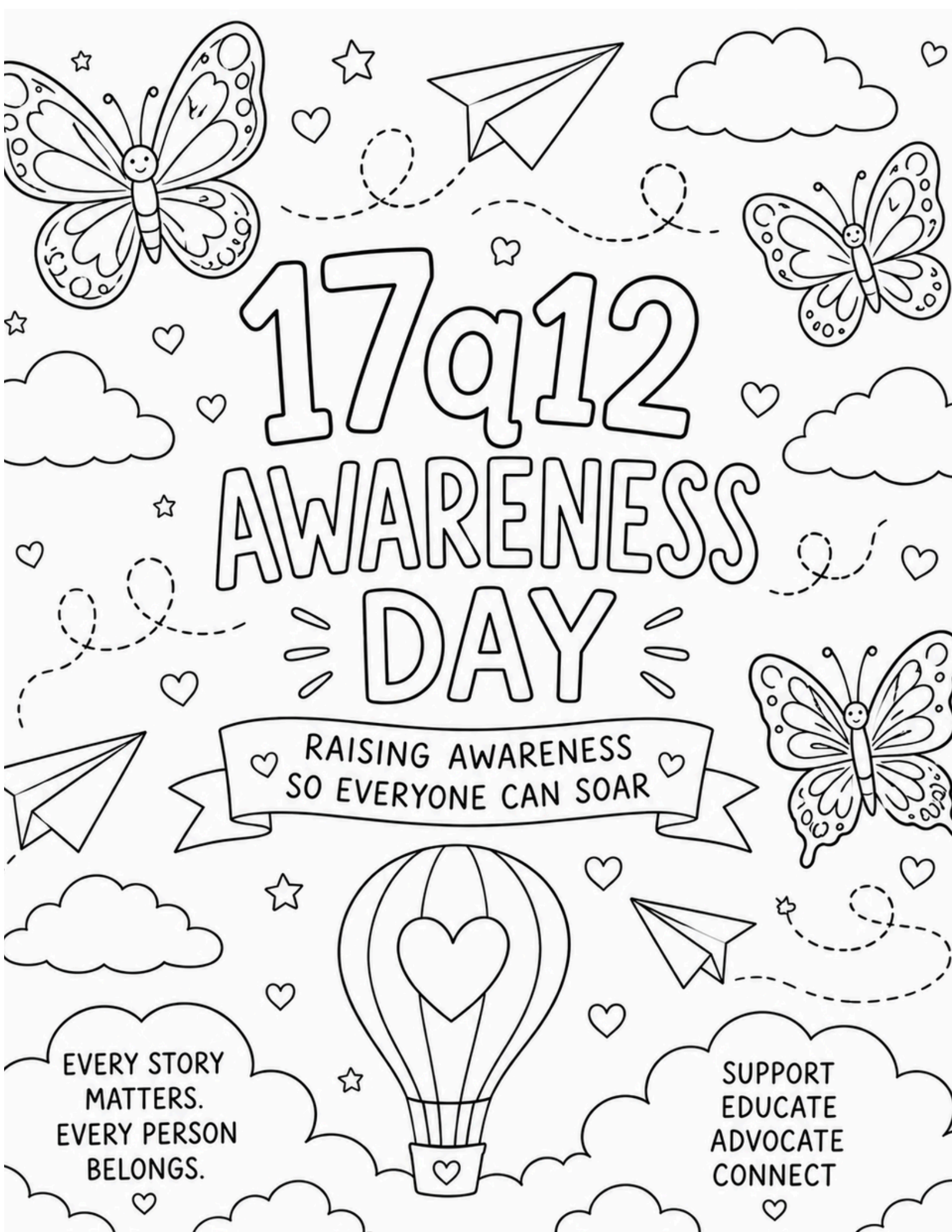
What is 17q12 Duplication Syndrome?

17q12 duplication syndrome is a rare genetic condition caused by an extra copy (duplication) of a small section of chromosome 17. This duplicated region contains ~15 genes, including HNF1B, although it is not yet clear which genes are responsible for the features seen in affected individuals.

The condition is highly variable. Some individuals have significant developmental or medical challenges, while others have very mild symptoms, or no obvious symptoms at all. Even people within the same family who have the duplication can be affected very differently.

IMPORTANT TO REMEMBER

- Not everyone with 17q12 duplication has symptoms.
- The condition has variable expressivity, meaning symptoms can range from none to significant, even within the same family.
- Early developmental support and individualized medical care can greatly improve outcomes.
- Regular follow-up allows healthcare providers to identify and address new concerns as they arise.


17q12
AWARENESS
DAY
RAISING AWARENESS
SO EVERYONE CAN SOAR
EVERY STORY
MATTERS.
EVERY PERSON
BELONGS.
SUPPORT
EDUCATE
ADVOCATE
CONNECT