



22q11.2 Deletion Syndrome

Practical Implications for Behavior and Learning

Atlanta Georgia
April 21 - 22, 2006

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Websites

<http://www.geocities.com/mitorelvis/vcfs.html>

Email your questions to Robert and Susan at mitorelvis@juno.com

[22Q11.2 The International 22q11.2 Deletion Syndrome Foundation](#)

April 2006 [Practical Implications for Behavior and Learning](#)

[Ability](#) VCFS has a dedicated area. Conference 41. You have to sign up to participate because this site is intended to be a safe haven for children and teens to chat as well.

[VCFS Velo-Cardio-Facial Syndrome Educational Foundation](#)

[Message Board](#) A second message board dedicated to VCFS

[yourhealth.com](#) Some VCFS information but primarily a general health site

[VCFS information out of the UK 22Q11](#)

[Another VCFS site](#) More general information

[Philadelphia newsletter](#) Last entry was 1999, but there is still some good information

[Robert Shprintzen, Ph.D.](#) Contact information

[LINKS](#) to more Links

[Family Village](#) Family village sites always seem to have good information.

[Summary](#) Information.

[Easy to read](#) description of the syndrome etc.

[Presentation of Atlanta conference material.](#) (2004)

Will add this presentation material shortly (2006)

General Agenda

Friday, April 21, 2006

7:30 - 8:30 AM Continental Breakfast

8:30 - 9:30 AM Overview of the 22q11.2 Deletion Syndrome
Donna M McDonald-McGinn, MS, CGC

9:30 - 10:15 AM Symptoms Versus Causes: Putting Genetic Syndromes Into Context in the School Setting
Brenda Finucane, MS, CGC

10:15 - 10:30 AM Break

10:30 - 11:15 AM Cognitive and Educational Profile of Students with the 22q11.2 Deletion Syndrome
Edward M Moss, PhD

11:15 - 12:15 PM Behavioral and Psychiatric Features of 22q11.2 Deletion Syndrome Across the Lifespan
Anne S Bassett, MD

12:15 - 12:30 PM Questions and Answers

12:30 - 1:30 PM Lunch

1:30 - 2:15 PM Psychiatric Profile of Children with the 22q11.2 Deletion Syndrome
Jacob AS Vorstman, MD

2:15 - 2:30 PM Break

2:30 - 4:00 PM Concurrent Breakout Sessions (choose one):

Session A: Mathematics Instruction for Elementary-aged Children with Nonverbal Learning Disabilities
Eileen F Witcher, MEd and Cheri Settanni, MEd

Session B: Social Stories: Effective Tools for Behavior and Education
Barbara Haas-Givler, MEd, BCBA

Session C: Advocating for Children with the 22q11.2 Deletion At School and in the Community
Mark Abissi

General Agenda

Saturday, April 22, 2006

7:30 - 8:30 AM Continental Breakfast

8:30 - 8:45 AM Announcements

8:45 - 9:15 AM A Model for Servicing the Complex Needs of Children with 22q11.2 Deletions

Lisa J Kobrynski, MD

9:15 - 10:00 AM Mental Health Screening in Adolescents With the 22q11.2 Deletion Syndrome

Joseph F Cubells, MD, PhD and Opal Y Ousley, PhD

10:00 - 10:15 AM Break

10:15 - 11:00 AM Family Perspectives on Raising a Child With the 22q11.2 Deletion Syndrome

Karlene Coleman RN, MN, CG

- Introduction Southeast 22q Support Group Parents Panel

11:00 - 11:45 AM Positive Exposure: Redefining Beautiful

Rick Guidotti

**12 - 4 PM: Spring Fling luncheon and entertainment
hosted by the *Southeast 22q Support Group*
All Are Welcome! RSVP to Reserve Lunch.**

Conference Objectives

- ◇ As a result of this conference, participants will be able to:
 - recognize the spectrum of physical, behavioral, and cognitive features seen in people with deletions of chromosome 22q11.2
 - distinguish behavioral diagnoses, like ADHD and autism, from causal diagnoses, such as 22q11.2 deletion syndrome
 - apply educational and behavioral strategies for working with people who have 22q11.2 deletion syndrome
 - access informational and therapeutic resources on 22q11.2 deletion syndrome

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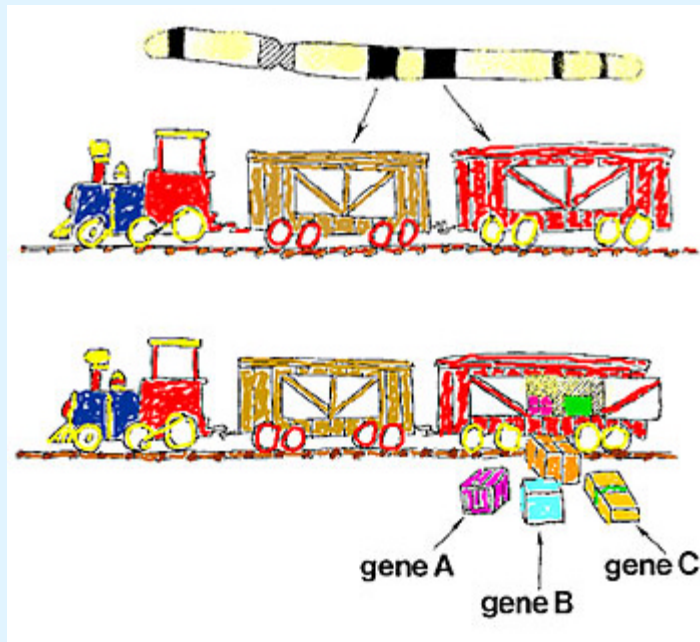
Target Audience

This conference was of interest to parents, special educators, psychologists, genetics professionals, nurses, social workers, and others involved in the care of children and adults with the 22q11.2 deletion syndrome.

The focus was on practical and understandable information.

Children's Hospital Of Philadelphia study of 600 patients

- Males 49% / Females 51%
- 4% have succumbed due to related complications
- Young database, 69% are 10 and younger
- Only 7% of cases are familial
- The size of the deletion does not seem to be influencing clinical findings
- New cases occur equally between sperm and egg
- In familial cases parent of origin does not influence clinical findings



Common Significant Medical Problems

◇ Immunodeficiency	77%
◇ Congenital Heart Disease	76%
◇ Palatal Differences	76%
◇ Hypocalcaemia	49%
◇ Renal Abnormalities	36%
◇ Feeding and Swallowing Problems	35%

Most
normalize by 1
year of age

- ◇ Often resolves in newborn period
- ◇ May reoccur during stress, illness (puberty)
- ◇ Adult incidence is higher now than in previous studies
- ◇ Should be periodically monitored

Autoimmune Disease as an Emerging Issue

	22Q11	General Population
◇ Idiopathic Thrombocytopenia	6/600	1/5000
◇ Juvenile Rheumatoid Arthritis	5/600	1/1000
◇ Neutropenia	1/600	1/1,000,000
◇ Graves Disease	1/600	1/25,000

A Little More Detail

- ◇ Prevalence
 - ◇ 2003 data
 - ◇ 1.7 per 10,000 births
- ◇ Speech and language Issues
 - ◇ 96% of people with VCFS are language delayed
- ◇ Psychiatry, Behavior and Neuroscience
 - ◇ Schizophrenia is prevalent in this population (23% of VCFS population vs. 1.3% of general population) Canadian study
 - ◇ seek mental health advice early
- ◇ Psychiatric disturbance in adults with VCFS
 - ◇ develop in late teens or older
 - ◇ Ionized calcium levels are a clue
 - ◇ Manage your child's stress levels to delay or reduce the on set of psychiatric issues

Treatable Psychiatric Illnesses (newest data based on less biased sample)

	22Q11	General Population
◇ None	40%	
◇ Major Depression	17%	5.8%
◇ Anxiety Disorder	13%	4.1%
◇ Attention Deficit Disorder	10%	-----
◇ Substance Use Disorder 17%	16%	
◇ Schizophrenia	23%	1.3%

Per: Dr. Ann Bassett et al

Neuropsychological Issues in Young Children

- ◇ Motor milestone delays
- ◇ Emergence of language delays
- ◇ Features of Frank Autism

All Benefit From Early Intervention Strategies

Watch for:

- ◇ Withdrawal
- ◇ Depression
- ◇ Falling Grades
- ◇ Loss of interest in usual activities

Nonverbal Learning Disorders

Strengths

Verbal IQ
Rote Verbal Learning
Word Reading (phonics)
Immediate Auditory Attention
Simple Focused Attention

Challenges

Visual Perception
Visual Motor Integration
Mathematics
Complex Verbal Memory (stories)
Fine and Gross Motor
Visual Memory

What Does It Mean

Will be talkative but will not necessarily convey a coherent message

Insensitive to nonverbal cues

Conformity to rules but is rigid in novel situations

Poor understanding of non-literal language

Absence of spontaneous and implicit empathy

Social approaches based on explicit, verbal, one-sided communication about narrow interests

Limited insight

Practical Issues For Parents

- ◇ You can make a significant positive difference
 - Knowledge is power
 - Balance hard work toward goals with realistic expectations
- ◇ Take care of yourself and your spouse
 - Put on your own oxygen mask first
- ◇ Take behavioral symptoms seriously
- ◇ The squeaky wheel gets the grease

Practical Issues For People with 22Q11

- ◇ Speak up if something is bothering you, seek help from:
 - Family and trusted loved ones
 - Your primary care doctor(s)
 - Seek specialty care as required

- ◇ Set realistic but challenging goals

- ◇ Follow your treatment plan
 - Take your medications
 - Discuss effects with your doctor
 - Attend your appointments

Summary of School Setting

- ◇ Educational and behavioral diagnoses, not genetic Diagnoses determine eligibility and services in school settings
- ◇ Individuals with 22Q11 deletion often meet the criteria
- ◇ Use these diagnoses for everything they're worth
- ◇ Educators are unlikely to be familiar with 22Q11 but they are likely to be able to provide excellent service

From a Parent's Perspective: Coordinate Your Child's Care

- ◇ Be proactive - "you are the professional parent"

Have an end (goal) in mind and work backwards

- ◇ Recognize that this goal will change over time

IEP (Individual Educational Plan)

- ◇ An IEP meeting can include:
 - Parents / family
 - Resource teacher
 - Regular classroom teacher
 - Principal
 - Speech, Occupational, Physical therapist(s)
 - Member(s) of the district school board

- ◇ Keys to a successful meeting:
 - Stay focused
 - Know your rights, and stay current on those rights
 - Know what you want
 - Don't make unreasonable requests
 - Do not become emotionally charged
 - Do not commit to anything in the meeting

- ◇ Remember that IEPs may be reopened

- ◇ Document all meetings including attendance

- ◇ Educate the audience about the key points surrounding 22Q11 and education

Classroom Strategies

- ◇ Well defined schedule, consistent rules and routines
- ◇ Clear presentation of assignments
 - Single step precise instructions
 - Verbal or verbal reinforcement
 - Extra processing time, pause during instructions
- ◇ Eliminate distractions
- ◇ Preferential seating
- ◇ Explicit identification of assignment goals and sub goals
- ◇ Frequent monitoring of work pace and accuracy
- ◇ Immediate performance feedback

Beware of IQ Testing Scores

>12 point split between verbal and performance
IQ observed in 65% of 22Q11 children tested

Sample (real) data:

Full Scale score = 87

but

Verbal IQ = 111

Performance IQ = 65

Proper analysis of the data can lead to better educational strategies

Extra Curricular Activities

(as gathered from various families' experiences)

More Successful

Therapeutic Horseback Riding
Swimming
Golf
Tennis
Piano
Voice

Challenging

Team Sports
Large Group Settings
Noisy Activities

Outcomes

29 of 42 people graduated high school

Occupations of adults include:

- ◇ Chef
- ◇ Farmer
- ◇ Security guard
- ◇ Maintenance workers
- ◇ Office workers
- ◇ Delivery persons
- ◇ Homemakers

3 Young Adults, identified during adolescence, are currently being tracked:

- ◇ All are enrolled in college
- ◇ 2 of 3 passed the "standard" college entrance exam
- ◇ 1 just graduated with a 3.0 GPA

Adults in the 22Q11 population tend to:

- ◇ Have difficulty understanding the ramifications of the diagnosis and compliance with their treatment recommendations
- ◇ Have a poor understanding of the recurrence risks
- ◇ Generally require a social safety net in the form of an unaffected relative and/or social worker

Other Areas Of Discussion

Strategies for teaching Mathematics

Advocating in an Educational Setting

Social Stories



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Great On Line Discussion Group

To join, visit the site listed below and subscribe:

<http://listserv.icors.org/SCRIPTS/WA-ICORS.EXE?SUBED1=vdfs&A=1>

Our Personal Highlights / Notes

- ◇ Opportunity to interact with highly knowledgeable parents and professionals one on one
 - ☆ Knowledgeable parents
 - ☆ Medical doctors with specific areas of expertise
 - ☆ Psychiatric specialists
- ◇ Personal accounts of fairly high degrees of independence
- ◇ Advocate, advocate, advocate (educational systems)

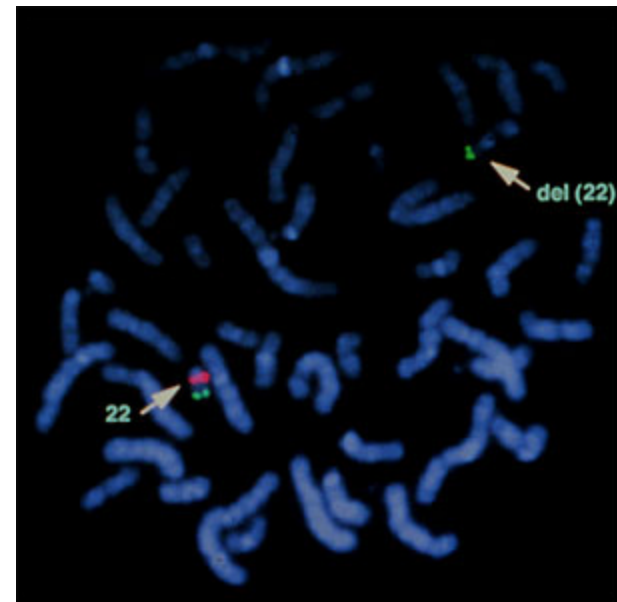
All in all a very educational and helpful experience. We highly recommend to any of you who can, to attend....

Annual International Meetings of The Velo- Cardio-Facial Syndrome Education Foundation to be held in:

VCFSEF International Scientific
Conferences
for 2006:

Strasbourg France
July 7 – 9

Brisbane, Australia
Nov 2 – 4



Planning for a 2007 Meeting in Texas