The 8th Biennial International 22q11.2DS Conference

Magic of Making a Difference.

The International 22q11.2 Foundation, Inc.
www.22q.org

Disney's Yacht and Beach Club Resorts
LAKE BUENA VISTA, FLORIDA, USA
JULY 6-10, 2012
# Conference at a Glance

**7:00 AM - 5:00 PM REGISTRATION OPEN DAILY — Asbury Rotunda**

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<td>9:00 AM—3:30 PM</td>
<td>8:00 AM—5:30 PM</td>
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<td>Family Program</td>
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<td>Professional Pre-Conference Symposium</td>
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<td>Sessions IX—XV</td>
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<td>International Brain and Behavior Consortium</td>
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<td>Grand Harbor Ballroom South</td>
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<td>6:30 PM—8:30 PM</td>
<td>12:30 PM—1:45 PM</td>
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<td>Speakers Reception with Group Photo Atlantic Dance Hall</td>
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<td>Lunch 22q Style Salons 5—8</td>
<td>12:45 PM—4:30 PM</td>
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<td>Opening Reception for All Asbury Rotunda</td>
<td>10:00 AM—11:30 AM</td>
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<td>Lunch International Style</td>
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<td>Combined Family &amp; Professional Program</td>
<td>Session A</td>
<td>Older Children, Adult and Adolescent Workshops</td>
<td>10:00 AM—11:30 AM</td>
<td>1:30 AM—6:00 PM</td>
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<td>Grand Harbor Ballroom South</td>
<td>Children’s Programs and Adult and Adolescent Workshops</td>
<td>* Capture the Moment</td>
<td>Simultaneous Family Program</td>
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<td>* Making Memories</td>
<td>* Working Things Through Together</td>
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<td>Sessions 12—14</td>
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<td>Welcome</td>
<td>* Getting to Know You</td>
<td>1:30 PM—3:00 PM</td>
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<td>Grand Harbor Ballroom South</td>
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<td>Parade of Nations</td>
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<td>Session D</td>
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<td>Angelo DiGeorge Memorial Medal</td>
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<td>Adult and Adolescent Workshops</td>
<td>Older Children, Adolescents and Adults with 22q11 deletion Syndrome Workshop</td>
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<td>5:30 PM—6:30 PM</td>
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<td>* Riding Things Through with 22q</td>
<td>* Put on a Happy Face</td>
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<td>Session 1</td>
<td>Simultaneous Family Programs</td>
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<td>* Challenges with Solutions</td>
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<td>Adult and Adolescent Workshops</td>
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<td>Session E</td>
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<td>Session 2</td>
<td>* One for All and All for Fun</td>
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<td>Older Children, Adolescents and Adults with 22q11 deletion Syndrome Workshop</td>
<td>22q Barbeque Character Dinner (ticket required) Marina/Boardwalk</td>
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<td>Grand Harbor Ballroom South</td>
<td>* Now What?</td>
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<td>* Put on a Happy Face</td>
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<td>* Mind Over Matter</td>
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# Combined Family & Professionals Program

**Friday, July 6th**

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<th>Time</th>
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| 8:00 AM | **REGISTRATION OPENS** - Disney’s Yacht and Beach Club Resorts,  
**ASBURY ROTUNDA AND LOBBY** |
| 4:00 PM | **WELCOME RECEPTION** – Light Refreshments,  
Disney’s Yacht and Beach Club Resorts,  
**ASBURY ROTUNDA AND LOBBY** |

**“22q and TuTu” Silent Auction and Shoppe for Everything 22q – Opens, ASBURY ROTUNDA AND LOBBY**

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| 5:00 PM | **OPENING REMARKS** – Disney’s Yacht and Beach Club Resorts,  
**GRAND HARBOR BALLROOM SOUTH**  
Anne Bassett, Carol Cavana,  
Donna McDonald-McGinn, Wendy Rose  
Welcome and Opening Remarks – Program Chair and Organizing Committee  
Parade of Nations  
Setting the Programmatic Stage |
| 5:15 PM | **ANGELO DIGEORGE MEMORIAL AWARD PRESENTATION** – Peter Scambler and Anne Bassett         |

**Session 1: Pulling Together for the Long Haul**  
Disney’s Yacht and Beach Club Resorts,  
**GRAND HARBOR BALLROOM SOUTH**  
Moderators: Carol Cavana and Wendy Rose  
5:30 to 6:30 PM

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| 5:30 PM | Carol Cavana  
*Making Newborn Screening a Reality – Why Families Care* |
| 5:35 PM | Wendy Rose  
*Detection – Care – Cure: A Common Goal* |
| 5:40 PM | Jill Stein  
*Supporting Regional Clinical Centers of Excellence – Why and How* |

**Session 2: Newborn Screening, Prevalence and Why It Matters**  
Disney’s Yacht and Beach Club Resorts,  
**GRAND HARBOR BALLROOM SOUTH**  
Moderators: Anne Bassett and Peter Scambler  
6:30 to 8:00 PM

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<th>Time</th>
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| 5:45 PM | Michelle Breedlove-Sells  
*Benefits of Summer Camp - Priceless!* |
| 5:50 PM | Maria Kamper  
*It’s a Small World - International Collaborations* |
| 5:55 PM | Sheila Kambin  
*The Miracle of Social Media - “22q at the Zoo” Worldwide Awareness Day* |
| 6:00 PM | Brenda Finucane  
*The Same Name Campaign – Practice Makes Perfect* |
| 6:05 PM | Karen Ruckman-Lindsay  
*Living with 22q - One Journey Instrumental Performance* |
| 6:20 PM | **DISCUSSION**                                                                            |
| 6:30 PM | Donna McDonald-McGinn  
*Where Have We Been? Where Are We Going? How Can Newborn Screening Help Us Along This Journey?* |
| 6:40 PM | Anne Bassett  
*Where Have We Been? Practical Guidelines for Managing Patients – A Document for Change* |
| 6:45 PM | Michael E. Mitchell  
*Impact of Early Detection of 22q11.2DS Related Congenital Heart Disease: Implications for Newborn Screening* |
| 6:50 PM | Julie Wootton  
*Remembering Max: In Hopes of Making a Difference Beyond the United Kingdom* |
| 6:55 PM | Sheila Kambin  
*The Odd Man Out! When the Patient Does Not Fit the Profile – One Family’s Diagnostic Odyssey* |
Saturday, July 7th

FAMILY PROGRAM

9:00 AM  REGISTRATION OPEN —
Disney’s Yacht and Beach Club Resorts, ASBURY ROTUNDA AND LOBBY

Session A: Simultaneous Programs 10:00 AM – 11:30 AM

CHILDREN’S PROGRAM:

Making Memories!
Disney’s Yacht and Beach Club Resorts, ASBURY HALL D

“22q and Sassy Doo”! (Children - Ages 3 years and older)

“I’m a Super Hero”! (Children – Ages 3 years and older)

Tweens and Teens – Video Gaming! (Children – Ages 8 years and older)

SIMULTANEOUS ADOLESCENT AND ADULT WORKSHOP OPTIONS:

Getting to Know You! —
Disney’s Yacht and Beach Club Resorts

Choose from one of the following simultaneous sessions:

A.1 Sibshop – A Program for Adolescent Siblings -
Barb Haas-Givler, Brenda Finucane, Alison Blackmon and
Justin Weinberg, ASBURY HALL A

A.2 Breaking the Ice – 22q Bingo!
(Adolescents and Adults with 22q11.2DS) – Wendy Rose, Michelle
Breedlove-Sells, Ayne Lawlor, Missy Disibio, Kayla Disibio,
Lauren Dodge, Joseph Lukac, and Meg Maguire, ASBURY HALL B

A.3 Mom’s Make a Difference So Focus On You! —
Zumba Dancing With International Meet and Greet!
Swati Patel, Carol Cavana, Julie Wootton, Maria Kamper, Anne Lawlor,
Mindy Dodge, Danielle Krijger and Barbi Weinberg,
THE ADMIRAL YACHT POOL

A.4 Dad’s Morning Out – Taking Time to Unwind (Or Wind Up!)
- Joe Uccello, Marc Weinberg, Louis Cavana and Dennis Green,
BEACH VOLLEYBALL COURT

A.5 Grandparents Need Attention Too!
— Alice Bailey and Louise Amlie-Wolf, SALON 8

(Family Program continued)
Saturday, July 7th
(Family Program continued)

Session B: Simultaneous Programs
1:30 PM – 3:00 PM

WORKSHOP OPTIONS:
Choose from one of the following simultaneous sessions:

One For All and All For Fun!
Disney’s Yacht and Beach Club Resorts
B.1 Scavenger Hunt (Adolescents and Adults) — Alice Bailey, Swati Patel, Missy Disibio, Kayla Disibio, Lauren Dodge, Joseph Lukac, and Ayne Lawlor, ASBURY ROTUNDA

Now What? —
Disney’s Yacht and Beach Club Resorts
B.2 Customize the Learning Environment: Effective Strategies to Support Behavior and Learning
(Parents, Grandparents and Caregivers) — Barb Haas-Givler and Meg Maguire, ASBURY HALL A

Dealing with Behavioral Differences During Adolescence and Adulthood
(Parents, Grandparents and Caregivers) — Raquel Gur, Daneen Whinna and Margaret Souders, ASBURY HALL B

Legal Zoom – How to Navigate Special Needs Trusts, Insurance Claims and Educational Appeals
(Parents, Grandparents and Caregivers) - Marc Weinberg, Michael Rama and Marilyn Cohen, SALON 8

Sunday, July 8th
FAMILY PROGRAM

9:00 AM REGISTRATION OPEN -
Disney’s Yacht and Beach Club Resorts, ASBURY ROTUNDA AND LOBBY

Session C: Simultaneous Programs
10:00 AM – 11:30 AM

WORKSHOP OPTIONS:
Choose from one of the following simultaneous sessions:

Capture the Moment -
Disney’s Yacht and Beach Club Resorts
C.1 Photography for Beginners
(Older Children, Adolescents and Adults)
[Please bring a camera] — Debbie Reynolds, Michelle Breedlove-Sells, Ivy Reynolds, Nadia Zomorodian and Alice Bailey, ASBURY HALL D

Working Things Through - Together -
Disney’s Yacht and Beach Club Resorts
C.2 Go to Sleep! Hands-On Demonstration to Help Fall and Stay Asleep
(Adults, Parents, Grandparents and Caregivers — Children are welcome and encouraged to attend with their Parent/Caregiver) - Margaret Souders, Daneen Whinna and Swati Patel, ASBURY HALL C

C.3 Puberty and Emerging Sexuality
(Parents, Grandparents and Caregivers) — Natalie Blagowidow, Sheila Kambin and Louise Amlie-Wolf, ASBURY HALL A

C.4 Sharing the Diagnosis Across the Lifespan
(Parents, Grandparents and Caregivers) — Betsy Leech, Brenda Finucane and Kelly Schoch, ASBURY HALL B

Session D: Simultaneous Programs
1:30 PM – 3:00 PM

WORKSHOP OPTIONS:
Choose from one of the following simultaneous sessions:

Riding Things Through with 22q -
Disney’s Yacht and Beach Club Resorts
D.1 Speak Out – A Program for Adolescents with 22q11.2DS —
Brenda Finucane, Daneen Whinna and Joseph Lukac, ASBURY HALL C

D.2 The Birds, the Bees and Internet Safety
(Adolescents and Adults with 22q11.2DS) —
Natalie Blagowidow, Sheila Kambin, Missy Disibio, Ayne Lawlor and Kayla Disibio, ASBURY HALL D

Challenges With Solutions -
Disney’s Yacht and Beach Club Resorts
D.3 Feeding, Swallowing and Speech
(Parents, Grandparents and Caregivers) —
Peggy Eicher, Maria Mascarenhas, Cindy Solot and Marilyn Cohen, ASBURY HALL A

D.4 Navigating IEP’s and Creating Positive School Partnerships
(Parents, Grandparents and Caregivers) —
Ed Moss, Donna Cutler-Landsman and Barb Haas-Givler, ASBURY HALL B
Sunday, July 8th
(Family Program continued)

7:30 PM  “22Q BARBEQUE” - CHARACTER DINNER FOR ALL!
Disney’s Yacht and Beach Club, MARINA/BOARDDWALK

Monday, July 9th
FAMILY PROGRAM

7:00 AM  REGISTRATION –
Disney’s Yacht and Beach Club Resorts,
ASBURY ROTUNDA AND LOBBY

Session 3: 22q – What’s it to You?
GRAND HARBOR BALLROOM SOUTH

8:00 AM  GOOD MORNING FLORIDA!
DAILY REMARKS –
Anne Bassett, Carol Cavanaugh,
Donna McDonald-McGinn & Wendy Rose

8:10 AM  Carol Cavanaugh, Sheila Kambin & Wendy Rose
“It Takes A Village”
Honoring Heroes Amongst Us
Inaugural “Unsung Hero Award”

8:20 AM  Rick Guidotti, Positive Exposure
“Through the Looking Glass” – Recognizing Value in Difference

8:50 AM  DISCUSSION

9:00 AM  Donna McDonald-McGinn and Anne Bassett
Nuts and Bolts – What Are the 22q11.2
Deletion and Duplication Syndromes?

9:45 AM  DISCUSSION

10:00 AM  COFFEE BREAK – ASBURY LOBBY

Session E: Simultaneous Program
10:00 AM – 11:30 AM

WORKSHOP OPTION:
Put On a Happy Face -
Disney’s Yacht and Beach Club Resorts

E.1  Supporting Self Esteem and Sharing
Your Life Perspective With the World
(Older Children, Adolescents and Adults) -
Rick Guidotti, Brenda Finucane and
Alice Bailey, ASBURY HALL A

Session 4: Of Mice and Men –
What Can Basic Science Tell Us?
GRAND HARBOR BALLROOM SOUTH
Moderators: Peter Scambler & Bernice Morrow

10:30 AM  Peter Scambler
Understanding the Developing Head and Neck

10:45 AM  Bernice Morrow
The Heart and Why it is Affected

11:00 AM  Anthony Lamantia
Behavior in Mice as a Clue to Behavior in Humans

11:15 AM  DISCUSSION

Session 5 – Drug Discovery and Treatment Outlook
GRAND HARBOR BALLROOM SOUTH
Moderators: Anthony Lamantia and Christian Marshall

11:30 AM  Christian Marshall
What Can Studies in Autism tell us about 22q11.2DS?

11:45 AM  Paul Wang
Lessons from Fragile X and Down Syndrome Too!

12:15 PM  DISCUSSION

12:30 PM  LUNCH 22Q STYLE – CHECK TABLE MARKERS FOR SEATING!
Disney’s Yacht and Beach Club Resorts, SALONS 5-8

1:15 PM  2012 FAMILY GROUP PHOTO
GRAND HARBOR BALLROOM SOUTH

Session F: Simultaneous Program
1:30 PM – 3:00 PM

WORKSHOP OPTION:
Mind Over Matter –
Disney’s Yacht and Beach Club Resorts

F.1  Care for the Caregiver: Use of
Mindfulness Based Stress Reduction for Parents of Adolescents with
Autistic Spectrum Disorders
(Parents, Grandparents and Caregivers)
- Chris Moriconi, Margaret Souders, and
Alice Bailey – ASBURY HALL A

(Family Program continued)
Monday, July 9th
(Family Program continued)

Session 6: Healthcare Guidelines -
There's an App for That I
GRAND HARBOR BALLROOM SOUTH
Moderators: Elizabeth Goldmuntz and Kathleen Sullivan

1:30 PM Elizabeth Goldmuntz
The Heart
1:40 PM Kathleen Sullivan
Infection and Related Problems
1:55 PM Maria Mascarenhas
Digestion
2:05 PM Peggy Eicher
Feeding
2:15 PM Solveig Oskarsdottir
Ear, Nose and Throat
2:25 PM Brian Forbes
The Eye as a Window to the Brain
2:35 PM DISCUSSION

Session 7: Healthcare Guidelines -
There's an App for That II
GRAND HARBOR BALLROOM SOUTH
Moderators: Nancy Unanue and Alex Habel

2:50 PM Nancy Unanue
Thyroid Disease and Growth Issues
3:00 PM Alex Habel
Growth Charts
3:10 PM Lorraine Katz
Calcium Problems
3:20 PM Michele Lambert
Bleeding
3:30 PM Rosario Mayro
Teeth
3:40 PM Dino Colo
Bones
3:50 PM Annique Hogan
Keeping It All Together By Establishing a Medical Home
4:00 PM DISCUSSION
4:20 PM AFTERNOON TEA – ASBURY LOBBY

Session 8: 22q It Starts With You -
an International Perspective
GRAND HARBOR BALLROOM SOUTH
Moderators: Julie Wootton and Maria Kamper

4:40 PM Julie Wootton
Max Appeal's Activity Programmes for Young People in the United Kingdom
4:50 PM Maria Kamper
Establishing Good Working Partnerships with Teachers in Australia
5:00 PM Anne Lawlor
Need for Informational Leaflets in Ireland
5:10 PM Daniella Krijger
What Difference? A View From the Netherlands
5:20 PM Mindy Dodge
Rolling Up Our Sleeves For Good in the USA
5:30 PM Fred Weiner
The Value in Establishing 22q11.2DS Specific Summer Camps – Lessons from Dragonfly Forest
5:40 PM DISCUSSION
6:00 PM ADJOURN – DINNER ON YOUR OWN
9:00 PM MOVIE ON THE BEACH
FOR HOTEL GUESTS,
Disney's Yacht and Beach Club Resorts

Tuesday, July 10th
FAMILY PROGRAM

7:00 AM REGISTRATION –
Disney's Yacht and Beach Club Resorts, Asbury Rotunda and Lobby

Session 9: What a Difference a Palate Makes!
GRAND HARBOR BALLROOM SOUTH
Moderators: Christina Persson and Hillary Joyce

8:00 AM Christina Persson
Speech
8:15 AM Oksana Jackson
Palate
8:30 AM Margaret Souders
Postoperative Apnea – Caution: Sleep Impacts Everything!
8:40 AM Meg Maguire
Elective Surgery – Points to Ponder
8:50 AM DISCUSSION
Tuesday, July 10th
(Family Program continued)

Session 10: Early Childhood Development
GRAND HARBOR BALLROOM SOUTH
Moderators: Brenda Finucane and Ann Swillen

9:15 AM Brenda Finucane
Symptoms Versus Causes

9:30 AM Ann Swillen
Birth to Five
The Developing Brain
Motor Milestones versus Cognitive Ability
The Autistic Spectrum
Helping Children to Learn

10:00 AM Cindy Solot
Delays in Emergence of Language – Overcoming Obstacles

10:15 AM DISCUSSION

10:30 AM COFFEE BREAK – ASBURY LOBBY

Session 11: Education – The School Years
GRAND HARBOR BALLROOM SOUTH
Moderators: Barbara Haas-Givler and Ed Moss

11:00 AM Barbara Haas-Givler
Setting the Stage for Success: Effective Strategies to Enhance Social Skills

11:15 AM Tony Simon
Interventions Matter – Supporting Academic Achievements While Minimizing Stress

11:30 AM Carrie Bearden
The Brain Can Be Taught!

11:45 AM Jennifer Janusz
Executive Function Skills – Implications for Students

12:00 PM Ed Moss
Overcoming Academic Challenges Associated with Non-Verbal Learning Differences

12:15 PM DISCUSSION

12:45 PM LUNCH INTERNATIONAL STYLE! ASBURY A-C

Session 12: The Brain, Behavior and Transitions to Adulthood
GRAND HARBOR BALLROOM SOUTH
Moderators: Natalie Blagowidow & Sheila Kambin

1:30 PM Anne Bassett
Treating the Treatable – Emotions, Behavior and Thinking

2:00 PM Raquel Gur
Neuropsychiatric Features – Challenges and Approaches

2:15 PM DISCUSSION

2:45 PM Solveig Oskarsdottir
Educational Crossroads
Independent Living
Life Skills
Parenting
Grand Parenting

3:00 PM Natalie Blagowidow
Reproductive and Genetic Counseling Questions and Answers

3:15 PM DISCUSSION

3:30 PM AFTERNOON TEA – ASBURY LOBBY

Session 13: Two Sides of the Same Coin
GRAND HARBOR BALLROOM SOUTH
Moderators: Solveig Oskarsdottir and Sixto Garcia-Minaur

4:00 PM Donna McDonald-McGinn
Atypical 22q11.2 Deletions
The 22q11.2 Duplication Syndrome

4:10 PM Ann Swillen
Developmental Issues in children with 22q11.2 duplication syndrome

4:20 PM Cindy Solot
Speech and language in children with 22q11.2 duplication syndrome

4:30 PM DISCUSSION

Session 14: 22q – It’s Time You Knew - Rapid Fire Update
GRAND HARBOR BALLROOM SOUTH
Moderators: Anne Bassett and Donna McDonald-McGinn

(Family Program continued)
Tuesday, July 10th
(Family Program continued)

4:45 PM
Bernice Morrow
State of the International 22q11.2 Consortium — Why It Matters
Developing an International Registry — Why and How

4:55 PM
Ann Swillen
The Newly Established International Brain Behavior Consortium — How It Can Help

5:00 PM
Anne Bassett
Healthcare Guidelines — Keeping Them Current and Available for All

5:05 PM
Hillary Joyce
Standards in the United Kingdom

5:10 PM
Donna McDonald-McGinn
Establishing Clinical Centers of Excellence

5:15 PM
Michelle Breedlove-Sells
Resiliency Matters — And Awareness Too!

5:20 PM
Sheila Kambin
United We Stand

5:25 PM
Carol Cavanaugh
Celebrating Success and Continuing to Work Together

5:30 PM
DISCUSSION

6:00 PM
CLOSING REMARKS

MEETING ADJOURNS — NEXT UP?
SEE YOU IN 2014 IN MALLORCA, SPAIN!

Saturday, July 7th
PROFESSIONAL PROGRAM

7:00 AM
REGISTRATION OPEN,
Disney’s Yacht and Beach Club Resorts,
ASBURY ROTUNDA AND LOBBY

Session III -Of Mice and Men
GRAND HARBOR BALLROOM SOUTH
Moderators: Bernice Morrow and Peter Scambler

8:00 AM
Peter Scambler
Craniofacial Development

8:20 AM
Silvia E. Racedo*
Mouse Model Approach for Studying Human Congenital Heart Disease (CHD)

8:30 AM
Anthony Lamantia
Brain and Behavior

8:50 AM
Noboru Hiroi
Alterations of Social Behavior Through Genetic and Environmental Manipulation of the 22q11.2 Gene SEPT5 in the Mouse Brain

9:00 AM
Laurie R. Earls*
Age-Dependent MicroRNA Modulation of Ca2+ Dynamics and Synaptic Plasticity in Mouse Models of 22q11.2DS

9:10 AM
Christian Marshall
CNV’s in Autism, Intellectual Disability, and Neuro-Psychiatric Disease Lessons for 22q11.2DS

9:30 AM
DISCUSSION

9:45 AM
COFFEE BREAK –
SALONS 5-6

Session IV – Advances in Molecular Genetics
GRAND HARBOR BALLROOM SOUTH
Moderators: Anthony Lamantia and Christian Marshall

10:00 AM
Bernice Morrow
Human and Model Organism Approaches to Find Genetic Modifiers for 22q11.2DS

10:10 AM
Anne Chun-Hui Tsai
Chromosomal Microarray Analysis (CMA) in Patients with 22q11.2DS
Saturday, July 7th
(Professional Program continued)

10:20 AM Beata A. Nowakowska*
Targeted Re-sequencing of the Remaining
22q11.2 Region in Atypical Patients with
22q11.2DS

10:30 AM Beverly S. Emanuel
22q11.2DS: Second Hit CNVs and
Conotruncal Heart Defects

10:40 AM Estefania Pinero Martos*
Additional CNVs in 22q11.2: Meiotic Instability
in the Parent of Origin?

10:50 AM Flora Tassone
Decreased DGCRRB Levels Lead to Altered
MicroRNA Processing in Children with 22q11DS

11:00 AM DISCUSSION

Session V – Two Sides of the Same Coin?
Dups and Atypicals
GRAND HARBOR BALLROOM SOUTH
Moderators:
Damien Heine Suner and Hiroshi Kawame

11:15 AM Damien Heine Suner
Mutations in TBX1 & Clinical Characterization
of Two Patients with Mental Retardation and a
Rare Variant in the 5'UTR of the TBX1 Gene

11:25 AM Donna McDonald-McGinn
Phenotypic Findings in the Atypical Nested
22q11.2 “B-D” Deletion
A Report on 50 Individuals with 22q11.2
Duplication – A Genetic Counseling Conundrum

11:35 AM Sulagna Saitta
Molecular Analysis of Patients whose Clinical
Features Overlap 22q11.2DS

11:45 AM Ann Swillen
Neuro-Behavioural Deficits in Children (2-12
years) with 22q11.2 Duplication

11:55 AM Cynthia Solot
Description of Speech and Language in Eight
Children with 22q11.2 Duplication Syndrome

12:00 PM DISCUSSION
12:15 PM LUNCH -
SALONS 5-6

*Denotes Junior Investigator

Session VI – Healthcare Guidelines:
There’s an App for That!
GRAND HARBOR BALLROOM SOUTH
Moderators: Elizabeth Goldmuntz and
Kathleen Sullivan

1:00 PM Elizabeth Goldmuntz
Current Trends in Cardiology

1:10 PM Julia Kozlova*
22q11.2DS Study on Patients in the First Year of
Life in the Perinatal Cardiological Centre -
Moscow, Russian Federation

1:15 PM Kathleen Sullivan
Immunologic Update and Related Issues

1:30 PM Nancy Unanue
Thyroid Disease and Growth Hormone Deficiency

1:40 PM Lorraine Katz
Calcium and Parathyroid Hormone Levels in Youth

1:50 PM Anne Bassett
Potential Effects on Cognitive Development of
Neonatal Hypocalcemia

1:55 PM DISCUSSION

2:10 PM Michele Lambert
Hematologic Manifestations

2:20 PM Peggy Eicher
Feeding and Swallowing Issues

2:30 PM James Kelly*
Gut Inflammation

2:40 PM Dino Colo*
Scoliosis in 22q11.2DS
Orthopaedic Manifestations in 22q11.2DS

2:50 PM DISCUSSION

3:05 PM AFTERNOON TEA -
SALONS 5-6

Session VII – Structural and Functional
Brain Anomalies
GRAND HARBOR BALLROOM SOUTH
Moderators: Solveig Oskarsdottir & Peggy Eicher

3:25 PM David Lynch
Structural Brain Findings in 129 Individuals with
22q11.2DS

3:35 PM Stephan Eliez
Abnormal Course of Cortical Maturation

(Professional Program continued)
Saturday, July 7th  
(Professional Program continued)

3:45 PM  Elliott Beaton  
*Reduced Amygdala Volume and Elevated Anxiety in Children*

3:55 PM  Carrie Bearden  
*MRI Resting State Functional Connectivity*

4:05 PM  Yi Deng*  
*Altered Hippocampal Connectivity and Midline Brain Anomalies in Children with 22q11.2DS*

4:15 PM  DISCUSSION

**Session VIII – The Topic of Reproduction**  
**GRAND HARBOR BALLROOM SOUTH**  
Moderators: Natalie Blagowidow and Sheila Kambin

4:30 PM  Natalie Blagowidow  
*Prenatal Clues to the Diagnosis, Testing, Preimplantation Genetic Diagnosis, and Reproductive Issues in 22q11.2DS*

4:50 PM  Gregory Costain*  
*Reproductive Issues in Adults with 22q11.2 Deletion Syndrome*

5:00 AM  Louise Amlie – Wolf*  
*Renal Anomalies, Polyhydramnios and Single Umbilical Artery as Prenatal Clues to the Diagnosis of 22q11.2DS*

5:10 PM  Dana Faux*  
*Assessment of Parental Disclosure of a 22q11.2DS Diagnosis and Implications for Clinicians*

5:15 PM  DISCUSSION

5:30 PM  ADJOURN

6:30 PM  "22Q HULLABALOO" SPEAKER RECEPTION – ATLANTIC DANCE HALL  
*AT DISNEY’S BOARDWALK*

7:00 PM  2012 PROFESSIONAL GROUP PHOTO – ATLANTIC DANCE HALL  
*AT DISNEY’S BOARDWALK*

Sunday, July 8th  
PROFESSIONAL PROGRAM

7:00 AM  REGISTRATION OPEN,  
Disney’s Yacht and Beach Club Resorts,  
ASBURY ROTUNDA AND LOBBY

**Session IX – The Significance of Case Reports**  
**GRAND HARBOR BALLROOM SOUTH**  
Moderators: Karlene Coleman and Elaine Zackai

8:00 AM  Elaine Zackai  
*The Unique Challenge of Dual Diagnoses in 22q11.2DS*

8:05 AM  Karlene Coleman  
*22q11.2DS with Concomitant Diagnoses of Neurofibromatosis and Waardenburg Syndrome in Two Families*

8:10 AM  Paula Goldenberg  
*Left Ventricular Non-Compaction Cardiomyopathy (LVNC) and 22q11.2DS: Two Rare Diseases in One Individual or Syndromic Association?*

8:15 AM  Helena Ly*  
*Evaluation of Possible Association Between Leg Pain, Hypocalcemia and Muscle Weakness in Children with 22q11DS*

8:20 AM  Meredith Workman*  
*Cryptotia in 22q11.2DS: Case Report and Review of Common External Ear Anomalies*

8:25 AM  Ariela Nachmany  
*Platbyasia in 22q11.2DS*

8:30 AM  Adrian Baylis  
*Hypenasal Speech in the Presence of “Adequate” VP Closure in 22q11.2DS: A Case Report*

8:35 AM  Eva Chow  
*Familial 22q11.2 Duplications in a Father and Son Pair with Variable Phenotype but with Features Commonly Associated with 22q11.2DS*

8:40 AM  Hiroshi Kawame  
*Differential Diagnosis of 22q11.2DS: q23.1 Microdeletion Syndrome*

8:45 AM  DISCUSSION

**Session X – ENT, Speech and Language**  
**GRAND HARBOR BALLROOM SOUTH**  
Moderators: Brian Sommernad and Christina Persson

*Denotes Junior Investigator
<table>
<thead>
<tr>
<th>Time</th>
<th>Speaker</th>
<th>Title</th>
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<tbody>
<tr>
<td>11:05 AM</td>
<td>Maria Niarchou</td>
<td>Cognitive Function and Psychopathology in Children with 22q11.2DS</td>
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<tr>
<td>11:15 AM</td>
<td>Stephen Hooper</td>
<td>Longitudinal Neuropsychological Outcomes in Preadolescent Children with 22q11.2DS</td>
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<td>11:25 AM</td>
<td>Kathleen Angkutsiri</td>
<td>Autism in 22q11.2DS: Diagnostic Dilemmas of Dual Diagnoses</td>
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<td>11:30 AM</td>
<td>Sasja Duijff</td>
<td>Developmental Aspects of Psychiatric Symptoms in Youngsters with 22q11.2DS; A Focus on Psychosis</td>
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<tr>
<td>11:40 AM</td>
<td>Nancy Butcher</td>
<td>Adult Functioning in 22q11.2DS: Characterization &amp; Predictors of Adaptive Functioning in 100 Adults</td>
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<tr>
<td>11:50 AM</td>
<td>Therese Van Amelsvoort</td>
<td>22q11.2DS in Moderate and Severe Learning Disabled Adults - Relation between Psychosis and Cognitive Deterioration</td>
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<td>11:55 AM</td>
<td>Ren Evers</td>
<td>COMT val/met Polymorphism and Startle Reactivity in Adults with 22q11.2DS</td>
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<tr>
<td>12:00 PM</td>
<td>Linda Campbell</td>
<td>The Relationship Between Antisaccade, Prepulse Inhibition, and Executive Dysfunction in 22q11.2DS</td>
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<tr>
<td>12:05 PM</td>
<td>Heather Shapiro</td>
<td>Impaired Cognitive Control and its Relationship to Anxiety, Adaptive Functioning, and Risk for Psychopathology</td>
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<tr>
<td>12:10 PM</td>
<td>Doron Gothelf</td>
<td>S-Adenosyl-L-Methionine (SAME) for the Treatment of Neuropsychiatric Symptoms in 22q11.2DS: A Double-Blind Placebo Controlled Trial</td>
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<tr>
<td>12:15 PM</td>
<td>DISCUSSION</td>
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<td>12:30 PM</td>
<td>LUNCH</td>
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**Session XI – Cognition, Behavior, Development and Functioning**

**GRAND HARBOR BALLROOM SOUTH**

Moderators: Ann Swillen and Paul Wang

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<tr>
<td>10:45 AM</td>
<td>Ann Swillen</td>
<td>Cognitive Profile Associated with the 22q11.2DS</td>
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<td>10:55 AM</td>
<td>Sasja Duijff</td>
<td>Cognitive and Behavioural Trajectories in 22q11.2DS from Childhood into Adolescence: Preliminary Results of a Prospective 6-year Follow-up Study</td>
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<td>12:15 PM</td>
<td>DISCUSSION</td>
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**Session XII – Psychiatric Illness, Possible Predictors and Amelioration**

**GRAND HARBOR BALLROOM SOUTH**

Moderators: Anne Bassett and Therese Van Amelsvoort

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<th>Time</th>
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<tr>
<td>1:15 PM</td>
<td>Paul Wang</td>
<td>Drug Discovery: Genes to Pathways to Therapeutics - Translational Research Lessons from Fragile X</td>
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</table>
Sunday, July 8th  
(Professional Program continued)

1:35 PM  Anne Bassett  
Psychosis and Related Neuropsychiatric Expression Across the Lifespan

1:45 PM  Joseph Cubells  
Simultaneous Assessment of Schizophrenia-Spectrum, Autism-Spectrum and Other Axis I Diagnoses in Adolescents and Young Adults with 22q11.2DS

1:55 PM  Maude Schneider*  
Patterns of Psychotic Symptoms in 22q11.2DS and Their Association with Cognitive Functioning - A Cluster Analysis

2:05 PM  Raquel Gur  
Brain and Behavior in 22q11.2DS

2:15 PM  Vandana Shashi  
Predictors of Anxiety Symptoms and Disorders in Children with 22q11.2 Deletion Syndrome: Further Understanding of the Underlying Pathophysiology

2:20 PM  Tony Simon  
Reduced Resolution of Spatial and Temporal Processing in Children with Chromosome 22q11.2DS and Implications for Functioning

2:30 PM  Petra Klaassen*  
Behavioral Problems in Children with 22q11DS - Comparing the Teachers' and Parents' Perspectives

2:40 PM  DISCUSSION

Session XIII – Remediation  
GRAND HARBOR BALLROOM SOUTH  
Moderators: Eva Chow and Tony Simon

2:55 PM  Wendy Kates  
Computerized Cognitive Remediation in Adolescents: A Preliminary Report

3:05 PM  Stephan Eliez  
Vis-à-vis: Socio-Emotional Remediation

3:15 PM  Eva Chow  
Transition to Psychosis in a Clinical Sample of Adolescents with 22q11.2DS  
Aripiprazole in Patients with 22q11.2DS and Schizophrenia

3:25 PM  Margaret Souders  
Sleep Behaviors and Solutions

3:30 PM  Betsy Leech  
Fostering Resilience in Families

3:35 PM  Eileen Marrinam  
Social Communication Correlates of Psychiatric Disorders in 22q11.2DS

3:40 PM  Donna Cutler-Landsman  
Changes Over Time - Educational Implications

3:45 PM  DISCUSSION

4:00 PM  AFTERNOON TEA -  
SALONS 5-6

Session XIV – Multidisciplinary Matters  
GRAND HARBOR BALLROOM SOUTH  
Moderators: Sixto Garcia-Minaur and Alex Habel

4:15 PM  Alice Bailey*  
22q11.2DS Centers of Excellence – How and Why

4:20 PM  Joan Atkin  
An Interdisciplinary Team Approach for a 22q11.2DS Center

4:25 PM  Alan Fung  
Strategic Planning for a Multidisciplinary Adult 22q11.2 Deletion Syndrome Clinic in a Canadian General Hospital Setting  
Coordinating and Managing a Clinic for Adults with 22q11.2 Deletion Syndrome

4:30 PM  Clodagh Murphy  
Transition of Care from Child to Adult Mental Health Services

4:35 PM  Annette Hogan  
Caring for the Pediatric Patient with 22q11.2DS: The Role of the Primary Care Physician and the Medical Home

4:40 PM  Meg Maguire  
Navigating the Electronic Record and Developing Perioperative Order Sets

4:45 PM  Robert Hopkin  
Need for Hospitalization Analysis from Birth to Age 10

4:50 PM  Maria Mascarenhas  
Feeding and Gastrointestinal Manifestations are the Most Difficult Issues for Families to Navigate

4:55 PM  Brian Forbes  
Why an Eye Exam is Included in the Guidelines - Ophthalmologic Findings in Children
Sunday, July 8<sup>th</sup>  
(Professional Program continued)

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<tr>
<td>5:00 PM</td>
<td>Rosario <strong>Mayro</strong></td>
<td>Demystifying Dental Issues</td>
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<tr>
<td>5:05 PM</td>
<td>Alex <strong>Habel</strong></td>
<td>Specific Growth Curves for 22q11.2DS</td>
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<td>5:15 PM</td>
<td>DISCUSSION</td>
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**Session XV – Moving Ahead**  
**GRAND HARBOR BALLROOM SOUTH**

Moderators:  
Anne Bassett and Donna McDonald-McGinn

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<tr>
<td>5:30 PM</td>
<td>Bernice <strong>Morrow</strong></td>
<td>International Phenotypic Modifier Consortium Outcomes Update</td>
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<tr>
<td>5:35 PM</td>
<td>Ann <strong>Swillen</strong></td>
<td>International Brain-Behavior Consortium Update</td>
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<tr>
<td>5:40 PM</td>
<td>Sixto <strong>Garcia-Minaur</strong></td>
<td>Developing Registries</td>
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| 5:45 PM | Anne **Bassett** and Donna **McDonald-McGinn** | Challenges for the Future  
Clinical Guidelines - Keeping Them Current and Available for All!  
Opportunities for Cross-Fertilization  
Developing a Professional Society  
Establishing Criteria for Clinical Centers of Excellence |
| 6:00 PM | DISCUSSION       |                                                 |
| 6:15 PM | Anne Bassett, Damien **Heine Suner**, and Sixto **Garcia-Minaur** | Junior Investigator Award  
Closing Remarks |
| 6:30 PM | PROFESSIONAL PROGRAM CONCLUDES | SEE YOU IN 2014  
IN MALLORCA, SPAIN! |

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<tr>
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| 7:30 PM | **"22Q BARBEQUE" CHARACTER DINNER FOR ALL**  
*Denotes Junior Investigator* |

*Denotes Junior Investigator*