



# 22q11.2 Deletion



Bringing the 22q11.2 Deletion  
Into the 21st Century  
*Second International Conference for  
Families and Professionals*

June 22-25, 2000  
Penn Tower Hotel  
Philadelphia, Pa., U.S.A.

Sponsored by

 The Children's Hospital of Philadelphia  
*A pediatric healthcare network*



## WEEK AT A GLANCE

### THURSDAY

### FRIDAY

### SATURDAY

### SUNDAY

7:00 a.m. - 8:00 a.m.  
Continental  
Breakfast,  
CHOP, 1<sup>st</sup> floor,  
Stokes Rotunda

7:00 a.m. - 8:00 a.m.  
Continental  
Breakfast,  
CHOP, 1<sup>st</sup> floor,  
Stokes Rotunda

7:00 a.m. - 8:00 a.m.  
Continental  
Breakfast,  
CHOP, 1<sup>st</sup> floor,  
Stokes Rotunda

7:30 a.m. - 6:00 p.m.  
Registration  
Penn Tower Hotel,  
2<sup>nd</sup> floor, Ballroom  
Foyer

7:30 a.m. - 6:00 p.m.  
Registration  
Penn Tower Hotel,  
2<sup>nd</sup> floor, Ballroom  
Foyer

7:30 a.m. - 1:00 p.m.  
Registration  
Penn Tower Hotel,  
2<sup>nd</sup> floor, Ballroom  
Foyer

8:00 am - 1:00 p.m.  
Conference in  
Session  
Penn Tower Hotel,  
2nd floor, Ballroom

8:00 am - 1:15 p.m.  
Conference in  
Session  
Penn Tower Hotel,  
2nd floor, Ballroom

8:00 am - 1:15 p.m.  
Conference in Session  
Penn Tower Hotel,  
2nd floor, Ballroom

12:00 p.m. - 8:00 p.m.  
Registration  
Penn Tower Hotel,  
2<sup>nd</sup> floor, Ballroom  
Foyer

1:15 p.m. - 1:30 p.m.  
Special Performance

1:15 p.m.  
Conference Adjourns

1:00 p.m. - 2:15 p.m.  
\* General Lunch,  
Location on ticket

1:30 p.m. - 2:30 p.m.  
\* General Lunch,  
Location on ticket

3:00 p.m. - 4:00 p.m.  
Special Presentation  
for Parents and  
Non-genetics  
Professionals  
Penn Tower Hotel,  
2nd floor, Ballroom

2:15 p.m. - 5:45 p.m.  
Conference in Session  
Penn Tower Hotel,  
2nd floor, Ballroom

2:30 p.m. - 6:30 p.m.  
Conference in Ses-  
sion: Concurrent Ses-  
sions  
See Program for  
Specific Location

6:00 p.m. - 8:35 p.m.  
Welcome and  
Historical  
Overview  
Penn Tower Hotel,  
2nd floor, Ballroom

6:00 p.m.  
Star Spangled  
Celebration



The Second International Conference Program

Thursday, June 22, 2000

Noon Conference Registration Penn Tower Hotel, 2<sup>nd</sup> floor, Ballroom Foyer

Special Presentation Penn Tower Hotel, 2<sup>nd</sup> floor, Ballroom

3 p.m. Special Presentation for Parents and Non-genetics  
Professionals: What is the 22q11.2 Deletion?  
*Donna M. McDonald-McGinn, M.S., C.G.C.*  
*Philadelphia, Pennsylvania, U.S.A.*

IS1

General Conference Session Penn Tower Hotel, 2<sup>nd</sup> floor, Ballroom

6 p.m. Welcome and Introduction  
*Donna M. McDonald-McGinn, M.S., C.G.C.*

The Sunshine Singers

Historical Overview

Moderators: *Donna M. McDonald-McGinn, M.S., C.G.C.*  
*Claude Stoll, M.D.*

6:15 p.m. DiGeorge Syndrome  
*Angelo DiGeorge, M.D.*  
*Philadelphia, Pennsylvania, U.S.A.*

IS2

6:35 p.m. Velocardiofacial Syndrome  
*Rosalie Goldberg, M.S., C.G.C.*  
*Bronx, New York, U.S.A.*

IS3

6:55 p.m. Conotruncal Anomaly Face Syndrome  
*Atsuyoshi Takao, M.D.*  
*Tokyo, Japan*

IS4



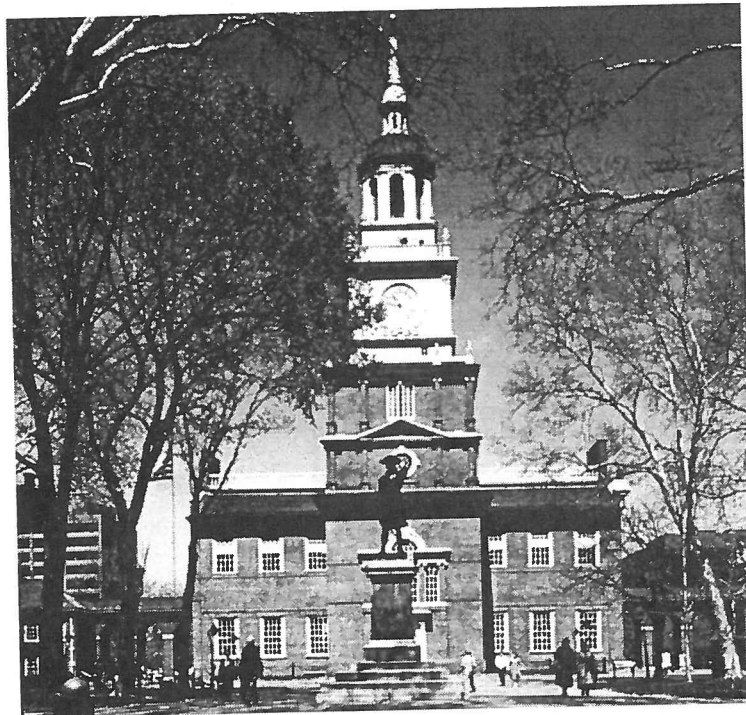


- 7:15 p.m. **The Philadelphia Story: Clinical and Molecular Findings in the 22q11.2 Deletion**  
*Elaine H. Zackai, M.D.*  
*Beverly S. Emanuel, Ph.D.*  
*Philadelphia, Pennsylvania, U.S.A.*
- 7:55 p.m. **Clinical Findings in the Belgium Population**  
*Koen Devriendt, M.D., Ph.D.*  
*Leuven, Belgium*
- 8:15 p.m. **The 22q11 Deletion Syndrome: A Multidisciplinary Project in Sweden**  
*Sólveig Óskarsdóttir, M.D.*  
*Göteborg, Sweden*
- 8:35 p.m. **Adjourn**

IS5

IS6

IS7





Friday, June 23, 2000

7:00 a.m. Continental Breakfast CHOP, Main Building, 1<sup>st</sup> floor, Stokes Rotunda

7:30 a.m. Conference Registration Penn Tower Hotel, 2<sup>nd</sup> floor, Ballroom Foyer

8:00 a.m. General Conference Session I Penn Tower Hotel, 2<sup>nd</sup> floor, Ballroom

Moderators: Bruno Marino, M.D.  
Atsuyoshi Takao, M.D.

8 a.m. Cardiac Abnormalities: An International Perspective

*Bruno Marino, M.D.*

*Rome, Italy*

*Elizabeth Goldmuntz, M.D.*

*Philadelphia, Pennsylvania, U.S.A.*

*Atsuyoshi Takao, M.D.*

*Tokyo, Japan*

IS8

IS9

IS10

8:45 a.m. Tetralogy of Fallot (TOF) Associated With  
Chromosome 22q11 Deletion in Adolescents and  
Young Adults

*Momma, Matsuoka, Takao*

*Tokyo, Japan*

A1

8:55 a.m. 22q11 Microdeletions in Adults With Familial  
Tetralogy of Fallot

*Hokanson, Pierpont, Hirsch, Moller*

*Minneapolis, Minnesota, U.S.A.*

A2

9:05 a.m. Identifying the Cardiac Patient With a 22q11  
Deletion: Cardiac and Extracardiac Features

*Shores, Jawad, Driscoll, Stouffer, Kenton,*

*Hoess, Mitchell, Goldmuntz*

*Philadelphia, Pennsylvania, U.S.A.*

A3



9:15 a.m.	<b>Pulmonary Atresia/VSD (PA-VSD): Prevalence of Deletion 22q11 in the Different Anatomic Patterns</b> <i>Anaclerio, Marino, Toscano, Gitto, Digilio, Giannotti, Dallapiccola</i> <i>Rome, Italy</i>	A4
9:25 a.m.	<b>Outcomes of Cardiothoracic Surgery</b> <i>J. William Gaynor, M.D.</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	IS11
9:40 a.m.	<b>Questions and Answers</b>	
9:50 a.m.	<b>Break</b>	Penn Tower Hotel, 2 <sup>nd</sup> floor, Assembly Room
<b>Moderators:</b> Kathleen Sullivan, M.D., Ph.D John Riski, Ph.D., F.A.S.H.A., C.C.C.-S.		
10:15 a.m.	<b>Immunodeficiency and Rheumatologic Findings in the 22q11.2 Deletion Syndrome</b> <i>Kathleen Sullivan, M.D., Ph.D.</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	IS12
10:35 a.m.	<b>T-Cell Receptor Repertoire and Function in DiGeorge/Velocardiofacial Syndrome</b> <i>Pierdominici, Marziali, Giovannetti, Oliva, Rosso, Marino, Digilio, Giannotti, Novelli, Aiuti, Pandolfi</i> <i>Rome, Italy</i>	A5
10:45 a.m.	<b>The 22q11 Deletion Syndrome: A Multidisciplinary Project in Sweden: Immunological Aspects</b> <i>Óskarsdóttir, Fasth</i> <i>Göteborg, Sweden</i>	A6
10:55 a.m.	<b>Immunologic Characteristics of Catch 22 Syndrome</b> <i>Ryu, Jo, Kil, Lee</i> <i>Taejon, Republic of Korea</i>	A7



11:05 a.m.	<b>Natural History of the Immunodeficiency in Chromosome 22q11.2 Deletion Syndrome</b> <i>Sullivan, McDonaldMcGinn, Zackai, Jarwad</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	A8
11:15 a.m.	<b>Graves Disease in Patients With a 22q11.2 Deletion</b> <i>Kawame, Adachi, Tachibana, Masuno, Kurosawa, Ochiai</i> <i>Kanagawa, Japan</i>	A9
11:25 a.m.	<b>Questions and Answers</b>	
11:35 a.m.	<b>Endocrinology of the 22q11 Deletion Syndrome</b> <i>Stuart Weinzeimer, M.D.</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	IS13
11:50 a.m.	<b>Feeding Issues: Old Challenges, New Solutions</b> <i>Peggy Eicher, M.D.</i> <i>Yardley, Pennsylvania, U.S.A.</i>	IS14
12:10 p.m.	<b>Palatal Abnormalities</b> <i>Richard Kirschner, M.D.</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	IS15
12:25 a.m.	<b>Outcomes of Speech Measures Using Instrumentation</b> <i>John Riski, Ph.D., F.A.S.H.A., C.C.C.-S.</i> <i>Atlanta, Georgia, U.S.A.</i>	IS16
12:40 p.m.	<b>Videofluoroscopic Evaluation of Velopharyngeal Function in Children With Velocardiofacial Syndrome</b> <i>Rommel, Vantrappen, Hermans, Smet, Devriendt, Feenstra, Fryns</i> <i>Leuven, Belgium</i>	A10
12:50 p.m.	<b>Questions and Answers</b>	
1 p.m.	<b>Lunch</b>	See your Ticket for Location



2:15 p.m. General Conference SessionII Penn Tower Hotel, 2<sup>nd</sup> floor, Ballroom

Moderators: Linda D'Antonio, Ph.D.  
Richard Kirschner, M.D

- 2:15 p.m. Ear, Nose and Throat Findings IS17  
*Ian Jacobs, M.D.*  
*Philadelphia, Pennsylvania, U.S.A.*
- 2:30 p.m. The 22q11 Deletion Syndrome— A11  
A Multidisciplinary Project in Sweden:  
Audiological Aspects  
*Jönsson, Óskarsdóttir, Hanner*  
*Göteborg, Sweden*
- 2:40 p.m. Tracheal Agenesis in the 22q11.2 Deletion A12  
*Mable, McDonald-McGinn, Russell, Zackai*  
*Philadelphia, Pennsylvania, U.S.A.*
- 2:50 p.m. Questions and Answers
- 3 p.m. Communication Issues  
*Nancy Scherer, Ph.D.* IS18  
*Johnson City, Tennessee, U.S.A.*  
*Linda D'Antonio, Ph.D.* IS19  
*Loma Linda, California, U.S.A.*  
*Cynthia Solot, M.A., CCL-S* IS20  
*Philadelphia, Pennsylvania, U.S.A.*
- 3:40 p.m. 22q11 Deletion Syndrome— A13  
A Multidisciplinary Project in Sweden:  
Speech and Language Aspects  
*Persson, Óskarsdóttir*  
*Göteborg, Sweden*
- 3:50 p.m. Questions and Answers





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4 p.m.      Break      Penn Tower Hotel, 2<sup>nd</sup> floor, Assembly Room

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Moderators: Paula Goldenberg  
Kimberly Grace, M.S.

4:20 p.m.      **Exceptional Parents**  
*Joseph M. Valenzano Jr.*  
*Oradel, New Jersey, U.S.A.*

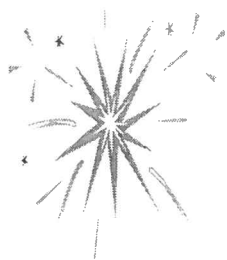
IS21

4:45 p.m.      **International Parents Panel**  
*Paula Goldenberg, U.S.A.*  
*Julie Squain, U.K.*  
*Dominique Pfeiffer, France*  
*Alex Bakalinsky, Ukraine*  
*Josie Snyders, Canada*

5:30 p.m.      **Faces of Sunshine**

5:45 p.m.      **Adjourn**

6 p.m.      **Red, White and Blues: A Star Spangled Celebration**  
at the Leonard and Madlyn Abramson Pediatric Research Center



Red, White and Blues:  
A Star Spangled Celebration

From many countries near and far  
From all across the nation  
We gathered here for thought and  
cheer  
And now a celebration

Philly food and spirits now  
Mummers and face painting  
Dancing while the night is young  
Until the band stops playing!





Saturday, June 24, 2000

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7:00 a.m. Continental Breakfast CHOP, Main Building, 1<sup>st</sup> floor, Stokes Rotunda

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8:00 a.m. General Conference Session I Penn Tower Hotel, 2<sup>nd</sup> floor, Ballroom

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Moderators: Ann Swillen, M.S.  
Eva Chow, M.D.

- |           |  |      |
|-----------|--|------|
| 8 a.m.    | <b>Neurological Manifestations of Patients with 22q11 Deletions</b><br><i>David Lynch, M.D., Ph.D.</i><br><i>Philadelphia, Pennsylvania, U.S.A.</i>  | IS22 |
| 8:15 a.m. | <b>Functional MRI Results</b><br><i>Stephan Eliez, M.D., Ph.D.</i><br><i>Stanford, California, U.S.A.</i>  | IS23 |
| 8:30 a.m. | <b>Structural and Functional Neuroanatomy in the 22q Deletion Syndrome</b><br><i>Carrie Bearden, M.D.</i><br><i>Philadelphia, Pennsylvania, U.S.A.</i>   | IS24 |
| 8:45 a.m. | <b>The Association Between White Matter Anomalies and Cognitive Functioning in Velocardiofacial Syndrome</b><br><i>Kates, Burnette, Kaufmann, Pearlson</i><br><i>Baltimore, Maryland, U.S.A.</i>                     | A14  |
| 8:55 a.m. | <b>Brain Abnormalities Associated With Deletion at 22q11: Implications for Psychosis</b><br><i>van Amelsvoort, Daly, Critchley, Robertson, Henry, Suckling, Owen, K. Murphy, D. Murphy</i><br><i>London, England</i> | A15  |
| 9:05 a.m. | <b>Questions and Answers</b>   |      |



9:15 a.m.	<b>Aspects of Development</b> <i>Ann Swillen, M.S.</i> <i>Leuven, Belgium</i>	IS25
	<i>Edward Moss, Ph.D.</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	IS26
		A16
9:45 a.m.	<b>An Investigation of the Neuropsychological Profile in Velocardiofacial Syndrome</b> <i>Henry, van Amelsvoort, Morris, Owen, K. Murphy, D. Murphy</i> <i>London, England</i>	
		A17
9:55 a.m.	<b>Cognitive Development and Behavioral Manifestations in Children With Velocardiofacial Syndrome</b> <i>Freier, Freeman, D'Antonio, Ermshar, Vaz</i> <i>Loma Linda, California, U.S.A.</i>	
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10:05 a.m.	<b>Questions and Answers</b>	
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10:15 a.m.	<b>Break</b>	<b>Penn Tower Hotel, 2<sup>nd</sup> floor, Assembly Room</b>
Moderators:	<b>Anne S. Bassett, M.D., FRCPC</b> <b>Kieran Murphy, M.D., M.R.C. Psych</b>	
10:40 a.m.	<b>Overview of Developmental and Educational Needs</b> <i>Marsha Gerdes, Ph.D.</i> <i>Michael Woodin, Ph.D.</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	IS27
11:10 a.m.	<b>Educating the Educators</b> <i>Brenda Finucane M.S., C.G.C.</i> <i>Elwyn, Pennsylvania, U.S.A.</i>	IS28
11:25 a.m.	<b>Questions and Answers</b>	
11:35 a.m.	<b>Psychiatric Issues</b> <i>Anne S. Bassett, M.D., FRCPC</i> <i>Toronto, Canada</i> <i>Kieran Murphy, M.D., M.R.C. Psych</i> <i>London, England</i> <i>Naushad Jessani, M.D.</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	IS29 IS30 IS31



- 12:20 p.m.    **The 22q11 Deletion Syndrome—  
A Multidisciplinary Project in  
Goteborg, Sweden: Some Neuropsychiatric  
and Neuropsychological Aspects**    **A18**  
*Niklasson, Rasmussen, Óskarsdóttir, Gillberg*  
*Göteborg, Sweden*
- 12:30 p.m.    **Neurobehavioral Phenotype of the 22q11.2 Deletion  
Syndrome**    **A19**  
*Bearden, Woodin, Moss, Wang, Emanuel,*  
*McDonald-McGinn, Zackai*  
*Philadelphia, Pennsylvania, U.S.A.*
- 12:40 p.m.    **Early Signs of Psychiatric Problems in Patients with  
Velocardiofacial Syndrome**    **A20**  
*Vogels, Swillen, Devriendt, Fryns*  
*Leuven, Belgium*
- 12:50 p.m.    **Neurocognition in 22q11 Deletion Syndrome;  
Adults With and Without Schizophrenia**    **A21**  
*Chow, Watson, Janiszewski, Young, Weksberg, Bassett*  
*Toronto, Canada*
- 1 p.m.                            **Questions and Answers**
- 1:15 p.m.    **Special Performance by Michelle Kelly**  
*“Just One Day” and “In My Hands”*
- 
- 1:30 p.m.    **Lunch**                            **See your Ticket for Location**
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## Concurrent Sessions

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### I. Basic Science Research Sessions

Penn Tower Hotel, 2<sup>nd</sup> floor, Ballroom

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Moderators: Peter Scambler, M.D.  
Marcia Budarf, Ph.D.

2:30 p.m. **Animal Models**

*Antonio Baldini, M.D.*

*Houston, Texas, U.S.A.*

*Raju Kucherlapati, Ph.D.*

*Bronx, New York, U.S.A.*

*Roger Reeves, Ph.D.*

*Baltimore, Maryland, U.S.A*

IS32

IS33

IS34

3:30 p.m. **Questions and Answers**

3:40 p.m. **Mechanisms of the Deletion**

*Beverly S. Emanuel, Ph.D.*

*Philadelphia, Pennsylvania, U.S.A.*

*Bernice Morrow, Ph.D.*

*Bronx, New York, U.S.A.*

IS35

IS36

4:20 p.m. **Questions and Answers**

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4:30 p.m. **Break**

Penn Tower Hotel, 2<sup>nd</sup> floor, Assembly Room

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Moderators: Antonio Baldini, M.D.  
Beverly S. Emanuel, Ph.D.

4:45 p.m. **Genes in the Region**

*Peter Scambler, M.D.*

*London, England*

*Marcia Budarf, Ph.D.*

*Montreal, Quebec, Canada*

IS37

IS38

5:25 p.m. **Questions and Answers**





- 5:35 p.m. **Functional Analysis of Promoter Region of UFD1L, A Gene Commonly Deleted in DiGeorge and Velocardiofacial Syndromes** A22  
*Amati, Conti, Amicucci, Botta, Dallapiccola, Novelli*  
*Rome, Italy*
- 5:45 p.m. **Isolation and Molecular Characterization of a Gene (PAQ1/2) Deleted in DiGeorge and Velocardiofacial Syndromes** A23  
*Novelli, Mittler, Amati, Berti, Conti, Bengala, Botta, Dallapiccola, Meistererst*  
*Rome, Italy*
- 5:55 p.m. **ZNF74, a Gene Deleted in DiGeorge Syndrome, is Expressed in Thymus and Parathyroids, two glands which development is perturbed in patients** A24  
*Rarassard, Bazinet, Mallet, Aubry*  
*Paris, France and Montreal, Quebec*
- 6:05 p.m. **Elucidation of the DiGeorgelike Phenotype on Chromosome 10p** A25  
*Van Esch, Groenen, Fryns, Thakker, van de Ven, Devriendt*  
*Oxford, England*
- 6:15 p.m. **The 22q Deletion Syndromes: From Diversity to a Single Gene Theory** A26  
*De Decker, Lawrenson*  
*Cape Town, South Africa*
- 6:25 p.m. **Questions and Answers**
- 6:30 p.m. **Adjourn**



## Concurrent Sessions

### II. Psychosocial Issues

### Multiple Locations

#### 2:30 p.m. Psychosocial Issues

- |      |  |      |
|------|--|------|
|      |  | IS39 |
| A.   | <b>Developmental Stimulation for Your Child:<br/>Birth Through 5 Years</b><br><u>CHOP, Main Building, 1<sup>st</sup> floor, Stokes Auditorium</u><br><i>Marsha Gerdes, Ph.D.</i><br><i>Philadelphia, Pennsylvania, U.S.A.</i>  |      |
| B.   | <b>Financial Planning</b><br><u>Leonard and Madlyn Abramson Pediatric Research Center, 1<sup>st</sup> floor, 123-A/B</u><br><i>James Kalil, Ph.D.</i><br><i>Wilmington, Delaware, U.S.A.</i>   | IS40 |
| C.   | <b>Supporting the Unaffected Sibling:<br/>A Workshop for Parents</b><br><u>CHOP, Richard D. Wood Pediatric Ambulatory Care Center, Wood Clubs</u><br><i>Laura Bedrossian, B.S.</i><br><i>Philadelphia, Pennsylvania, U.S.A.</i><br><i>Kimberly Grace, M.S.</i><br><i>San Diego, California, U.S.A.</i> | IS41 |
| D.   | <b>Self Esteem: Support for the Older Child<br/>or Adult With the Deletion</b><br><u>Leonard and Madlyn Abramson Pediatric Research Center, 1<sup>st</sup> floor, 123-C</u><br><i>Brenda Finucane, M.S., C.G.C.</i><br><i>Elwyn, Pennsylvania, U.S.A.</i>  | IS42 |
| <br> |  |      |
|      | <b>3:45 p.m.</b>   |      |
| E.   | <b>Educational Interventions</b><br><u>CHOP, Main Building, 1<sup>st</sup> floor, Stokes Auditorium</u><br><i>Edward Moss, Ph.D.</i><br><i>Paul Wang, M.D.</i><br><i>Michael Woodin, Ph.D.</i><br><i>Philadelphia, Pennsylvania, U.S.A.</i><br><i>Ann Swillen, M.S.</i><br><i>Leuven, Belgium</i>      | IS43 |





Sunday, June 25, 2000

7:00 a.m. Continental Breakfast CHOP, Main Building, 1<sup>st</sup> floor, Stokes Rotunda

8:00 a.m. General Conference Session I Penn Tower Hotel, 2<sup>nd</sup> floor, Ballroom

Moderators: Koen Devriendt, M.D., Ph.D.  
Angelo DiGeorge, M.D.

8:00 a.m. Gastrointestinal and Nutritional Aspects IS50  
*David Piccoli, M.D.*  
*Philadelphia, Pennsylvania, U.S.A.*

8:15 a.m. Urologic Manifestations IS51  
*Douglas Canning, M.D.*  
*Philadelphia, Pennsylvania, U.S.A.*

8:30 a.m. Dental Issues IS52  
*Rosario Mayro, D.M.D., D.D.S.*  
*Philadelphia, Pennsylvania, U.S.A.*

8:45 a.m. Associated Ophthalmologic Abnormalities IS53  
*Jane Edmonds, M.D.*  
*Philadelphia, Pennsylvania, U.S.A.*

8:55 a.m. The 22q11 Deletion Syndrome— A27  
Ocular and Facial Characteristics  
*Stömland, Óskarsdóttir, Borén, Aring, Hellström*  
*Göteborg, Sweden*

9:05 a.m. Questions and Answers

9:10 a.m. Auxological Evaluation in Patients With A28  
Deletion 22q11  
*Digilio, Marino, Cappa, Cambiaso, Giannotti, Dallapiccola*  
*Rome, Italy*



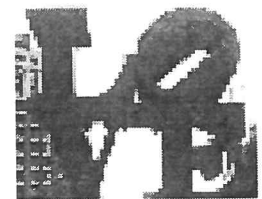
9:20 a.m.	<b>Height and Head Circumference in 122 Patients With the 22q11.2 Deletion</b> <i>Jawad, Stouffer, McDonald-McGinn, Zackai</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	A29
9:30 a.m.	<b>Cephalofacial Morphometrics in Children With the 22q11.2 Deletion</b> <i>Minugh-Purvis, Kirschner, LaRossa,</i> <i>McDonald-McGinn, Zackai</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	A30
9:40 a.m.	<b>Craniofacial Anthropometric Analysis in Patients With 22q11.2 Microdeletion</b> <i>Philip, Guyot, Dubuc, Dutour</i> <i>Marseille, France</i>	A31
9:50 a.m.	<b>Questions and Answers</b>	
10 a.m.	<b>Congenital Diaphragmatic Hernia in the 22q11.2 Deletion</b> <i>Russell, McDonald-McGinn, Mahale, Howell,</i> <i>Kasperski, Wilkes, Meyer-Cohen, Husted,</i> <i>Schauer, Adzick, Zackai</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	A32
10:10 a.m.	<b>Deletion 22q11 and Oculo-auriculo-vertebral (Goldenhar) Spectrum</b> <i>Digilio, Marino, Toscano, Giannotti, Dallapiccola</i> <i>Rome, Italy</i>	A33
10:20 a.m.	<b>Craniosynostosis in the 22q11.2 Deletion Syndrome</b> <i>Gripp, McDonald-McGinn, Kirschner, Husted,</i> <i>Schauer, Bartlett, Stolle, Driscoll, Emanuel, Pasquariello,</i> <i>Whitaker, Zackai</i> <i>Philadelphia, Pennsylvania, U.S.A.</i>	A34
10:40 a.m.	<b>Questions and Answers</b>	
10:50 a.m.	<b>Break</b>	<b>Penn Tower Hotel, 2<sup>nd</sup> floor, Assembly Room</b>





Moderators: Brenda Finucane, M.S., C.G.C.  
Nicole Philip, M.D.

- 11:10 a.m. **The DiGeorge/Velocardiofacial Syndrome  
Portuguese Experience** **A35**  
*Gaspar, Nogueira, Macedo, Anjos, Lourenço,  
Pinto, Reis, Soares, Arroz, KaKu, Feijóó  
Lisbon, Portugal*
- 11:20 a.m. **22q11 Microdeletion and Clinico-genetic  
Correlation in Catch 22 Syndrome** **A36**  
*Kil, Lee, Chung  
Taejeon, Republic of Korea*
- 11:30 a.m. **Audit of 479 Referrals to a FISH Laboratory  
for the Detection of Deletion 22q** **A37**  
*Robson, St. Heaps, Smith  
Westmead, Australia*
- 11:40 a.m. **Questions and Answers**
- 11:50 a.m. **Prenatal Diagnosis** **IS54**  
*Deborah Driscoll, M.D.  
Philadelphia, Pennsylvania, U.S.A.*
- 12:10 p.m. **Navigating the Healthcare System** **IS55**  
*Allan Bombard, Aetna U.S.Healthcare  
Oakland, California, U.S.A.*
- 12:30 p.m. **Pediatric Perspective on Clinical Management:  
A Take Home Message** **IS56**  
*Patrick S. Pasquariello Jr., M.D.  
Philadelphia, Pennsylvania, U.S.A.*
- 12:50 p.m. **Professional and Parent Recognition Awards  
Acknowledgements  
Closing Remarks**  
*Donna M. McDonald-McGinn, M.S., C.G.C.  
Elaine H. Zackai, M.D.  
Philadelphia, Pennsylvania, U.S.A.*



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