International 22q11.2 Foundation

Newsletter December 2024

Greetings from the International 22q11.2 Foundation.

We hope you and your family are doing well during this festive season. As you prepare gifts and cards for your loved ones, we hope you will consider sharing this newsletter with them as a way of spreading awareness about 22q differences. If you need a quick list of points about 22q11.2 deletion and duplication syndromes, please have a look on page 5.

November was 22q Awareness Month. Our Foundation and other support organizations have been actively spreading the word about 22q differences. On November 22, many buildings were lit up in red (pages 2 and 3). Our social media posts celebrated individuals, highlighted new research findings, fundraised, and promoted resources (page 4).

Congratulations to our board member, Donna McDonald-McGinn for receiving a special award in Australia. On page 6 of this newsletter, you can read about the award as well as the wonderful event she helped start – 22q at the Zoo Worldwide Awareness Day.

We are happy to bring you a new series in Health Conditions Explained. The 4 topics of the Immune System Series as well as a list of all the info sheets published so far are on page 9. As usual, we also bring you the latest research news – this time on functioning and brain processes (pages 10 and 11).

We take this opportunities to share with you two excellent resources from support organizations in Europe: the Spanish group produced an educational handbook for professionals and therapists, while the Dutch group generated a documentary video called Surviving 22q11. Both resources are very useful – have a look on page 7.

Last but not least, we are excited to let you know that the clinical recommendation documents are now available in multiple languages (see page 8). More languages will be available in the new year, and we are grateful to all the individuals who have worked so hard to make the documents accessible.

Happy Holidays!

With best wishes,

The International 22q11.2 Foundation, Inc

We Lit Up the Night for 22q!

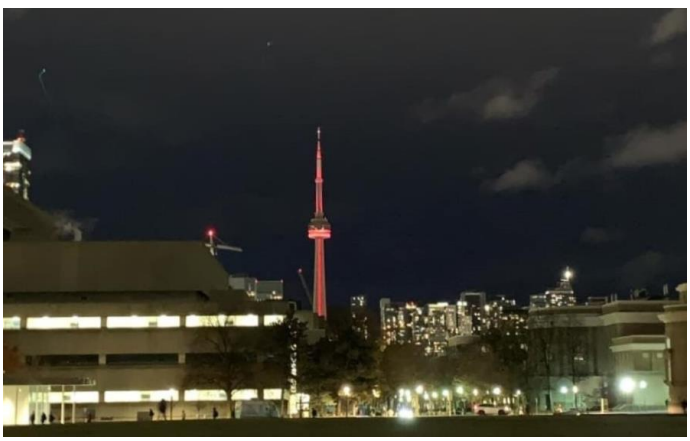
To increase public awareness of 22q differences, in 2018, La Asociación Síndrome 22q11 in Spain launched an awareness campaign entitled "**Luces por el 22q**" (Lighting the Night for 22q). Buildings and monuments were illuminated in **red** on November 22 (22/11 or 11/22 in North America) as a play on the name of the chromosome difference.

In 2024, close to 70 buildings were lit up in red in Spain (collage below). We are also bringing you the photos of some of the other buildings that were lit up on November 22nd, 2024.

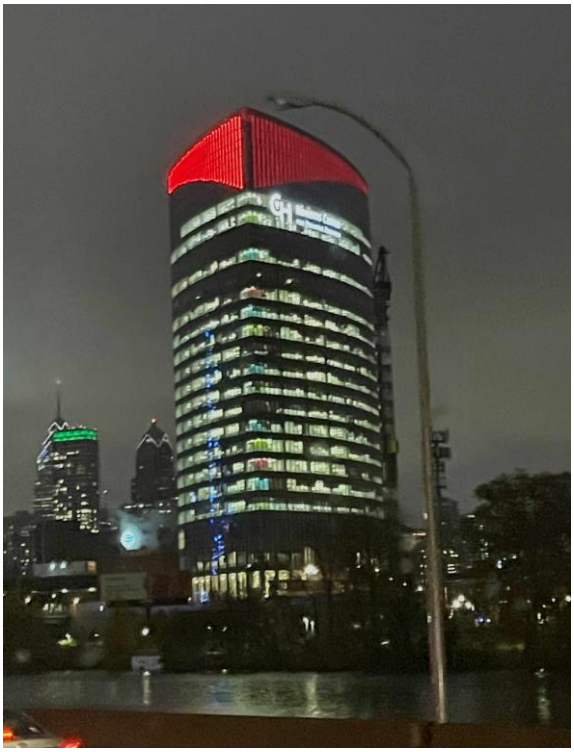
Close to 70 buildings in Spain were lit up in red

Ixelles Municipal Hall
Ixelles, Belgium

Podkarpackie Voivodeship Office
Rzeszów, Poland



CN Tower
Toronto, ON, Canada



Children's Hospital of Philadelphia (CHOP), Philadelphia, PA, USA

Sky Wheel
Helsinki, Finland

Oulu Academic Hospital
Oulu, Finland

If you would like to light up a building or monument in your city/town on November 22nd, 2025, we'll have a request letter ready for you on our website in the summer.

19th Annual Faces of Sunshine Calendar

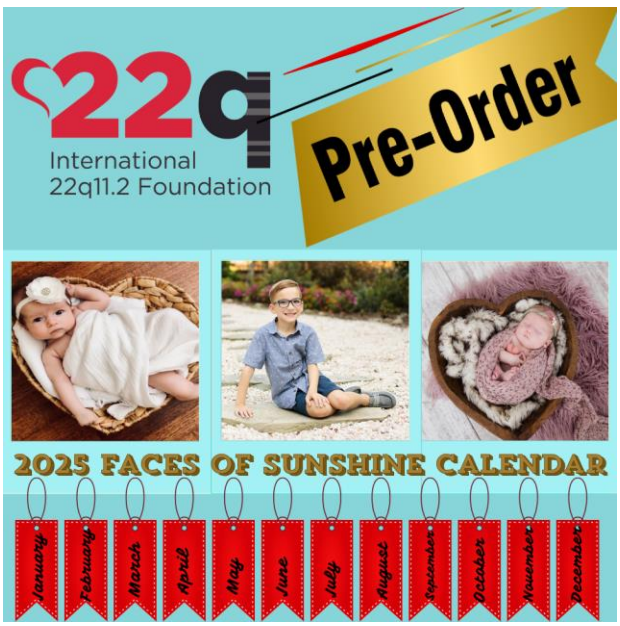
Many thanks to all who submitted photos for the 2025 calendar. The names of the featured individuals will be revealed on the **Calendar Campaign** webpage of the Foundation.

This calendar is a great way to show your 22q pride. We very much hope you enjoy the calendar as well as all of the beautiful faces of sunshine throughout the New Year!

Calendars may be **ordered via our website**. All proceeds benefit the work of the International 22q11.2 Foundation, Inc.

Thank you for your support!

Regards,
The International 22q11.2 Foundation, Inc.



We Spread the Word about 22q Differences!

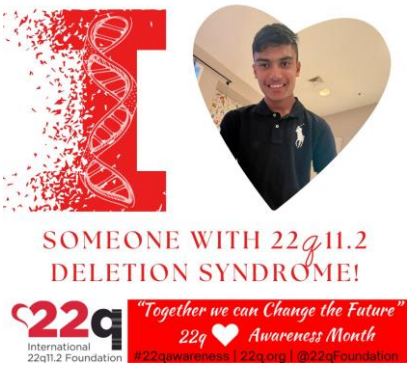
November was 22q Awareness Month.

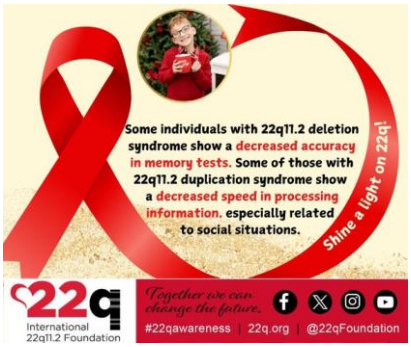
22q11.2 deletion and duplication syndromes are not well-known, even among medical professionals.

This November, staff members from the International 22q11.2 Foundation posted diligently on social media platforms to raise awareness for 22q differences. With help from our families and 22q experts, our posts celebrated individuals, highlighted new research findings, fundraised, and promoted resources. We need to let the world know what complex conditions these are, how much support is sometimes required, and how these amazing, beautiful children and adults overcome many obstacles to survive and thrive! We also remember the ones who are lost far, far too soon. We want the world to know all our stories and to recognize the beauty within these individuals.

Here are some posts from this year. You can click on each image for the original post on Facebook.

Many thanks to everyone who contributed!





Telling people about 22q

Not sure what to tell people about 22q11.2DS & 22q11.2Dup? Here are some points.

22q11.2 deletion syndrome (22q11.2DS)

1. Caused by a missing piece on chromosome 22q. One copy (out of 2) of some of the genes that usually tell the body how to form and function go missing
2. Happens in about 1 in 2148 babies, but not many people know about it
3. Used to be known by other names such as DiGeorge syndrome & velocardiofacial syndrome
4. Common features include:
    - Birth defects of the heart
    - Birth defects of the palate
    - Developmental delays
    - Intellectual disabilities
    - Anxiety disorders
    - Schizophrenia
    - Calcium level issues
    - Epilepsy
    - Immune deficiencies
    - Plus many others
5. Each person (even members within the same family) is affected differently in terms of symptoms and severities.
6. Usually arises as a new event in a family. Only about 10% of patients have a parent who is also affected.
7. Once a person has the deletion, he/she has a 50% chance of passing it onto the next generation at each pregnancy.

22q11.2 duplication syndrome (22q11.2DupS)

1. Caused by an extra piece on chromosome 22. There are 3 copies (instead of 2) of some of the genes that usually tell the body how to form and function
2. Happens in about 1 in 4000 babies, but not many people know about it
3. Much less is known about the duplication syndrome compared the deletion syndrome.
4. Common features include:
    - Birth defects of the heart
    - Birth defects of the palate
    - Developmental delays
    - Intellectual disabilities
    - Autism spectrum disorder
    - ADHD
    - Vision problems
    - Ear anomalies
    - Immune deficiencies
    - Plus many others
5. Each person (even members within the same family) is affected differently in terms of symptoms and severities.
6. Often runs in the family. About 60%-70% of patients has a parent with the duplication.
7. Once a person has the duplication, he/she has a 50% chance of passing it onto the next generation at each pregnancy.

To find out more, please visit www.22q.org.

Donna McDonald-McGinn Receives the Dedication to Service Award

Congratulations! Our very own International 22q11.2 Foundation Founding Board Member, Donna McDonald-McGinn, MS, LCGC receive the 2024 **Dr. Tony Lipson Memorial "Dedication to Service Award"** on the occasion of the 30th anniversary gala of 22q Australia and New Zealand.

Donna is the Director of the 22q and You Center at the Children's Hospital of Philadelphia. On the bottom left is a photo of Donna (center) with 22q Australia and New Zealand President, Maria Kamper (left), and the UK's Max Appeal Founder, Julie Wootton (right).

Donna also gave an outstanding lecture at the Sydney Children's Hospital, and was very honored to receive the award!

22q at the Zoo Worldwide Awareness Day

Sunday May 18th, 2025

See the 22q at the Zoo photos from:

2024 | 2023 | 2022

Did you know that **Donna McDonald-McGinn was the person who coined the event name "22q at the Zoo – Worldwide Awareness Day"**? That was in 2011!

Save the date for the 15th Annual 22q at the Zoo – Worldwide Awareness Day, which will take place on **Sunday, May 18th, 2025**. It is an opportunity for families, friends and professionals to socialize, network and raise public awareness of 22q11.2 syndromes.

If there were no 22q at the Zoo events in your area in 2024, you can host one in 2025! Please visit the Host a 22q at the Zoo Event section on our website for more information. We even have an event planner letter ready for you! No zoo? No problem. Consider gathering at a park, a farm, or even an aquarium.

If you have any questions, please email us: info@22q.org. Thank you.

Educational Handbook for 22q11.2DS

Learning difficulties and behavioural disorders are common among children with 22q11.2 deletion syndrome (22q11.2DS). Asociación Síndrome 22q11 (22q11 Syndrome Association in Spain) has produced an educational handbook for professionals and therapists. The handbook, which is 96 pages in length, provides information on the educational characteristics and learning needs of students with this diagnosis. Some of the topics covered include academic skills, relationship with peers, bullying, and educational transition.

It will help educational professionals improve the students' quality of life in school, ultimately reaching their potential and reaching their goals in terms of academic achievement and personal growth.

Click on a link below to see the Educational Handbook in English, Spanish, or Portuguese

22q11 Syndrome Educational Handbook

Guía Educativa Síndrome 22q11

Sindrome 22q11 Guía Educacional

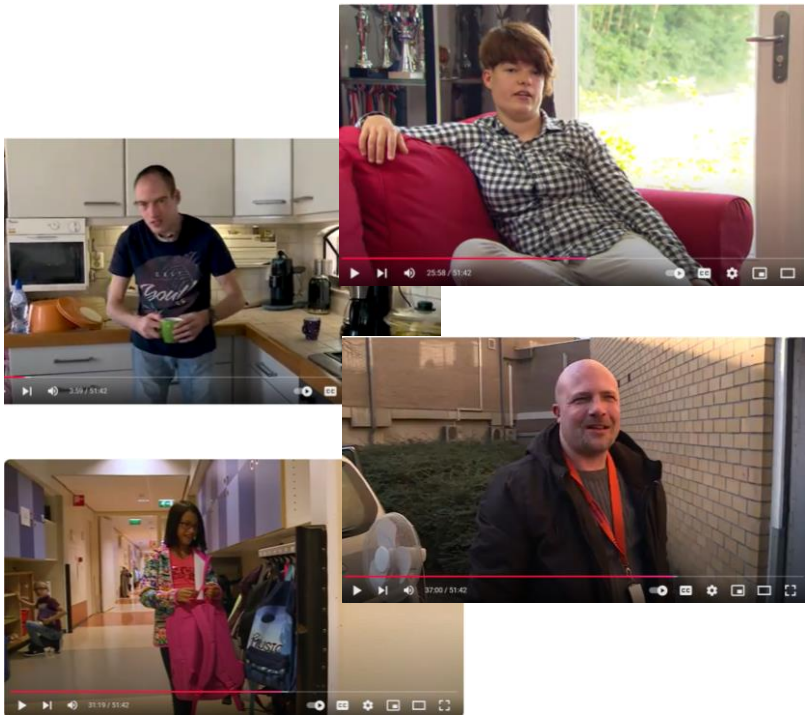
Documentary: Surviving 22Q11

Living with 22q11.2 deletion syndrome (22q11.2DS) has a major impact on daily life.

The Stichting Steun 22Q11 Foundation in the Netherlands made a beautiful Dutch documentary called Over Leven Met 22Q11 (Surviving 22Q11). It follows the lives of four individuals with 22q11.2DS over the span of a year, showing viewers how difficult it is to live with this genetic condition.

This video is about 52 minutes in length and covers topics such as:

- Choice of school
- Transition to independence
- Employment
- Housing
- Relationships
- Prenatal testing
- Receiving the diagnosis as an adult



The ages of the featured individuals were 9, 22, 26, and 38 years old. The conversations are in Dutch, but English subtitles are available.

Click to watch the video

Clinical Recommendations Now Available in More Languages

In 2023, an international team of experts, including members of our Medical Advisory Board, published clinical recommendation documents. The three sets of clinical recommendations documents took research results and applied the knowledge towards clinical care for children, adults, and fetuses with this condition. The links to all three documents have been posted on the 22q11.2 Society website.

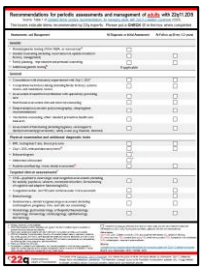
In order to provide equitable access to healthcare and expert knowledge to communities whose primary language is not English, we have begun to translate the documents into other languages. Please click the links on the right for the available documents.

All available translations are posted on the 22q11.2 Society website as they become available.

The clinical recommendations documents for children and adults each contains a useful table that lists the recommended periodic assessments. We have made the 2 tables into checklists in the form of fillable savable pdfs! Whenever you or your loved one completes an assessment, just put a check mark directly on the checklist on your computer.

↑ **Checklist for Children**

How to use the file directly on your computer



↑ **Checklist for Adults**

How to use the file directly on your computer

Clinical Recommendations 2023

Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome

- **English** (Original)
- Spanish / Español
- French / Français – in progress
- Simplified Chinese / 简体中文 – in progress

Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome

- **English** (Original)
- Spanish / Español
- French / Français
- Traditional Chinese / 繁體中文

Prenatal Screening and Diagnostic Considerations for 22q11.2 Microdeletions

- English (Original)

We thank the 22q11.2 Society as well as the following organizations and individuals for translating and reviewing the Spanish, French, and Traditional Chinese documents.

Project supervision and resource acquisition: Anne Bassett, Donna McDonald-McGinn, Erik Boot, Sólveig Óskarsdóttir

Project coordination – Joanne Loo

Spanish translation – Sixto Garcia-Minaur and Gabriela Repetto

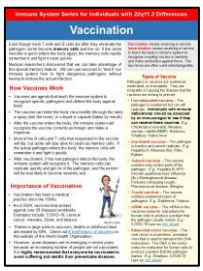
French translation – Khadijé Jizi, Sébastien Jacquemont, Maude Schneider, and Frédérique Bonnewijn

Traditional Chinese translation – University Health Network Translation Services, Joanne Loo, Wilson Loo, Jasmine Lee, and Brian Chung

Immune System Series – 4 Topics Available

Our newest series on the "**Health Conditions Explained**" section of our website focusses on the immune system. We have so far published information sheets on 4 immune system-related topics. Special thanks to Dr. Kathleen Sullivan, Immunologist and Joanne Loo, Programmatic and Educational Tool Developer.

- The Immune System
- Vaccination
- Immune Deficiency Related to T Cells
- Immune Deficiency Related to B Cells



Health Conditions Explained – Topics Published So Far

HEART SERIES

- The Heart and Normal Blood Flow
- Ventricular Septal Defect (VSD)
- Tetralogy of Fallot (ToF)
- Truncus Arteriosus (TA)
- Interrupted Aortic Arch (IAA)

DENTAL SERIES

- Let's Talk 22q Teeth – Info for Families
- Dental Health in Children with 22q – Info for Dentists

PALATE SERIES

- The Velopharynx
- Cleft Palate & Submucous Cleft Palate
- Velopharyngeal Dysfunction: Introduction and Causes
- Velopharyngeal Dysfunction: Diagnosis
- Velopharyngeal Dysfunction: Surgery
- Velopharyngeal Dysfunction: Speech Issues

22q GLOSSARY

- Third Edition

MENTAL HEALTH SERIES

- Mental Health and 22q11.2 Deletion Syndrome
- Mental Health and 22q11.2 Duplication Syndrome
- What Parents Can Do for Their Children
- Attention Deficit Hyperactive Disorder
- Autism Spectrum Disorder
- Anxiety Disorders
- Psychotic Disorders [Including schizophrenia]
- Mood Disorders
- Mental Health Q&A (Spring 2023)

SPEECH SERIES

- Introduction to Speech
- Consonants and Vowels in English
- Speech Disorders in Individuals with 22q11.2 Deletion Syndrome – an Overview
- Speech Disorders in Individuals with 22q11.2 Duplication Syndrome – an Overview
- Voice Disorders
- Speech Sound Disorders
- Motor Speech Disorders
- How Parents and Professionals Can Help

GI SERIES

- GI Issues in 22q11.2 Deletion Syndrome
- The Digestive System
- Feeding Difficulties
- Swallowing and Dysphagia
- Constipation
- Gastroesophageal Reflux Disease (GERD)
- Esophageal Dysmotility
- Gastroparesis
- Cholelithiasis (Gallstones)
- Nausea and Vomiting
- Autoimmune Issues in the GI System
- Inguinal Hernias
- Nonalcohol Fatty Liver Disease (NAFLD)

IMMUNE SYSTEM SERIES

Newly published in December 2024

- The Immune System
- Vaccination
- Immune Deficiencies Related to T Cells
- Immune Deficiencies Related to B Cells

More topics will be available next year.

How Well Do Adults with 22q11.2DS Function?

Cognitive, adaptive and daily life functioning in adults with 22q11.2 deletion syndrome.
Vingerhoets C, Ruiz-Fernandez J, von Scheibler E, Vergaelen E, Volbragt N, Soons N, Serrarens C, Vogels A, Boot E, van Amelsvoort T, Swillen A. BJPsych Open. 2024 Nov 11;10(6):e203. doi: 10.1192/bjo.2024.745. PMID: 39523675.

This study aimed to assess the cognitive abilities, adaptive functioning, and functional outcomes of adults with 22q11.2DS.

Methods

- Review of clinical and research records to gather information on:
  - Cognitive ability (full scale IQ or "FSIQ")
  - Adaptive functioning (VABS II)
  - Intellectual disability (ID)
  - Mental health conditions
  - Functional outcomes: living situation, work, and marital status

Study participants

- 250 adults with confirmed 22q11.2DS from the Netherlands and Belgium
- Includes 57 adults who lived in a residential center for people with ID
- Age range: 16 to 69 years

Characteristics of participants

- 80% participants had **borderline to mild intellectual disability**, and >10% had moderate to severe intellectual disability
- 65% had **low adaptive functioning** and handle things like a 9- to 11-year-old child
- 52% had at least 1 mental health issue or neurodevelopmental disorder

Functional outcome of participants

- 37% lived with parents or family, 21% lived with spouse/their own family, and 11% lived independently
- Only 7.8% worked full-time, while **40% worked part-time or had an adjusted job in a protected environment**
- 76% were single, and 20% were married or in a long-term relationship

Keep in mind: over 20% of the participants in the study were recruited from a live-in facility for people with intellectual disability and may have lower functioning.

Cognitive functioning – ability of the brain to think, learn, understand and use language, remember, pay attention, reason, and make decisions

Adaptive functioning – personal and social skills to go through daily life and respond to demands from the environment. E.g. communication and social participation

Intellectual disability (ID) – a condition related to the brain / nerves that affects cognitive and adaptive functioning

Predictors of adaptive functioning

↑ **Overall adaptive functioning**
- Being female
- ↑ FSIQ score
- No autism spectrum disorder (ASD)

↑ **Communication**
- ↑ FSIQ score
- ↓ Anxiety
- No ASD

↑ **Socialization**
- Being female
- ↑ FSIQ score
- No ASD

↑ **Daily living skills**
- ↑ Age
- Being female
- ↑ FSIQ score
- No ASD

These do NOT predict adaptive functioning levels
- Depression
- Psychosis
- ADHD

Main message

- It is recommended that adults with 22q11.2DS have **adequate and up-to-date assessments** of their **IQ**, **adaptive functioning**, **functional outcomes** (living situation, job), and **mental health**.
- Having a complete profile lets the adults, their caregivers, and their healthcare providers **set realistic goals** to improve functioning. If necessary, the healthcare professionals can suggest **interventions**.
- Each adult with 22q11.2DS is different, and the **support needs to be tailored**.

22q Differences and Brain Processes

Distinct neurocognitive profiles and clinical phenotypes associated with copy number variation at the 22q11.2 locus

O'Hora KP, Kushan-Wells L, Schleifer CH, Cruz S, Hoftman GD, Jalbrzikowski M, Gur RE, Gur RC, Bearden CE. Autism Res. 2023 Dec;16(12):2247-2262. doi: 10.1002/aur.3049. Epub 2023 Nov 23. PMID: 37997544; PMCID: PMC10872774.

This study aimed to look at how individuals with 22q differences perform in terms of thinking, reasoning, planning, switching between tasks, responding to situations, self-inhibiting, and remembering things. Using a range of tests, the researchers examined neurocognitive profiles for **22q11.2DS** and **22q11.2DupS**.

Neurocognition are the brain processes that allow people to **think** and **reason**. These processes include the ability to stay focused, remember facts, understand, and use information, etc.

A **neurocognitive profile** is the overall big picture of the strengths and weaknesses of a person's abilities to think and reason.

Assessment Methods

- The online **Penn Computerized Neurobehavioral Battery (CNB)** – this test has been used for cognitive testing in different populations, including in individuals with **22q11.2DS**. It assesses memory, attention, reasoning, motor speed and social cognition..
- Additional interviews and scales of mental health risks, functioning, behavior, and autism symptoms.

Study participants

- 126 individuals who have been participating in studies at the University of California at Los Angeles (UCLA)
- Participants include:
  - 55 with confirmed **22q11.2DS**
  - 30 with confirmed **22q11.2DupS**
  - 41 individuals from the general public with similar age and social characteristics as the test groups
- Age range: 4 to 50 years

Neurocognitive Profiles of 22q11.2DS & **DupS**

- The neurocognitive profiles were different for individuals with **22q11.2DS** vs. **22q11.2DupS**. The differences can be used to tell apart their genetic condition.
- Some individuals with **22q11.2 deletion syndrome** showed **decreased accuracy in memory** tests.
- Some individuals with **22q11.2 duplication syndrome** showed a **decreased speed in processing information**, especially related to social situations.
- Among individuals with **22q11.2DupS** (but not those with **22q11.2DS**):

  ↓ **Social cognition speed** was associated with
  - ↑ severe mental health issues
  - ↓ overall functioning
  - ↑ social problems
- While typically-developing individuals performed better as they got older, individuals with **22q11.2DS** and **22q11.2DupS** **did not show an age-related improvement**.
- Among individuals with autism, those with **22q11.2DS** were less accurate on memory and complex thinking compared to those with **22q11.2DupS**.

Main message

- Individuals with **22q11.2 deletions** and **duplications** perform differently compared to those without 22q11.2 differences. They also show unique patterns of cognitive strengths and weaknesses.
- Individuals with **22q11.2DupS** are more accurate but are slower than those with **22q11.2DS**.
- Faster social cognition speed in those with **22q11.2DupS** may relate to better clinical outcome.

We appreciate your financial support!

Our mission: to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships

At the International 22q11.2 Foundation, we help families that need important resources and information to meet the needs of loved ones with 22q. We work with top medical experts from around the world to build our understanding of 22q, improve treatment options for families, and conduct the necessary research for longer, healthier lives for patients.

Your support makes a difference!

Please visit the **Donate** page on our website for information on donating online, by mail, or via other methods. **Thank you**!

The mission of the **International 22q11.2 Foundation** is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships. This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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