2023

FREE WIFI

5TH EUROPEAN 22Q11 CONFERENCE DUBLIN

18TH - 19TH NOVEMBER 2023

Conference PROGRAMME

22q11 Europe

Fáilte Ireland

22q11 IRELAND

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WELCOME

Dear Participants,

As President of 22q11 Europe, I am delighted to welcome you to the European Conference on 22q11 Syndrome in Dublin, taking place on November 18th and 19th, 2023. We are organising this conference in collaboration with 22q11 Ireland, and together, we represent a united front for patients' associations across Europe. Our primary goal for this conference is to promote dialogue and collaboration between patient associations and the scientific community.

We aim to create an environment where both groups can share insights, findings, and experiences related to 22q11 syndrome. This event is a significant step towards building stronger connections and accelerating progress in researchand support for 22q11patients. I would like to express my heartfelt gratitude to our speakers and my warmest thanks to 22q11 Ireland, our host organisation and, in particular, to the organisation team. Over the next two days, I encourage you to engage, learn, and connect. Let's seize this opportunity to inspire progress, raise awareness, and work towards a more inclusive and supportive world for individuals living with 22q11 syndrome.

Welcome to Dublin and thank to each of you for your participation in the European Conference on 22q11 Syndrome. Your presence exemplifiesyour commitment to improving the lives of individuals with22q11 syndrome.
Thank you

Thank you.
Paul Havelange
22q11 Europe President

Dear Participants,

It is with great pleasure that we welcome everyone to Dublin for a second time to a 22q European conference. First time conference attendees can expect to learn a great deal about 22qDS, to share their reasons for being with us and to find new friends. We have so much to learn from each other! For those of us who attend these events on a more regular basis it is a time to catch up with old friends and to learn about what is new with 22q. Whatever the reason for attending, we hope that you find the conference informative and that you enjoy your stay with us here in Dublin.

With warm regards from Anne and 22q Ireland.

Thank you,
Anne Lawlor
Chairperson 22q11 Ireland

Anne Lawlor
Chairperson 22q11 Ireland

Dear Participants,

Welcome to the next European Conference on 22q11.2 Deletion Syndrome, an event dedicated to the most recent advancements in our understanding of this complex genetic disorder. As the Scientific Coordinator of this conference, I am thrilled to introduce our comprehensive program, which aims to bridge the gap between cutting-edge research and practical insights.

Our primary mission is to bring together families of individuals affected by 22q11.2 deletion syndrome and professionals working in the field, fostering collaboration and knowledge exchange. Over the two days, we will therefore hear insights from internationally renowned experts and young researchers in the field, as well as from parents and people affected by 22q11.2 deletion syndrome. We understand that 22q11.2 deletion syndrome presents unique challenges that evolve with age. Therefore, our program is divided into two streams to address the diverse needs of our attendees. The first stream will be dedicated to pediatric concerns and will cover various topics, such as family support, medical management, speech and language development, and educational strategies. The second stream will delve into the topics of independent living, entering the work environment, and mental health.

We are glad to welcome you in Dublin this November, where the exchange of knowledge and support will contribute to a better quality of life for individuals affected by 22q11.2 deletion syndrome and their families. Together, we can make a difference.

Pr. Maude Schneider
Scientific Coordinator of 22q11 Europe

Pr. Maude Schneider
Scientific Coordinator of
22q11 Europe

OUR TEAM

Jess Freeman

Magdalena Wojciech

Dr Aoife Lynam

WELCOME

We would like to welcome you all to Ireland and we hope you enjoy the conference.

DUBLIN

**A city that is as intimate as a village
and as friendly as an Irish pub.
Framed by mountains, centered on a river
and edged by a beautiful bay, the city's streets
and alleys are filled with vibrant art and historic buildings, hip cafés and traditional "old man" pubs, as Dubliners call them. Walk the streets and you'll feel the energy of over 1,000 years of history, as echoes of the Vikings mix with buzzing boutiques, cobbled streets reverberate with the sounds of buskers, and 18th century parks play host to festivals, film and food markets.
There are plenty things to see and do in Dublin.**

**Please see tourists attraction websites:
https://planmyhyattstay.com/
https://www.visitdublin.com/things-to-do
https://www.ireland.com
https://www.whereindublin.com**

The River Liffey
divides the city into the Northside and the Southside, with the city centre straddling the two. Though the centre of Dublin is fairly small, the broader city is made up of a series of villages, from the central neighbourhoods like Portobello and the Docklands, to the coastal districts of Sandymount and Clontarf. To the north, you'll find two of the city's oldest neighbourhoods, Stoneybatter and Smithfield, with cool coffee shops and gastropubs. In the south, the suburbs of Rathmines and Ranelagh are great for café hopping, brunch and people watching in the Georgian squares

ABOUT DUBLIN

It is also worth noting that the city is divided into numbered postal districts, from 1 to 24. All even numbereddistricts are South of the Liffey and all odd-numbered ones are to the North. Dublin 1 is the area around and including O'Connell St, while Grafton Street and the South city centre are in Dublin 2. The sole exception is Dublin 8, which extendsfrom the Liberties to the North of the river and includes part of the area around the Phoenix Park.

WHAT TO SEE IN DUBLIN

Trinity College Dublin

Is Ireland's highest ranked university, and one of Dublin's most popular visitor attractions, due in equal measure to its history and
architecture. A walk through the cobbled stones of Trinity College Dublin will bring visitors back to the 18th century, when the magnificent Old Library building was constructed. Inside is housed the Book of Kell’s - a 9th-century gospel manuscript famous throughout the world.

Phoenix Park

Is one of the largest public parks in any capital city in Europe, comprising 1750 acres. It was originally formed as a royal hunting Park in the 1660s and opened to the public in 1747. A large herd of wild fallow deer remainto this day. There are over 14 kilometres (9 miles) of paths to explore.
www.phoenixpark.ie

Guinness Storehouse

Tells the story of Ireland's most famous export, the Guinness beer, from its humble beginnings in 1759 to its global recognition today. The Storehouse is not only the home of Guinness but also a celebration of Irish culture, creativity, and entrepreneurship
www.guinness-storehouse.com/

TRANSLATION SERVICE

Notes for the Presenters:

Info for the attendees of the conference in relation to Translation services:

The following languages are available:

- Arabic
- Chinese Simplified
- Chinese Traditional (Preview)
- Czech
- Danish
- Dutch (Preview)
- English
- Finnish
- French
- French (Canada)
- German
- Greek
- Hebrew
- Hindi
- Hungarian (Preview)
- Italian
- Japanese (Preview)
- Korean (Preview)
- Norwegian
- Polish (Preview)
- Portuguese (Brazil)
- Portuguese (Portugal)
- Romanian (Preview)
- Russian (Preview)
- Slovak (Preview)
- Spanish
- Swedish
- Thai (Preview)
- Turkish (Preview)
- Ukrainian
- Vietnamese (Preview)

To make sure your live captions are as accurate as possible, try to follow these best practices:

• Speak clearly, slowly, and directly into the mic. As your distance from the mic increases, captions may become less accurate.
• Avoid having multiple people speak at the same time.
• Make sure if someone is speaking for example a question from the audience that they speak into a connected microphone

To provide captions on the large screen in English please follow the same instructions listed below under “On the day of the event (Attendee)”

TRANSLATION SERVICE

Info for the attendees of the conference in relation to Translation services:

If you require translation services you will need to take the below actions:

• Provide us with your email address ahead of time.

o This will allow us to send you the invite to the Teams meeting, if you have provided your email address but have not received the Teams meeting invite before the event, please check your spam folder, failing this please speak to a volunteer upon your arrival.

• Bring your own device that has internet connectivity

• Bring your own headphones

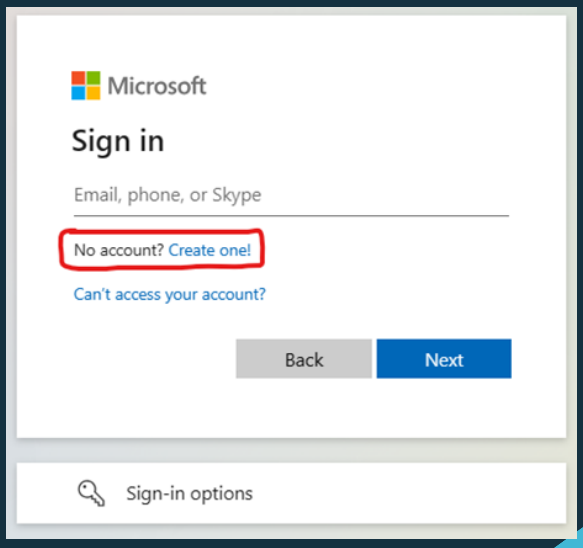
• Install Teams on your device.

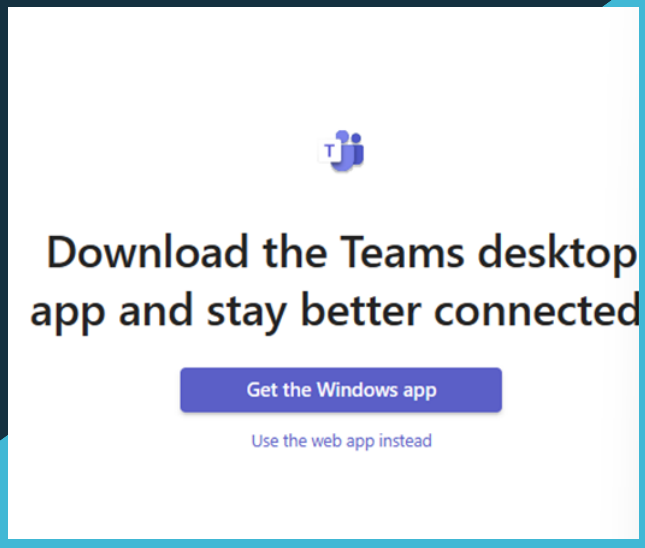
o If this is an apple or android device please download the app from the app store

o If this is a laptop you can download Teams at: https://teams.microsoft.com/

 You can log in with a Microsoft account such as hotmail.com or outlook.com. If if you don't have one you can use the create an account link and use your non Microsoft email address, your contact number OR create a new Microsoft email address

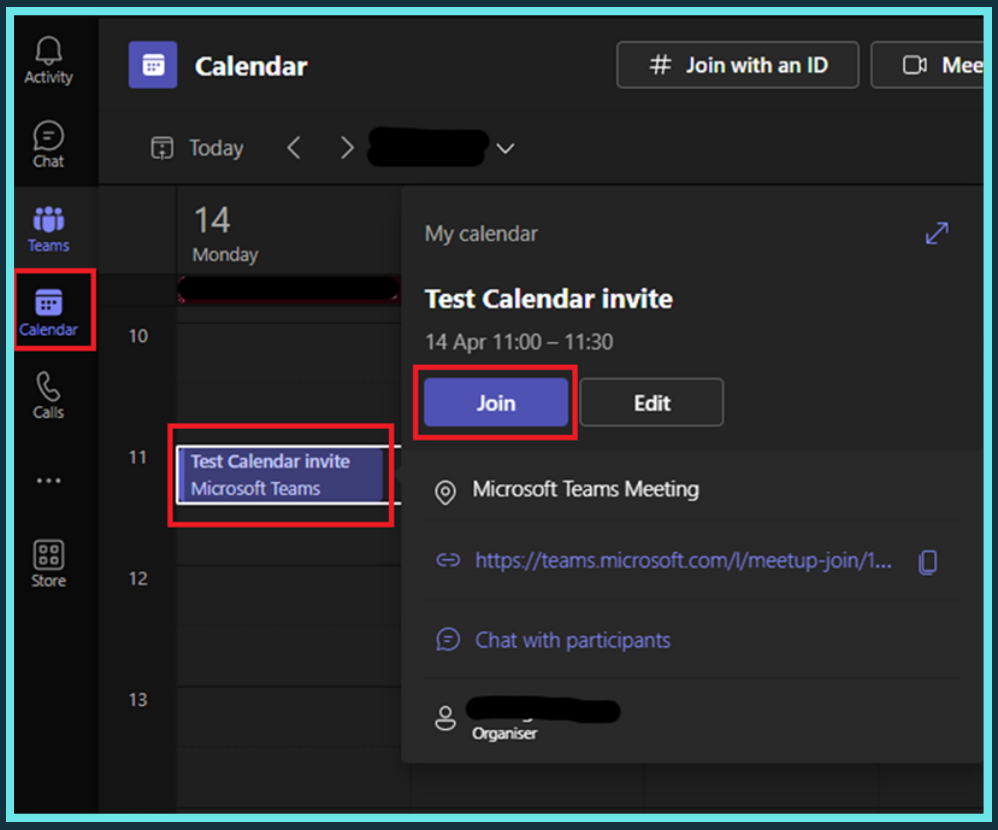
 Once you are logged in, follow the on screen instructions and then choose "Get the Windows app"



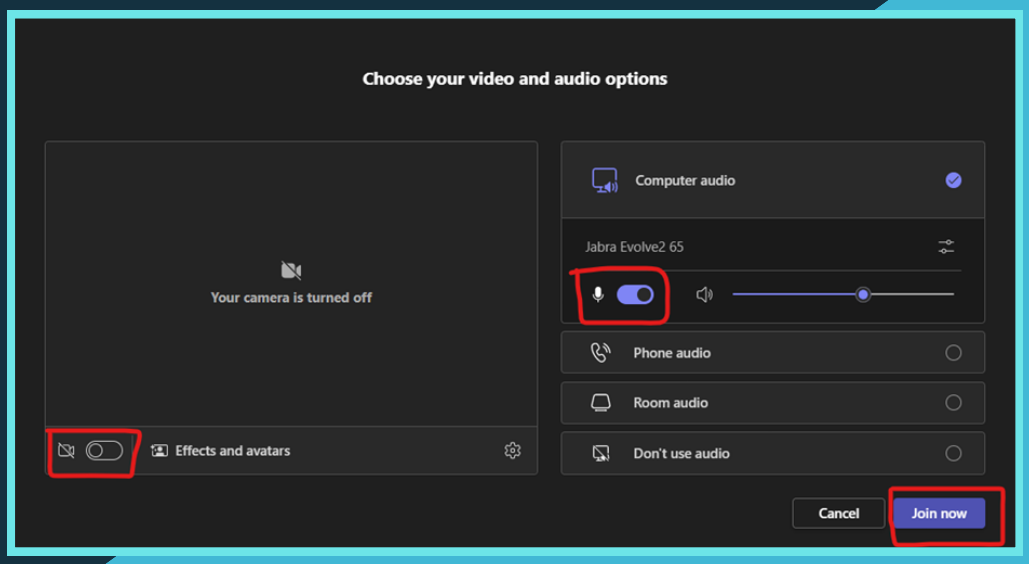


On the day of the event (Attendee)

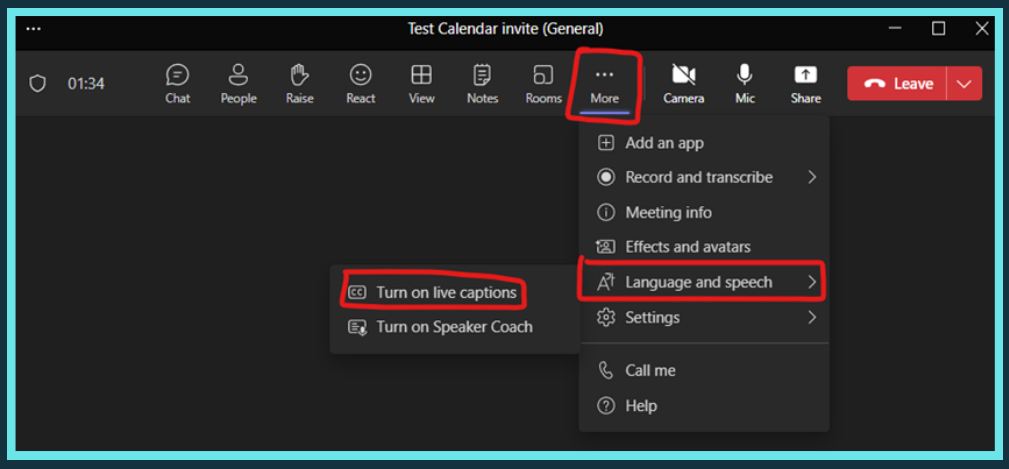
1. Using your device, open the Teams app, (Log in if you have not already done so and connect your headphones)
2. Navigate to the Calendar tab and locate your invite, click on the invite and select “Join”



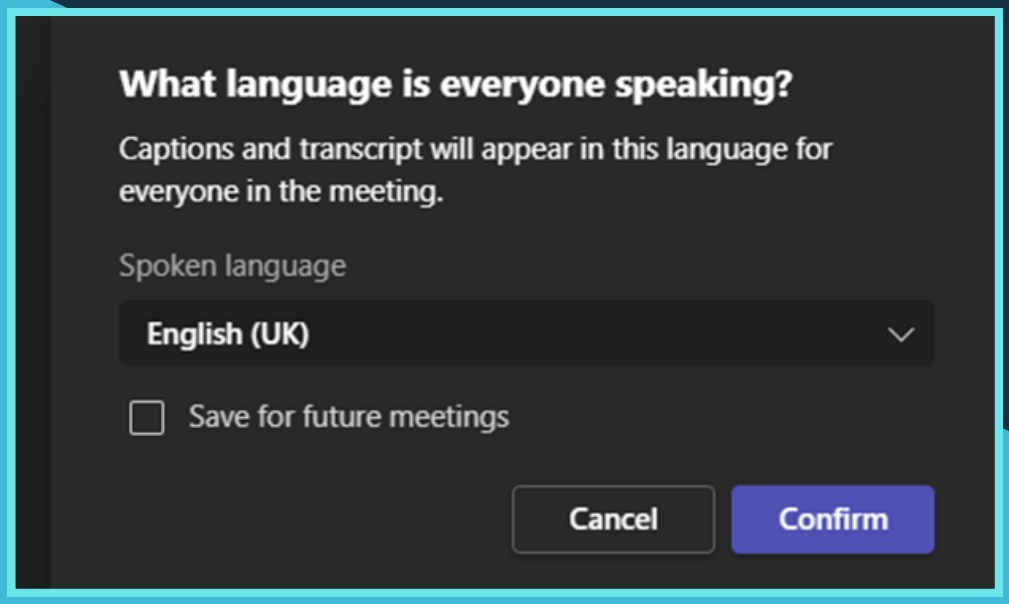
3. Toggle your mic and camera to off and select “Join now”



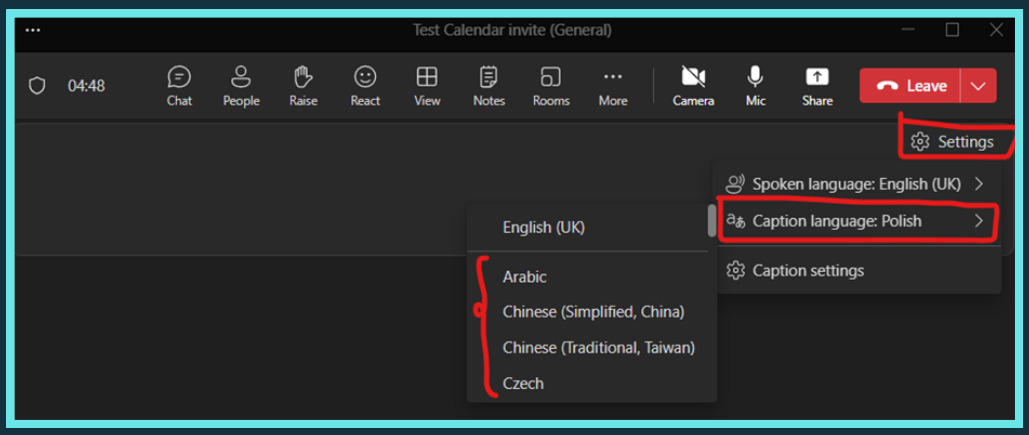
4. Select “… More” followed by “Language and Speech” followed by” Turn on Live captions”



5. If asked “What Language is everyone speaking?” please select English (UK) and choose Confirm



6. Once this is completed a new box will appear either at the top or the bottom of the screen. Once visible, click on “Settings” followed by “Caption Language” and finally choose your language.



Please Note:

Microsoft Teams technology strives for high accuracy in its operations; however, variations in dialects might occasionally result in errors, which is a common occurrence with most online technologies. Despite its best efforts to be precise, the platform may encounter challenges in understanding diverse linguistic nuances, leading to occasional inaccuracies in interpretation or transcription.

AGENDA

Day One: 18th November 2023

Time	Presentation	Speakers
9:00am	Everyone together.	
9:15am	Opening words.	*Paul Havelange, Stephan Eliez, and Anne Lawlor*
9:30am	General introduction.	*Maude Schneider*

Location: Room One (0–15 years old)		
Time	**Presentation**	**Speakers**
10:10am	Parent perspective: "What I know now that I would have liked to know when my child was younger."	*Vesna Vujicic*
10:25am	Opening presentation: What is 22q11DS and new guidelines for the management of children and adolescents.	*Donna McDonald-McGinn*
11:05am	**Break**	
11:30am	Care Coordination: a transferable model of care.	*Suzanne Kelleher and Wesley Mulcahy*
12:00pm	SESSION A - Short presentations (6x8' talk + 10' Q&A).	Samuel Chawner Ruth Garcia-Rodriguez and Charlotte James Jill Arganbright Ahmed Khan Dieuwke Mink van der Molen Jente Verbesselt
13:00pm	**Lunch**	
14:00pm	Speech and language and communication issues.	*Nicole Prendeveille*
14:25pm	Cognitive difficulties and impact on daily life.	*Ania Fiksinski*
14:50pm	Entering school.	*Louise Bottcher*
15:20pm	**Break**	
16:00pm	WORKSHOP - Q&A (language, cognition, development & school)	*Ania Fiksinski, Donna McDonald-McGinn, and Suzanne Kelleher*

Location: Room Two (16+ years)		
Time	**Presentation**	**Speakers**
10:10am	Parent perspective: "What I know now that I would have liked to know for the transition process to adulthood."	*Anne Lawlor*
10:25am	New guidelines for the management of adults with 22q11DS.	*Erik Boot*
11:05am	**Break**	
11:30am	SESSION. B - Short presentations (6x8' talk + 10' Q&A)	Robin de Croon Farnaz Delavari Silas Forrer Maria Gudbrandsen Julie Husmann Caren Latrèche
12:00pm	Towards coordinated care for adults (round table).	
13:00pm	**Lunch**	
14:00pm	Experience from the YEEP (Young Experts by Experience Panel) Group.	*YEEP Members*
14:25pm	Promoting autonomy.	*Maude Schneider*
14:50pm	From literature review to co - production: The public patient involvement (PPI) journey with the patient organization 22q11 Ireland.	Lorna Kerin
15:20pm	**Break**	
16:00pm	WORKSHOP - Q&A (cognition, autonomy, & work).	*Maude Schneider, Erik Boot, and Wesley Mulcahy.*

Time	Presentation	Speakers
16:30pm	Everyone together in the main room	
16:30pm	The importance of self-care for parents.	*Sasja Duijff*

End of Day One

Day Two: 19th November 2023

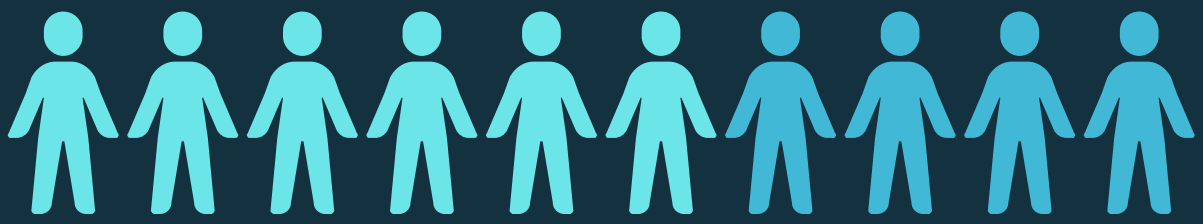
Location: Room One (0–15 years old)		
Time	**Presentation**	**Speakers**
9:00am	How to take care of the whole family.	*Maria Caples*
9:30am	Psychiatric issues in children and adolescents.	*Franziska Radtke*
10:00am	WORKSHOP - Q&A (family & psychiatry).	*Fraziska Radtke, Maria Caples, and Fiona Mc Nicholas*
10:30am	**Break**	
11:00am	SESSION C: Short presentations (5x8' talk and 10' Q&A)	Jill Arganbright Femke van den Helder Rae Lyz Yee Hayfa Soobratty and Fiona McNicholas *Jacob Vorstman*

Location: Room Two (+16 years)		
Time	**Presentation**	**Speakers**
9:00am	Psychiatric issues in adults.	*Boris Chaumette*
9:30am	Medication and supplements: Facts and fiction.	*Stephan Eliez*
10:00am	WORKSHOP - Q&A (psychiatry)	*Stephan Eliez and Boris Chaumette*
10:30am	**Break**	
11:00am	Short presentations (5x8’ talk and 10’ Q&A)	Michiel Houben, Marta Sousa Santos, and Carina Sauter

Time	Presentation	Speakers
11:45am	Everyone together	
11:45am	What's new in 22q11 and novel perspectives.	*Francesco Papaleo*
12:15pm	The interplay between genetics and mental health.	*Jacob Vorstman*
12:45pm	Sleep issues.	*Corrado Sandini and Sam Chawner*
13:15PM	Closing words.	*Paul Havelange*

13:30pm Conference Finished

SPEAKERS



DR ANIA M. FIKSINSKI, PH.D.

Clinical psychologist
University Medical Center Utrecht

Dr Ania M. Fiksinski, Ph.D.

Dr Ania Fiksinski (Ph.D.) is a psychologist and researcher based in the Netherlands. She is currently working as a psychologist and postdoctoral researcher at the Wilhelmina Children's Hospital, University Medical Center Utrecht (the Netherlands).

Ania obtained her PhD degree in February 2021, which focused onindividuals with 22q11.2 deletion syndrome.
Ania remains closely affiliated with The Dalglish Family 22q Clinic (Canada) and Maastricht University. She acts as an Advisor to The 22q11.2 Society and has close links to national and international patient advocate organisations. Her research focuses on understanding cognitive and behavioral trajectories in the context of a pathogenic genetic variant such as the 22q11.2 deletion

DR BORIS CHAUMETTE, MD, PH.D.

Associate Professor at Paris Cité University
Adjunct Professor at McGillUniversity

Dr Boris Chaumette, MD, Ph.D.

Dr Boris Chaumette (MD, Ph.D.), defended his thesis in psychiatry and his thesis in neurobiology in 2016 in Paris. He is an Associate Professor at Paris Cité University and Adjunct Professor in the Department of Psychiatry at McGill University (Montreal, Canada). Since 2019, he leads the Reference Center for Rare Psychiatric Diseases at GHU Paris Psychiatry and Neurosciences. This center provides psychiatric support for patients with a rare disease, includingpatients with deletion 22q11. His research activities focus on the genetics and epigenetics of psychiatric disorders at the Institute of Psychiatry and Neurosciences of Paris (INSERM U1266) and aim to better understandGene x Environment interactions in the emergence of psychosis in adolescence.

DONNA M. MCDONALD-MCGINN, MS, LCGC

Professor of Clinical Pediatrics
University of Pennsylvania

Donna M. McDonald-McGinn, MS, LCGC

Donna M. McDonald-McGinn is a Professor of Clinical Pediatrics at the Perelman School of Medicine of the University of Pennsylvania and serves as Chief of the Section of Genetic Counseling, Associate Director of Clinical Genetics, Research Scientist, andDirector of the22q and YouCenter at the Children's Hospital of Philadelphia. Prof. McDonald-McGinn is a pioneering leader in holisticcare for individuals with chromosome 22q11.2 differences and related conditions. As a Founding Board Member of the International 22q11.2 Foundation and Founding Trusteeand current Chairof the 22q11.2 Society, Prof.McDonald-McGinn has spent her career supporting education, multidisciplinary care, collaborative outcomes research, and family-centered rhyming awareness and friendship buildingevents such as 22q and Boo, 22k for 22q, and 22q at the Zoo.

MS ELISHA MINIHAN

Researcher with University College Dublin
Group Facilitator with 22q11 Ireland

Elisha Minihan

Elisha Miniham holds a Bachelor of Arts joint honours degree in Psychology and Philosophy from the National University of Ireland Galway, and a Masters in Psychology from the University of Limerick. Elisha is currently working as a Psychology Assistant withthe Health ServiceExecutive (HSE) in Primary Care Psychology. Additionally, Elisha has beenworking as a researcher with University College Dublin from 2019 to present and has published both quantitative and qualitative research. In January 2023, Elisha startedthe position of group facilitator for 22q11 Ireland YEEP group.

DR ERIK BOOT MD, PH.D.

Physician for individuals with
an intellectual disability

Dr Erik Boot MD, Ph.D.

Dr Erik Boot is a physician specialised in intellectual disability medicine and works at the multidisciplinary 22q11.2 clinics for adults at ‘s Heeren Loo and Maastricht, the Netherlands. He obtained his MD at the University of Amsterdamin 1999, completed his medical specialty training at the Erasmus University in Rotterdam in 2004, and completed his Ph.D. thesis on adults with 22q11.2 deletionsyndrome at the University of Amsterdam in 2010. From 2014 to 2016, Dr. Boot trainedas a post-doctoral fellow at The Dalglish Family 22q Clinic, Toronto, Canada; an interdisciplinary clinic devoted to adults with 22q11.2deletion syndrome and their families. Dr. Boot has (co-)authored over 80 peer-reviewed journal articles and book chapters; the majority related to microdeletion 22q11.2. He is a member of the scientific advisory committee of the Dutch 22q11.2 family network (Stichting Steun 22Q11), and advisor for the 22q11.2 Society.

DR CORRADO SANDINI

Resident in Child and Adolescent Psychiatry
Foundation Research Fellow, University of Geneva

Dr Corrado Sandini, MD, Ph.D.

Dr Corrado studied medicine at the Universityof Genova, Italy. He pursued a Ph.D. in Neuroscience at the University of Geneva focusing on the developmental of computational approaches, namely dynamic network analysis, to characterise neurodevelpmental and clinical pathways of vulnerability to psychosis in 22q11DS. Since his Ph.D., Corrado has divided his time as a resident in child and adolescent psychiatry and research activity.Dr Corrado has been supported by a clinical scientist grant of the NNCR Synapsy to study the role of sleep disturbances in 22q11DS. Corrado has recentlybeen awarded the SNSF Ambizione Fellowship, to study the contribution of sleep disturbances in contributing to affective comorbidities in ADHD using an ecological digital phenotyping approach and network analysis techniques.

PROF. FIONA MCNICHOLAS

Consultant Child & Adolescent Psychiatrist
Full Professor of Child & Adolescent Psychiatry
University College Dublin

Prof. Fiona McNicholas

Prof McNicholas, a Consultant in Child and Adolescent Psychiatry in Lucena Clinic, Rathgar and Our Lady'sHospital for Sick Children, Crumlin, has a diverse background. Trained at Guys Hospital in Psychiatry and Great Ormond Street Hospital, London, she pursued a research fellowship in Stanford University and returned as visiting Professor. She was Assistant Professor at Columbia University prior to her appointment as full professor and academic lead in University College Dublin. Prof. McNicholas' main clinical work is eating disorders, 22Q11DS, and acute psychiatry presentations via her role in paediatric liaison psychiatry. She is interested in increasing knowledge about, access to, and delivery of, evidence-based services within child and adolescent mental health services (CAMHS), along with clinician wellbeing. She contributes to postgraduate training within psychiatry and other disciplines. She provides regular information sessions for families,schools and hosts a UCD weekly webinar on mental health.She is recipient of many researchgrants and has published over 300 articles, book chapters and co-authored a book on mental health in child and adolescents, a guide for teachers, with an upcomingbook for GPs.

DR FRANCESCO PAPALEO

Senior Researcher and Group Leader
Genetics of Cognition laboratory
Istituto Italiano di Tecnologia

Dr Francesco Papaleo

Dr Francesco Papaleo (born in Scicli, RG, November (1977) is a tenured senior researcher, group leader of the Genetics of Cognition laboratory, at the Istituto Italiano di Tecnologia (IIT), Genova, Italy.The goal of our research effort is to investigate the mechanisms underlying social and cognitive processes which are altered in neurodevelopmental and psychiatric disorders. To reach this goal, his laboratory uses a cross-disciplinary approach includingdetailed studies in genetically modified mice and parallel clinical investigations. Dr Papaleo spearheaded his line of research through previous researchexperience at the University of Padova (Italy, 1996-2002), University of Bordeaux (France, 2002-2005) and at the National Institute of Mental Healthin Bethesda (USA,2005- 2010).

FRANZISKA RADTKE, MD

Head of child and adolescent psychiatric special outpatient unit
Centerfor Deletion & Duplication Syndrome 22q11

Franziska Radtke, MD

Dr Radtke is a physician at the Clinic for Child and Adolescent Psychiatry, Psychosomatics and Psychotherapy at theUniversity Hospital of Würzburg. She specialises in caring for patients and their families within the interdisciplinary Centerfor Deletion and Duplication Syndrome 22q11 (ZEDE22q11). Dr Radtke provides counselling, diagnostics and,if necessary, therapyregarding possible mental illnesses. Beyond her clinical work, Dr Radtke actively contributes to research on the causes of the syndrome, from the gene to the cell to the disease,Dr Radtke is enthusiastic about sharing the latest scientific insights and findings with others.

PROF. LOUISE BØTTCHER

Associate Professor, Aarhus University

Prof. Louise Bøttcher

Prof. Louise Bøttcher is a trained psychologist but has worked as a researcher since 2005. Her research interest has focused on the interplay between disability and social and cultural conditions for development. More specific projects have focused on learning with cerebral palsy, the role of social condition for communication technologies used in augmented and alternative communication and development of independence in young people with severe disabilities. A key publication is Bøttcher & Dammeyer (2016). Development and learning of young children with disabilities. A Vygotskian perspective. Springer: New York.

DR SAMUEL CHAWNER

Medical Research Foundation Fellow, Centre for Neuropsychiatric Genetics and Genomics, Cardiff University

Dr Samuel Chawne

Dr Samuel Chawner is a researcher specialising in mental health amongst those with 22q11.2 Deletion Syndrome. Using his extensive background in conducting longitudinal cognitive and mental health investigations, Dr Chawner's research has centred on understanding the relationship between neurodevelopment and mental health outcomes among individuals with 22q11.2 Deletion Syndrome. His research has extended into sleep studies and early development, where he actively seeks to identify early indicators and markers of mental health challenges. Beyond his research endeavours, Dr Chawner has a strong commitment to public engagement and is a prominent and frequent speaker at conferences and workshops tailored to clinicians, researchers, and families affected by 22q11.2 Deletion Syndrome, actively sharing his expertise and insights in the field.

DR MARIA CAPLES

Lecturer in Intellectual Disability Nursing
University College Cork

Dr Maria Caples

Dr Maria Caples is a lecturer in Intellectual Disability Nursing at the School of Nursing and Midwifery, University College Cork. Her professional journey commenced in 1995 when she obtained her qualification as a Registered Nurse in Intellectual Disabilities. Following her registration, she gained experience as a staff nurse and clinical nurse manager until 2008, when she transitioned to University College Cork.Currently, Dr Caples is actively engaged in teaching across the BSc undergraduate nursing programmes. She holds a Ph.D. in Nursing, and her doctoral research explored the relationship between resilience factors and adaptation within families raising children with 22q11.2 Deletion Syndrome. Dr Caples' research interests encompass a broad spectrum, including the quality of life of parents and families caring for children with intellectual disabilities, the physical health requirements of individuals with intellectual disabilities, respite care, the intersection of aging and intellectual disability, and the resilience of families, with a particular emphasis on rare diseases. Notably, she is currently a member of a team contributing to an Erasmus+ Partnership initiative that explores Genomic Literacy and Ethical Competence in Nursing Education.

DR MAUDE SCHNEIDER

Assistant Professor
University of Geneva

Dr Maude Schneider

Dr Maude Schneider is assistant professor at the Facultyof Psychology and Educational Sciences of the University of Geneva since 2019 where she heads the Clinical PsychologyUnit for Intellectual and Developmental Disabilities.

Dr Maude Schneider is working as a clinician and researcher in the fieldof neurodevelopmental disorders. Since 2009, she is actively involved in the 22q11DS Geneva longitudinal cohort and is also conducting research on autism spectrum disorder. Using a combination of clinical and cognitive approaches as wellas digital phenotyping, her research aimsto explore the mechanisms underlying social and mental health difficulties of adolescents and young adults with neurodevelopmental conditions. She is also the scientificcoordinator of 22q11Europe and advisor for the 22q11.2 society.

MS NICOLE PRENDEVILLE

Team Leaderand Clinical ExpertSpeech and LanguageTherapist

Nicole Prendeville

Nicole Prendeville is a Speech and Language therapist with 18 years of experience working in paediatrics in the UK and the Republic of Ireland. As part of her rolewithin the Cleft Lip and Palate service at Great Ormond Street Hospital, she has worked with children and young people across the lifespan in the designated 22q11 clinic, in her own Speech and Language clinic and within the Velopharyngeal Investigations service. Nicole have worked with babies who have been diagnosed with 22q11D.S. to support their feeding and swallowing difficulties; with most of her work evaluating the communication skills of children during the preschool, school-age, and teenage years. Nicole has coordinated and taught the Cleft Palate modules on SLT undergraduate and Masters programmes at City University London and UCL as well as teaching/training community SLTs which has includedcase studies of children with 22q11D.S. with speech velopharyngeal dysfunction and speech disorders to increase awareness of this conditionin student SLTs.

DR SASJA DUIJFF, PH.D.

Psychologist | Infant Mental Health specialist | Mindfulness trainer VMBN cat.1

Dr Sasja Duijff, Ph.D.

Dr Sasja Duijff currently works in her own private practice, Care4Minds, and is a guestresearcher at the Radboud University Center for Mindfulness. She has been working with children with 22q11DS and their parentssince 2003 (of which 15 years at the 22q11 outpatient clinic at the Wilhelmina Children's Hospital, UMC Utrecht, The Netherlands). In her conversations with parents, she became aware of the challenges that parents with a child with medical complexity such as 22q11DS, face. This inspired her to adapt the evidence-based Mindful Parenting Training (by Susan Bögels) with themes that are pertinent to parents with a child with medical complexity. 'Care4Parents' is an accessible, practical training in which parents learn to recognize, understand, and manage stress in themselves and their response to it.

PROF. STEPHAN ELIEZ

Medical Doctor, Child & Adolescent Psychiatry
Professor, University of Geneva
Director, Fondation Pôle Autisme

Prof. Stephan Eliez

Professor Stephan Eliez is a medical doctor in child and adolescent psychiatry, a professor at the University of Geneva's School of Medicine (Department of Psychiatry), and the director of the Fondation Pôle Autisme. Prior to founding the Fondation Pôle Autisme, he held the position of director for 15 years at the Office médico-pédagogique of the State of Geneva, a public institution specialising in special education and child psychiatry services. Within his area of research, Professor Eliez is renowned for his specialization in cutting-edge brain imaging techniques. He leverages these advanced methodologies to gain deeper insights into the intricate relationship between cerebral structure and function, as well as the origins of learning difficulties and psychological conditions in children. His focus is particularly on children afflicted with conditions such as 22q11 deletion syndrome and autism. Following his medical education, Professor Eliez pursued further training in the field of neuroscience at Stanford University in California, where he spent a five-year period of study and research

DR SUZANNE KELLEHER

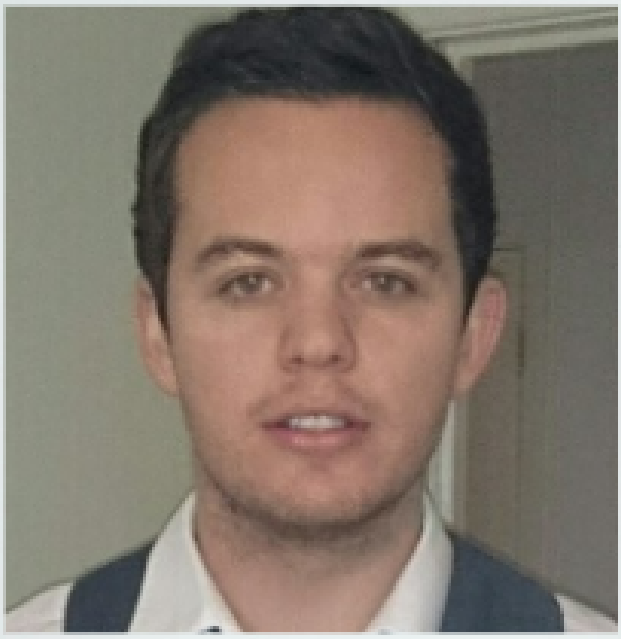
Consultant Pediatrician at Children's Health Ireland, with a special interest in Complex MedicalCare

Dr Suzanne Kelleher

Dr Suzanne Kelleheris a Consultant Pediatrician at Children's Health Ireland, with a special interest in Complex Medical Care. After graduating in 1992, Dr Kellehertrained in Ireland,Australia and the United Kingdom in General and Community Pediatrics. Dr Kelleher has worked as a consultantin the Central Remedial Clinic, Enable Ireland and in the Developmental Clinic in the Coombe Women and Infants University Hospital. Dr Kelleher is an honorary lecturer with RCSI and is a Fellow of the Royal College of Physicians in Ireland. Dr Kelleher sees all GeneralPediatric and developmental referrals, and has a special interest in Complex Medical Needs as well as having set up a multidisciplinary 22q11 clinic at CHI.

MR WESLEY MULCAHY

Clinical Specialist Occupational Therapist
Children's Health Ireland



Wesley Mulcahy

Wesley Mulcahy is a Clinical Specialist Occupational Therapist at Children's Health Ireland(CHI). After undertaking studies in Social Work, he completed his Masters in Occupational Therapyin 2010 and his Masters in Advanced ClinicalPractice in 2021.Wesley has worked in Enable IrelandDisability Services, specialising in Early Intervention before transferring to acute care,and was the senior therapist on the Paediatric Neurology and Neonates teams. Wesley was recently appointed the Complex Care Coordinator for children with 22q.11 DS,an innovative post,tasked to improvethe lives of children, young people and their families living with this rare disease.

YEEP GROUP

The YEEP group is made up of young adults with 22q who provide mutual support, forming a community of individuals with expertise gained through personal experiences. During the YEEP workshop on mental health and anxiety, two members, Áine and Ciara, will share their personal journeys with mental health. Their insights will foster an open and constructive conversation on these important topics.

SHORT ORAL PRESENTATIONS

Hearing loss in patients with 22q11.2 deletion syndrome: a review of 689 patients

Authors: JJill Arganbright, MD, Blaine (Terrance) Crowley, Meghan Tracy, CCRC, Janelle Noel-Macdonnell, PhD, Kim Gaiser, Lori Yaktine,AuD, Amanda Moore, AuD, Jamie Hamm, AuD, Bernice MarrowPhD, Hansoo Song,Srivats Narayanan, NikitaRaje, MD, Donna McDonald-McGinn, MS, CGC

Background: Hearing loss is considered common in children with 22q11.2 deletion syndrome (22q11.2). A few small studies have reported a 32-77% prevalence. Despite the prior studies examining hearing loss in patients with 22q11.2, there is an overall paucity of data regardingthe frequency, type, severity, and progression of hearing loss.

Methods: A retrospective chart review was completed. Data was combinedfor two large US-based 22q Centers based in tertiary care children's hospitals. Pediatric patients with a diagnosis of 22q11.2 deletion syndrome and documented audiologic testingwere included. Data collection included co-morbidities, results of all prior audiologic testing, radiologic temporal bone imaging, and otologic surgical procedures.

Results: 1,640 charts reviewed; 689 patients met inclusion criteria. Comorbidities included 87% speech delay and 25% cleft palate. In total, 2,539 audiograms were reviewed, of which 74% showed abnormalresults. For this cohort, 71% of patients had an abnormal audiogram. Hearing loss was most often mild and conductive; sensorineural hearingloss was less common, and a majoritydid not progress. Ear tube placement occurred in 42% of patients; of these, 55% went onto require multiple sets of ear tubes. Thirty-seven patients had temporal bone imagingwith 89% showing anomalies of the middle/inner ear.

Conclusion: This is the largest study to date describing hearing loss in children with 22q11.2. This study confirms a high frequency of hearing loss. The results highlight the importance of otolaryngology and audiology involvement in managing patients with 22q11.2, particularly as speech and language deficits may be exacerbated by these issues.

Early childhood development in 22q11.2 Deletion Syndrome

Authors: Samuel J.R.A. Chawner1,2, Amy L. Paine2, Matt J. Dunn3, Alice Walsh1, Poppy Sloane1, Megan Thomas1, Alexandra Evans1, Lucinda Hopkins-Jones1, Siske Struik4, IMAGINE-ID consortium, Jeremy Hall1, Jonathan T. Erichsen3, Susan R. Leekam2, Michael J. Owen1, Dale Hay2, Marianne B.M. van denBree1

Affiliations: 1Centre for Neuropsychiatric Genetics and Genomics,Division of Psychological Medicine and ClinicalNeurosciences, Cardiff University, Cardiff, UK
2Cardiff University Centre for Human Development Science, School of Psychology, Cardiff University, Cardiff, UK, 3School of Optometry, Cardiff University, Cardiff, UK
4Immunodeficiency Centre for Wales, University Hospital of Wales, Cardiff, UK

Abstract: Individuals with 22q11.2 Deletion Syndrome (22q11.2DS) are at increased risk of psychiatric conditions across the lifespan. Understanding development in early childhood may help identify early signs of psychiatric risk in 22q11.2DS.
Objective: To investigate the developmental profileof children aged 2 to 5 years with 22q11.2DS.

Methods: 32 children with 22q11.2DS and 12 siblingcontrols (siblings in the family who did not have 22q11.2DS) aged 2 -5 years old took part in the study, whichwas advertised through British Charity Max Appeal and via social media. Children underwent in-depth assessments across a range of areas of childhood development, including behavioural and emotional problems, social skills, language, motor skills, cognitive ability, and sleep problems. The impact of 22q11.2DS on development was estimated by comparisonwith control siblings.

Results: Children with 22q11.2DS experienced difficulties with behavioural and emotional problems,social skills, language, motor skills, cognitive ability and sleep problems comparedto control siblings. Developmental delays ranged between 8-16 months depending on the area of childhood development investigated. However, there was considerable variability, using cluster analysis we identified a subgroup of childrenwith 22q11.2DS (n=16) who showed more early signs of developmental and behavioural problems. Early motor and sleep problems were a marker of those childrenwho experienced more behavioural and emotional problems

Conclusion: Children with22q11.2DS show a range of developmental difficulties, though considerable differences exist between children. The presence of early developmental difficulties in 22q11.2DS indicates potential for early identification and intervention for behavioural and emotional problems in 22q11.2DS

The genetic puzzle: a mHealth application for individuals with 22q11 DS and their caregivers

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Background:Families with children with cognitive impairments due to CNVs experience communication challenges and lack of information, leading to tension. Bridging this communication gap is crucial,and mHealth applications hold promise.

Objective: The current study explored how adolescents with 22q11 DS and their families interact with mHealth applications to bridge the communication gap with the aim to inform the development of more impactfulinterventions. In this presentation, we will present the design and development of an mHealthapplication for individuals with 22q11 DS and their caregivers, along with key findings from its evaluation.

Methods: The application was designed and developed using an iterative, user-centered approach. Two phases, design, and evaluation were conducted. Feedback from medical and HCI expertswas gathered duringthe design phaseusing a low-fidelity prototype. The evaluation phase involved feedback from eight families through interviews, a trial period, and concluding interviews. Results were categorized through thematic analysis.

Results: The evaluation of the fully functional application revealed insights in four areas: overcoming usage barriers, stimulating conversation, providing information, and delivering added value. Families expressed gratitude and acknowledged the application's valuefor others.

Conclusions: The study confirms the need for user-friendly applications that bridge the communication gap betweenindividuals with cognitiveimpairments due to CNVs and their caregivers. Clinics and institutions are encouraged to develop similar applications. Considerations for developing applications for individuals with 22q11 DS and cognitive impairments are proposed, emphasising design principles, recognition in stimulating conversations, trusted sources for information, and personalized digital tools with continual benefits.

Neurodevelopmental trajectories in brain structure-function relationships in 22q11.2DS carriers

Authors: Silas Forrer1,2, Farnaz Delavari1,2, Corrado Sandini1, Dimitri Van De Ville2, Stephan Eliez1

Affiliations: 1 Developmental Imagingand Psychopathology Laboratory, University of Geneva School of Medicine, Geneva,Switzerland
2 Neuro-X Institute, École Polytechnique Fédérale de Lausanne, Geneva, Switzerland

Abstract: A recent network-neuroscience framework allows to quantify the relationship between functional andstructural connectivity in the brain,and demonstrate its variation across different regions, exhibiting local differences in accordance with regional specialization. Alterations in structure-function dependency have been associated with mental health problems in 22q11.2DS. However,at this stage it remains unclearwhether these alterations are innate or rather the outcome of a neuro-developmental process. We studied longitudinal structural-decoupling-index (SDI) maps in 22q11.2DS carriers, to assess their neurodevelopmental trajectory in comparison to typicallydeveloping healthy controls.

Methods: 139 healthy controls and 118 deletion carriers (in total438 scans, aged4.6 to 34) underwent longitudinally repeated magnetic resonance imaging. We combined brainactivity and structural connectivity measures to obtain regionalSDI maps. The SDI maps were then modeledover age-span using longitudinal multivariate analysis.

Results: We found a disturbed SDI trajectory in 22q11DS patients, observed predominantly in regions related to the central executive network (CEN), that are critically implicated in higher cognitive functioning. Our findings indicated that such alterations were already present during childhood development potentially representing early prognostic markers, as well as an atypical neurodevelopment of SDI during adolescence, which could relate to emerge of psychosis, thus represent a specific intervention target. These findings suggest that our neuroimaging technique to characterize structure-function dependency could represent a novel powerful biomarker to understand and predict vulnerability to mental health disturbances in 22q11DSSilas Forrer1,2, Farnaz Delavari1,2, Corrado Sandini1, Dimitri Van De Ville2, Stephan E In a next step, we will investigate whether such neurodevelopmental mechanisms could potentially be influenced by preventive pharmacological and non-pharmacological treatment strategies.

Service Evaluation of the 22q11.2DS Multidisciplinary Team Clinic at Great Ormond Street Hospital for Children

Authors: Dr Ruth Garcia-Rodriguez, Dr Lottie James, Isabella Mais

Abstract: 22q11.2DS is associated with high rates of ADHD,ASD, anxiety and psychosis. Timely intervention for these conditions is essential to mitigate their burden.

Great Ormond Street Hospital(GOSH) runs a monthly 22q11.2DSmultidisciplinary clinic (5-yearly reviews). Time-limited psychiatric assessment only enables screening for mental illness and psychoeducation. Symptomatic children requiring further assessmentare signposted to local services, which often feel ill-equipped to manage their complexity. There is no direct pathway for this clinic to refer internally to current GOSH neurodevelopment assessment services. This leads to delayed treatment.

Methods: We performed a service evaluation of the psychiatric component of our clinic to betterunderstand the rates of mental illness in our population, the sufficiency of localservices and whetheran internal referral pathway to expedite specialist care would be of value.30 patients, aged 4-17, were seen from June 2021-January 2022. Quantitative and qualitative data were gathered through medical record searches and telephone questionnaires. 15 children(50%) had a psychiatric diagnosis, with ASD being the most common. A further 9 (38%) children were on a local waiting list for assessment. Patients referred for local psychiatric assessment from our clinic had a more efficient and improved experience than those utilising the local referral system (via a GP). All patients experienced obstacles when trying to access local services: the assessment and diagnostic system was the most problematic.

Results: The results support the creation of a direct pathway into specialist psychiatric services to expedite care for these complex children.

Dysphagia in children with 22q11.2 Deletion Syndrome

Authors: Jana Ghulmiyyah, MD, Srivats Narayanan, Lauren Bartik, MS, CGC, Meghan Tracy, CCRC, Hung-Wen Yeh PhD,Jill Arganbright, MD

Background: Dysphagia is thoughtto occur commonly in children with22q11.2 deletion syndrome (22q11.2DS) with few prior small studies noting dysphagia in 36%-41% of patients.The purpose of this investigation is to more fully elucidate dysphagia in children with 22q11.2DS.

Methods: IRB approval was obtained, and a chart review was completed. Pediatric patients witha diagnosis of 22q11.2DS wereincluded. Data extracted included genetic diagnosis, co-morbidities, diagnosis of dysphagia, resultsof video fluoroscopic swallow studies (VFSS), andneed for gastrostomy tube (g-tube).

Results: A total of 166 charts were reviewed.Of these, 141 met inclusioncriteria. Co-morbidities included 60% with congenital heart disease and 15% with cleft palate. Of the cohort, 46% were evaluated by a feeding therapist for dysphagia, 28% underwent at least one VFSS, 19% required a hospital admission due to concerns around swallowing/feeding, and 23% required g-tube insertion. A total of 67 VFSS were reviewed, of which 64 (95%) reported dysphagia and/or airway penetrations. Recommendations of the VFSS results included 60% started on thickener, and 28% recommended no oral feeding. Thirteen patients had multiple VFSS, of which 61% showed no change or worsened results over time.

Conclusion: This data suggests dysphagia is common in children with 2q11.2DS and may not always improve over time. VFSS can be helpful in diagnosing dysphagia and determining safe alternative feeding methods. G-tube was required by nearly one-quarter of our patients. Screeningfor swallowing/aspiration concernsis important for children with 22q11.2DS to allow for accurate and timely diagnosis of dysphagia

Understanding and improving mental wellbeing support for **children and young people with 22q11.2 deletion syndro**me

Authors: Maria Gudbrandsen & Michelle Jaymana

Affiliation: Centre for Research in Psychological Wellbeing (CREW), Department of Psychology, University of Roehampton, London, SW15 5PJ, UK

Introduction: Individuals with 22q11.2 deletion syndrome (22q11.2DS) experience co-occurring physical and neurodevelopmental conditions and are at substantial risk of poor mental health (McDonald- McGinn et al., 2015; Schneider,2014). This exploratory study investigates stakeholder perspectives and experiences of existing mental wellbeing provision in the UK, with the aim to identify barriers and facilitators for tailoredmental wellbeing supportin the 22q11.2DS across both education and mental health.

Methods: A qualitative, participatory action research (PAR) study was conducted, involving collaboration among key stakeholders and prioritising CYP's authentic views. Participants included 10xCYP with 22q11.2DS, 14xparents, and 14xhealth professionals. Data collection methods comprise semi-structured interviews for CYP and parents and 3x focus groups for professionals. Question schedule was developed through consultation with YP with 22q11.2DS and their parent,as well as a consultant psychiatrist. A hybrid inductive-deductive Thematic Analysis was conducted on the integrated data.

Results: Still in preliminary stages,but common themes included aspectslike 1) a need for greater awareness of 22q11.2DS and the complex presentation; 2) lack of current support available across both education and mental health services, 3) difficulties around seeking support/know what is available; and 4) lack of support with transition and adults with 22q11.2DS.

Conclusion: These findings are essential for a better understanding of current barriers and facilitators to mental wellbeing in the UK and to create furtherawareness. These findingswill hopefully guide further studies, adapting current effective early interventions to the 22q11.2DS population, as well as more collaborative approach to treatmentacross professionals.

Lower extremity pain and exercise intolerance in children and adolescents with 22q11.2 deletion syndrome

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Affiliation: Dept. of Pediatrics, Wilhelmina Children's Hospital, University Medical Center Utrecht Utrecht, The Netherlands

Background: Throughout childhood and adolescence patients with 22q11.2 deletion syndrome (22q11DS) may experience exercise intolerance or lower extremity pain, with majorimpact on dailylife activities and on quality of life. Althoughthe occurrence and severity of these symptomsmay vary over individuals, an important proportion of patients above approximately 8 years appear to suffer from these problems. Given the variety of potential somatic manifestations associated with the syndrome, several medical factors could be involved. We planned to study:
(1) the prevalence of lower extremity pain and exercise intolerance in youth with 22q11DS and
(2) to what extent congenital heart defects (CHD), hypocalcemia and / or orthopedic problems are associated factors.

Methods: Data will be extractedfrom medical recordsin the national Dutch 22q11DSreference center in Utrecht. Approximately 150 children and adolescents from 8 through 18 years will be assessed. Patient and family reports on standardized routine care questions concerning the presence and severityof lower extremitypain and exerciseintolerance will be recorded, as well as information on the past medical history of CHD, the presence of hypocalcemia, and the presenceof distinct orthopedic problems (e.g., scoliosis, patella luxation, pes planes). We will carry out descriptive, bivariate, and multivariate analyses.

Results: We will present the results on the prevalence of lower extremity pain and exercise intolerance and the potentialassociations with CHD, hypocalcemia, and orthopedic problems.

Conclusion: We will discuss the magnitude and impact of lower extremitypain and / or exerciseintolerance in youth with 22q11DS and the potential clinical implications.

Organization of outpatient clinical care for patients with 22q11.2 deletion and duplication syndrome - differences and similarities across Europe

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Background: During growth and development children and adolescents with 22q11.2 deletion and duplication syndromemay display different medical, surgical, and psychosocial needs.During and after transition into adulthood new questions on (e.g.) occupational, relational, or residential issues, coping strategies and / or resilience may arise, next to medical challenges in adulthood. These needscan often (partially) be met by well-organized local, regional and / or national outpatient clinics. Differences in level of organisation, medical and paramedical specialty involvement, follow-up intervals and shared care solutions between distinct centers may have historical, pragmatic, or other origins, as well as distinct implications for patients. In this study we explore the design featuresof these outpatient CLINICS in different European centers.

Methods: Within the recently started ERN CRANIO working group on 22q11, we will carry out a short survey on design features of regional and national reference 22q11 centers within the European Union.

Results: We will present results on the level of organisation and specific design features over the included 22q11 centers.Relevant similarities and differences will be exemplified and put into international perspective.

Conclusion: We will discuss differences and similarities in organisational design of distinct European regional and national outpatient reference centers for patients with 22q11.2 deletion and duplication syndrome. Implications and recommendations for future (complex) patient care will be discussed.

A multi-method approach for the identification of social functioning profiles in adolescents and young adults with 22q11DS

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Affiliations: (a) Clinical Psychology Unit for Intellectual and Developmental Disabilities, Psychology and Sciences of Education faculty, University of Geneva, Geneva, Switzerland, (b) Department of Genetic Medicine and Development, Faculty of Medicine, University of Geneva, Geneva,Switzerland, (c) Developmental Imaging and Psychopathology Lab Research Unit, Faculty of Medicine, University of Geneva, Geneva, Switzerland.

Abstract: The 22q11 deletion syndrome (22q11DS) exhibits a wide range of symptoms, including psychiatric manifestations that become more common with age. The complexity of these symptoms often leads to social impairments. Becausestatic measures do not providean integral view of social functioning, which is highly context dependent, this study aims to use a combination of measures: the Ecological Momentary Assessment (EMA), parent-reported questionnaires, and direct observation to obtain a comprehensive understanding of social functioning in individuals with 22q11DS.

This study aims to characterize the various socialfunctioning profiles withinthe 22q11DS population. This characterization would enable more targeted therapeutic interventions based on specific social functioning phenotypes and their associated comorbidities. Furthermore, this methodology could provide insights into the impaired and preserved aspects of social functioning in this population across different phenotypes.

Methods: 70 adolescents and young adults with 22q11DS and an equal number of typically developing individuals (TD) have been alreadycollected. Participants completed the EMA protocol, which measuresthe frequency and subjective evaluation of social interactions several times a day during a week in the daily-life environment. Additionally, parent-report questionnaires (SRS and ERSSQ) assessed social skills, while direct observation (SSPA) during role plays quantified social appropriateness. The influence of mental health difficulties on social functioning will also be explored.

We expect heterogeneous social functioning impairments among participants with 22q11DS, while TD will exhibit a more homogeneous profile of good social functioning. We hypothesize that participants with 22q11DS will be clustered at least two subgroups, resulting in different social functioning profiles. Additionally, we expect to observe different associations with mental health difficulties in each subgroup.

Evaluation of Parents Plus parenting programme delivered to parents of children with 22q11.2DS

Authors: Ahmed Khan1, Veselina Gadancheva2, Wesley Mulcahy3, SuzanneKelleher3, Fiona McNicholas1,2

Affiliations: 1 UCD School of Medicine, Dublin, 2 Department of Psychiatry, Children's Health Ireland at Crumlin, 3 Department of General Paediatrics, Children's Health Ireland at Crumlin

Abstract: The aim of this study was to evaluate the effectiveness of Parents Plus Children's Programme (PPCP)as a parenting intervention for parents of children with 22q11.2DS who experience behavioural difficulties. PPCP is an evidence-based parenting programmedeveloped in Ireland and delivered widely in the community.

Methods: A group of eight parents of children aged 6-12 years with 22q11.2DS were invited to participate in PPCP that included 8 sessions delivered online. Five parents completed the programme. Questionnaires (Strengths and Difficulties Questionnaire SDQ, Parental Stress Scale PSS, Kansas Parental Satisfaction Scale KPSS) were administered at three different time points: prior to(T1), immediately after(T2), and at six-month follow-up(T3). Parents were asked to identifyindividual and child-related goals. SPSS vs27 was used for statistical analysis to determine significance.

Results: Mean baseline SDQ score was 18.2 (SD 5.07), PSS – 38.2 (SD 7.36), KPSS – 13.8 (SD 3.27). No significant difference at baseline T1 and post-intervention T2 SDQ, PSS and KPSS scores was found. Parents moved significantly towards achieving their individual and child-oriented goals. 6-monthly follow-up data collection is currently in progress. Parents found value in sharing theirexperiences with otherparents of children with 22q11.2DS and beingpart of a support group.

Conclusion: This pilot study demonstrated potential feasibility and benefits for parents attending the group. The small group sample is a limitation of this study and ongoing evaluation is planned.

Prospective Natural History Study of Idiopathic-like Scoliosis in Patients with 22q11.2 Deletion Syndrome, Starting Before its Pathological Onset

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Affiliations: (1) Department of Orthopedic Surgery, University Medical CenterUtrecht, Utrecht, The Netherlands, (2) Department of Biomedical Engineering, Eindhoven University of Technology, Eindhoven, The Netherlands.

Abstract: 50% of 22q11.2DS-patients develops an idiopathic-like scoliosis. We screen 22q11.2DS-patients biennially with radiographs for scoliosis from age 6-18. Currently, important information about scoliosis development in 22q11.2DS is unknown. This study will use our longitudinal database to inventory the natural history of scoliosis in 22q11.2DSand to explore whether this can be used for predictive modelling.

Methods: From the prospective registry, which includesradiographs before scoliosis onset,773 full-spine radiographs of 296 patients were examined. Cobb-angles were measured,with scoliosis development defined as a curve >10°. Age of onset, risk of progression to >30° and need for treatment were evaluated.

Results: 119(40%) of patients developed scoliosis >10°, 62(44%) of included girls and 57(37%) of included boys, 14(5%) progressed to >30°. Eight (3%) required surgical treatment (see figure).A total of 68 patients had radiographic follow-up until 16 years or older: 40(59%) had scoliosis with a Cobb angle>10°, 11(16%) >30° and 6(9%) required surgery. From all scoliosis patients, 31 had radiographs taken before the onset of scoliosis. In this group, the mean age of progression into a scoliosis (>10°) was 11.3±2.6 years and ranged from 5.6 – 15.4 years.

Conclusion: This prospective natural history study describes scoliosis development, starting before its onset. It demonstrates the age of onset and differentiates between patients without scoliosis, mild scoliosis, and severe progressive deformities. This provides the opportunity for future risk-profiling to distinguish between mild, stable and progressive scoliosis, which possibly could be extendedto the scoliosis population as a whole.

Using non-invasive brain stimulation to enhance working memory skills in youths with 22q11.2 deletion syndrome

Authors: Caren Latrèche1, † ,*, Valentina Mancini1, † , Vincent Rochas2, Johanna Maeder1, Lucia M. Cantonas3, Maude Schneider4, Christoph M. Michel2, Stephan Eliez1,5

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5 University of Geneva, School of Medicine, Switzerland

Abstract: Individuals with 22q11.2 deletion syndrome (22q11.2DS) often display cognitive difficulties in several domainsincluding memory. Yet,such cognitive deficits only show limited responseto current pharmacological treatment. Non-invasive brain stimulation (NIBS) is a safe intervention which applies low-intensity electrical currents to the scalp,with the aim to modulatebrain functions and enhance cognition. NIBS could thus represent a promising strategy to alleviate memory deficits in individuals with 22q11.2DS.

Method: We used transcranial current stimulation to improve visual working memory (WM) in 34 deletion carriers. We targeted the prefrontal and temporal cortices which underlie WM. First, we collected EEG and MRI data to personalize the frequency and the intensity of the stimulation to each participant (Day 1). Participants were randomized to either sham or real stimulation (Days 2 and 3) and then completed a WM task and a control task. After the stimulation, we assessed tolerability to stimulation and administered verbal and visuospatial WM tasks.

Results: Side effects were rare and of mild intensity. After the real stimulation, we found a significantly increased accuracy in the WM task but not in the control task, whereas no change was found after the sham stimulation. Moreover, we found improved verbal WM performance at least one-hour post-stimulation.

Conclusions: This is the first NIBS study in 22q11.2DS. We showed that the use of NIBS is safe and can effectively modify cognitive functions. Our findings support the application of repeated sessions of brain stimulation in 22q11.2DS.

Review of the mental health profile of 22q11.2DS children referred for a psychiatry assessment at Children's Health Ireland at Crumlin

Authors: Rae Lyz Yee1, Veselina Gadancheva2, Fiona McNicholas1,2

Affiliations: 1 UCD School of Medicine, Dublin,2 Department of Psychiatry, Children's Health Irelandat Crumlin (CHI at Crumlin)

Background: 22q11.2 Deletion Syndrome (22q11.2DS) is among the commonest microdeletion syndromes. In Ireland this equates to approximately 15 to 30 babies with 22q11.2DS being born per year. The syndrome caries high risk for neurodevelopmental and psychiatric comorbidities. The aim of this report is to summarise the profile of children with 22q11.2DS referred for a psychiatric assessment at CHI at Crumlin.

Methods: Descriptive report of the findings following a psychiatric review offered to children with 22q11.2DS between 2015-2023. In several cases,Child Behavioural Checklist 6-18y (CBCL) was completed by parents and data was extracted.

Results: A total of 95 children, age 3-18y, were referred to child psychiatry as part of the integrated coordinated 22q Clinic. 74 children were assessed while21 families did not attend or declined an assessment. 60 (81%) young people met the diagnostic criteria for at least one psychiatric condition. 34 (57%) presented with anxiety, depressive episode was diagnosed in 2 cases (3%). 18 (30%) childrenpresented with ADHD symptoms. 1 case of psychosis was detected.1 child was given a diagnosis of oppositional defiant disorder and 4 (7%) presented with autism. 14 (19%) cases did not meet the threshold for a diagnosis and psychoeducation was recommended. CBCL questionnaire scores were available for 36 children. The total difficulties score was in the clinical range in 19 (53%) cases and borderline – in 5 (14%). In 12 (33%) children the score was within the normal range.

Conclusion: Our data confirmthe importance of early psychiatric screening in the 22q population and timely treatmentto improve future outcomes.

Inversion and AT-rich repeat size polymorphisms can drive 22q11.2 rearrangements

Authors: Senne Meynants, Marta Sousa Santos, Lisanne Vervoort, Nicolas Dierckxsens, Erika Souche, and Joris R. Vermeesch
Department of Human Genetics, KU Leuven, Leuven, Belgium

Introduction: The 22q11.2 Deletion Syndrome (22q11.2DS) is the most common microdeletion disorder, with an estimated incidence of 1 in 2148 live births, being caused by rearrangements between the low copy repeats (LCRs) present in this locus. Due to the repetitive nature of the LCRs, along with their structural hypervariability in both copy number and organization, short-read sequencing has been insufficient to characterize them and to map the exact deletion breakpoints.

Methods: To gain insightsinto the mechanisms that drive rearrangements in patient- parent duos, we used a combination of Oxford Nanopore Technologies sequencing and fiber-FISH, a targetedoptical mapping technique.

Results: We successfully mapped the rearrangement site at the subunit level in two patients and further refined the breakpoint at the nucleotide level in three. This revealed that the rearrangements occur within multipleloci, which may contribute to the high incidence of the 22q11.2DS. Interestingly, in one family, the parent was seen to carry an inversionpolymorphism that seemed to be predisposing for the subsequent deleterious event happening in the proband.Furthermore, we observedcopy number variation of AT-rich-HSATI-AluY triplets resultingin fragment lengthsranging between 2,8 and 25 kb, which might as well influence the frequency of both deletions and translocations in the 22q11.2locus.

Conclusion: Taken together, our results support that both inversions and AT-rich repeat size polymorphisms can be drivers for 22q11.2 rearrangements.

Meta-analysis of prevalences of cardiovascular, psychiatric and palatal/dental symptoms in 22q11.2 deletion syndrome

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Background: The 22q11.2 deletion syndrome (22q11.2DS) is caused by a heterozygous deletion of chromosomal locus 22q11.21 and associated with multisystem symptoms, such as cardiovascular, psychiatric and palatal/dental. Little is known about the prevalence numbers in these symptoms.

Methods: Four separate systematic literature searches were conducted in autumn 2022 using the databases Pubmed, Web of Science and Cochrane library to retrieve studies in English and German. Those not focusing on 22q11.2DS or dealing with a combination of several genetic defects were excluded. Risk of bias was assessed, and Grading of Recommendations Assessment, Development and Evaluation (GRADE approach) was applied as additional quality assessment for the outcomes.

Results: The systematic searches identified 8476 studies, of which 60 were included (6 cardiovascular, 41 psychiatric, 13 palatal/dental focus). For studies with cardiovascular focus there was enough homogeneity between studies to perform a meta-analysis of prevalence: Tetralogy of Fallot (20 %), isolated ventricular septal defect (14%), pulmonary atresia with ventricular septal defect (10 %), interrupted aortic arch with ventricular septal defect (10 %) and truncus arteriosus (9 %). Psychiatric and palatal/dental symptoms were found to be prevalent in all studies analysed. However, there was high variability between studies regarding prevalence figures. The GRADE approach resulted in a very low quality of evidence for all examined symptoms.

Conclusion: While we can give first figures on prevalence of the studied symptoms, our analysis of the literature also shows that more high-quality studies with unified standards are needed to predict prevalence's with more certainty.

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Clinical profile of paediatric patients with 22q11.2 Deletion Syndrome attending CHI at Crumlin - retrospective chart review

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Background: 22q11.2 deletion syndrome (22q11.2DS) is the most common microdeletion syndrome with a prevalence of 1 in 2000 to 1 in 4000 live births. The clinical presentation is diverse affecting multiple systems and organs. Developmental delay, neurodevelopmental disorders, behavioural and psychiatric difficulties are common. In their journey patients navigate throughmultiple clinical specialties.

Abstract: The aim of this studywas to describe the healthcare needs and service utilisation of patients with 22q11.2DS attending a specialist paediatric 22q clinic.

Methods: Data was extracted following retrospective review of clinical files of patients who attended the clinicbetween October 2017 and May 2023. Additional search was done through the hospital Integrated Patient Management System. Data was collated on Microsoft Excel. SPSS vs 27 was used for descriptive statistical analysis.

Results: 175 patient records were reviewed. 15% of the patients were 0-5 years of age, 30% - 6-11 years, and 55% - over 12years. 95% of the patients had 22q11.2 deletion, 4% - 22q11.2 duplication and 1% - complex genetic rearrangement in the critical chromosomal region. 14.3% of the cases were familial. More than 50% of were seen by at least one other clinics in the hospital. 36.0% of the patients were active on the cardiology register, followed by dentistry (26.9%), audiology (21.7%), plastic surgery (19.4%), ophthalmology (18.9%), orthopedics(17.1%), immunology (14.3%) and other specialties.

Conclusion: A wide distribution of age groups attended the clinic. Cardiology is the top complex specialty link, but multidisciplinary involvement in 22q patient care is fundamental. Integrated care ensures patient centered holistic approach.

Speech outcomes after Pharyngoplasty in 22q11.2 Deletion Syndrome: Cranial Based Pharyngeal Flap and the Modified Honig procedure: Cranial Based Pharyngeal Flap and the Modified Honig procedure

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Introduction: Historically, the Modified Honig Procedure (MHP) was performed for velopharyngeal insufficiency (VPI) in Utrecht. Literature seemed to indicate better results for CranialBased Pharyngeal Flap(CBPF). Gradually switchto the CBPFin more severe cases with poor levatorfunction.

Method: Retrospective chart review.Children with 22q11DSand VPI and CBPF or MHP procedure between 2006-2021. Primary outcome measure: Improvement in speech understandability ≥12 months post-operatively. Secondary outcome measures: post-operative resonance, post-operative nasal emission, complications.

Results: Total 68 cases, male 35, female 33, average age operation 6 years (range 3-17). Previous palatal surgeryin 18/68 cases, 10 in CBPF group and 8 in MHP group.Pre-op levator function more limited in the CBPF group (assessed by nasendoscopy / video- fluoroscopy). Resonance improved in 82% in the CBPF and 83% in the Honig group. Some degree of remaining air-leakage was noted in 53% in the CBPF and 39% in the Honig group. According to parents' speechimproved in 78% after CBFP and 96% after MHP.

Discussion: Overall, no statistically significant differences in speech outcomes and complications between techniques. Limitations: retrospective study,small population size, non-homogenous groups. VPI in 22q11.2 is multifactorial, speech enhancing surgery is worthwhile. An individualapproach (tailored care) is advocated.

Language profiles of school-aged children with 22q11.2 copy number variants

Authors: Verbesselt, J. (1st corresponding author, presenter); Solot, C.B.; Van Den Heuvel, E.; Crowley, T.B.; Giunta, V.; Breckpot, J.; McDonald-McGinn, D.M.; Zink, I.; Swillen, A.

Background: Although it is known that copy number variants (CNVs) on chromosome 22, such as 22q11.2 deletion (22q11.2DS) and 22q11.2 duplication (22q11.2Dup) syndromes, are associated with higher risk for neurodevelopmental issues, few studies have examined the language skills across 22q11.2Dup nor compared them with the 22q11.2DS.

Methods: The current study aims to characterise language abilities in school-aged children aged 6-16 years with 22q11.2Dup (n = 29), compared to age-matched children with 22q11.2DS (n = 29). Standardised language tests were administered, assessing receptive and expressive language skills across different language domains.

Results: Results indicate that children with 22q11.2Dup demonstrate significantly more language problems compared to the general population. Mean language skills were not significantly different among children with 22q11.2 CNVs in this cohort. While children with 22q11.2DS demonstrated language difficulties starting at the word level, the most common language problems in children with 22q11.2Dup started at the sentence level. Importantly, both expressive and receptive language as well as lexico-semantic and morphosyntactic domains were impaired in children with 22q11.2 CNVs.

Conclusion: Early identification, therapeutic intervention, and follow-up of language impairments in children with 22q11.2Dup are recommended to support language development and to reduce longitudinal impact of language and communicative deficits.

UP NEXT.....

Belgium 2025

Welcome to Brussels for the 5th European conference, 10 years after the first one that also took place in Brussels. The start of the 22q11 Europe adventure!

Conference location: Auditoires Saint Luc (UCL)

Dates: 22nd &23rd November, 2025

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