

THE 8TH BIENNIAL INTERNATIONAL 22Q11.2DS CONFERENCE



The 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

The Week at a Glance

CONFERENCE AT A GLANCE

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

Friday July 6	Saturday July 7	Sunday July 8	Monday July 9	Tuesday July 10
<p>9:00 AM—3:30 PM</p> <p>Professional Pre-Conference Symposium</p> <p>International Brain and Behavior Consortium</p>	<p>8:00 AM—5:30 PM</p> <p>Professional Program Continues</p> <p>Sessions III—VIII</p> <p>Grand Harbor Ballroom South</p>	<p>8:00 AM—6:30 PM</p> <p>Professional Program Continues</p> <p>Sessions IX—XV</p> <p>Grand Harbor Ballroom South</p>	<p>8:00 AM—6:00 PM</p> <p>Family Program</p> <p>Sessions 3—8</p> <p>Grand Harbor Ballroom South</p>	<p>8:00 AM—12:45 PM</p> <p>Family Program Continues</p> <p>Sessions 9—11</p> <p>Grand Harbor Ballroom South</p>
<p>4:00 PM—5:00 PM</p> <p>Opening Reception for All Asbury Rotunda</p>	<p>6:30 PM—8:30 PM</p> <p>Speakers Reception with Group Photo Atlantic Dance Hall</p>	<p>10:00 AM—11:30 AM</p> <p>Simultaneous Family Programs</p> <p>Session C</p> <p>Older Children, Adult and Adolescent Workshops</p> <ul style="list-style-type: none"> • Capture the Moment • Working Things Through—Together 	<p>12:30 PM—1:15 PM</p> <p>Lunch 22q Style</p> <p>Salons 5—8</p> <p>1:15 PM</p> <p>2012 Family Group Photo</p>	<p>12:45 PM—1:30 PM</p> <p>Lunch International Style</p> <p>Asbury A—C</p>
<p>5:00 PM—5:30 PM</p> <p>Combined Family & Professional Program</p> <p>Grand Harbor Ballroom South</p> <ul style="list-style-type: none"> • Welcome • Parade of Nations • Angelo DiGeorge Memorial Medal 	<p>10:00 AM—11:30 AM</p> <p>Simultaneous Family Programs</p> <p>Session A</p> <p>Children's Programs and Adult and Adolescent Workshops</p> <ul style="list-style-type: none"> • Making Memories • Getting to Know You 	<p>1:30 PM—3:00 PM</p> <p>Simultaneous Family Programs</p> <p>Session D</p> <p>Adult and Adolescent Workshops</p> <ul style="list-style-type: none"> • Riding Things Through with 22q • Challenges with Solutions 	<p>10:00 AM—11:30 AM</p> <p>Simultaneous Family Program</p> <p>Session E</p> <p>Older Children, Adolescents and Adults with 22q11.2DS Workshop</p> <ul style="list-style-type: none"> • Put on a Happy Face 	<p>1:30 AM—6:00 PM</p> <p>Family Program Continues</p> <p>Sessions 12—14</p> <p>Grand Harbor Ballroom South</p>
<p>5:30 PM—6:30 PM</p> <p>Session 1</p> <p>Grand Harbor Ballroom South</p>	<p>1:30 PM—3:00 PM</p> <p>Simultaneous Family Programs</p> <p>Session B</p> <p>Adult and Adolescent Workshops</p> <ul style="list-style-type: none"> • One for All and All for Fun • Now What? 	<p>7:30 PM—9:00 PM</p> <p>22q Barbeque Character Dinner (Ticket required) Marina/Boardwalk</p>	<p>1:30 PM—3:00 PM</p> <p>Simultaneous Family Program</p> <p>Session F</p> <p>Adult Workshop</p> <ul style="list-style-type: none"> • Mind Over Matter 	<p>Meeting Adjourns</p> <p>See You in Mallorca, Spain in 2014!</p>
<p>6:30 PM—8:00 PM</p> <p>Session 2</p> <p>Grand Harbor Ballroom South</p>				

Combined Family & Professionals Program

Friday, July 6th

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**

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**

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*

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

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**

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*

Friday, July 6th
(Combined Family
& Professionals
Program continued)

- 7:00 PM Aoy **Mitchell**
*Newborn Screening –
How, Why and When?*
- 7:10 PM Laird **Jackson**
*22q11.2DS Array
Results in Consecutive
Prenatal Diagnostic
Screenings – A Window
Into Prevalence*
- 7:20 PM Melissa K.
Maisenbacher
*Prevalence of
22q11.2DS in Products
of Conception Lend
Insight Into
Non-Invasive Prenatal
Diagnostic Testing*
- 7:30 PM Thomas **Werge**
*Estimating Incidence
Rates and Clinical
Profiles Using Danish
Health Registers and
Biobanks*
- 7:40 PM Peter **Scambler**
Summing It Up
- 7:45 PM **DISCUSSION**
- 8:00 PM **ADJOURN –
DINNER ON
YOUR OWN**

Family Program

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
- B.3 Dealing with Behavioral Differences During Adolescence and Adulthood**
(Parents, Grandparents and Caregivers) –
Raquel **Gur**, Daneen **Whinna** and Margaret **Souders**, ASBURY HALL B
- B.4 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/BOARDWALK

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 Cavana,
Donna McDonald-McGinn & Wendy Rose

8:10 AM Carol Cavana, 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 Anniqne **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 Cavana**
*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!

Professional Program

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 DGCR8 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 **Amlië – 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**
Platybasia in 22q11.2DS
- 8:30 AM Adriane **Baylis**
Hypernasal 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: 2q33.1 Microdeletion Syndrome
- 8:45 AM DISCUSSION

Session X – ENT, Speech and Language GRAND HARBOR BALLROOM SOUTH

Moderators: *Brian Sommerlad and Christina Persson*

*Denotes Junior Investigator

Sunday, July 8th

(Professional Program continued)

- 9:00 AM **Brian Sommerlad**
What a Difference a Palate Makes - Evaluation, Surgery, Timing and Recovery
- 9:15 AM **Nicole Spruijt***
In Search for the Optimal Surgical Treatment for Velopharyngeal Dysfunction in 22q11.2DS: A Systematic Review
- 9:25 AM **Cynthia Solot**
Timing for Pharyngoplasty in Children with 22q11.2DS and Velopharyngeal Dysfunction – Why Should we Wait
- 9:35 AM **Christina Persson**
Acoustic and Perceptual Characteristics of Speech in 22q11.2DS: Measures of Voice Onset Time and Syllable Durations Related to Articulation and Prosody
- 9:45 AM **Carrie Stransky***
Critical Phenotypic Manifestations of 22q11.2DS in Patients Requiring Surgery for Velopharyngeal Insufficiency
- 9:55 AM **Adriane Baylis**
Motor Speech Deficits in the 22q11.2DS
- 10:00 AM **Meredith Workman***
Quantitative Nasopharyngoscopic Assessment of Velopharyngeal Closure in 22q11.2DS
- 10:05 AM **Solveig Oskarsdottir**
Important ENT considerations

10:15 AM **DISCUSSION**

10:30 AM **COFFEE BREAK - SALONS 5-6**

Session XI – Cognition, Behavior, Development and Functioning

GRAND HARBOR BALLROOM SOUTH

Moderators: *Ann Swillen and Paul Wang*

- 10:45 AM **Ann Swillen**
Cognitive Profile Associated with the 22q11.2DS
- 10:55 AM **Sasja Duijff***
Cognitive and Behavioural Trajectories in 22q11.2DS from Childhood into Adolescence: Preliminary Results of a Prospective 6-year Follow-up Study

- 11:05 AM **Maria Niarchou***
Cognitive Function and Psychopathology in Children with 22q11.2DS
- 11:15 AM **Stephen Hooper**
Longitudinal Neuropsychological Outcomes in Preadolescent Children with 22q11.2DS
- 11:25 AM **Kathleen Angkustsiri***
Autism in 22q11.2DS: Diagnostic Dilemmas of Dual Diagnoses
- 11:30 AM **Sasja Duijff**
Developmental Aspects of Psychiatric Symptoms in Youngsters with 22q11.2DS; A Focus on Psychosis
- 11:40 AM **Nancy Butcher**
Adult Functioning in 22q11.2DS: Characterization & Predictors of Adaptive Functioning in 100 Adults
- 11:50 AM **Therese Van Amelsvoort**
22q11.2DS in Moderate and Severe Learning Disabled Adults - Relation between Psychosis and Cognitive Deterioration
- 11:55 AM **Ren Evers**
COMT val/met Polymorphism and Startle Reactivity in Adults with 22q11.2DS
- 12:00 PM **Linda Campbell**
The Relationship Between Antisaccade, Prepulse Inhibition, and Executive Dysfunction in 22q11.2DS
- 12:05 PM **Heather Shapiro**
Impaired Cognitive Control and its Relationship to Anxiety, Adaptive Functioning, and Risk for Psychopathology
- 12:10 PM **Doron Gothelf**
S-Adenosyl-L-Methionine (SAME) for the Treatment of Neuropsychiatric Symptoms in 22q11.2DS: A Double-Blind Placebo Controlled Trial
- 12:15 PM **DISCUSSION**
- 12:30 PM **LUNCH - SALONS 5-6**

Session XII – Psychiatric Illness, Possible Predictors and Amelioration

GRAND HARBOR BALLROOM SOUTH

Moderators:

Anne Bassett and Therese Van Amelsvoort

- 1:15 PM **Paul Wang**
Drug Discovery: Genes to Pathways to Therapeutics - Translational Research Lessons from Fragile X

(Professional Program continued)

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 **Annique 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 8th

(Professional Program continued)

- 5:00 PM Rosario **Mayro**
Demystifying Dental Issues
- 5:05 PM Alex **Habel**
Specific Growth Curves for 22q11.2DS

5:15 PM **DISCUSSION**

Session XV – Moving Ahead

GRAND HARBOR BALLROOM SOUTH

Moderators:

Anne Bassett and Donna McDonald-McGinn

- 5:30 PM Bernice **Morrow**
International Phenotypic Modifier Consortium
Outcomes Update
- 5:35 PM Ann **Swillen**
International Brain-Behavior Consortium Update
- 5:40 PM Sixto **Garcia-Minaur**
Developing Registries
- 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!**

- 7:30 PM **"22Q BARBEQUE" CHARACTER
DINNER FOR ALL –**
Disney's Yacht and Beach Club Resorts
MARINA/BOARDWALK

Notes...

at a Glance

*Denotes Junior Investigator