Chromosome 22q11.2 Deletion Syndrome: Integrating Clinical and Research Experience

Society for Developmental Behavioral Pediatrics

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Disclosures

Tony J. Simon, Robin Hansen, Kathy Angkustsiri, Scott Akins, Ingrid Leckliter, Janice Enriquez, and Joel Stoddard have nothing to disclose.
Outline

Introductions and Overview
– Tony J. Simon, Ph.D. and Robin Hansen, M.D.

Medical Care and Genetic Mechanisms
– Kathy Angkustsiri, M.D. and Scott Akins, D.O.

An Explanatory Account of (Some) Cognitive Impairments
– Tony J. Simon, Ph.D.

Neuropsychological Assessment
– Ingrid Leckliter, Ph.D. and Janice Enriquez, Ph.D.
Outline

Psychiatric Comorbidities
  – Joel Stoddard, M.D.

Translational Clinic at the MIND Institute
  – All

Discussion/Questions
The Naming Issue

Far too many names, causing confusion, reducing identification

Growing preference: 22q11.2DS (chromosome 22q11.2 deletion syndrome)

DiGeorge syndrome (DiGeorge 1965)
Velocardiofacial syndrome (Shprintzen 1978)
Conotruncal anomaly face syndrome (Kinouchi)
Opitz G/BBB syndrome (Opitz 1972)
Cayler Cardiofacial syndrome (Cayler 1969)

From Donna McDonald-McGinn

90 signs & symptoms identified so why these three?

Oral: 69 - 100%
Ocular: 49 - 83%
Cardiovascular: 100%
Introduction

Cognitive Analysis and Brain Imaging Laboratory (ABIL)

Funded by:

• National Institutes of Health
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  – Clinical and Translational Science Center UL1 RR024146

Formation of the translational clinic

Very brief history, more later
Objectives

- Increase identification of children with the disorder
- Reduce some of the naming confusion
- Describe the associated medical symptoms
- Review guidelines for medical work-up & surveillance
- Increase understanding cognitive/behavioral profile
- Provide recommendations for educational resources and intervention
- Understand the mental health difficulties and potential treatments for children with 22q11.2DS
A Huge “Thank You”

...all of the children who took part in our studies and their families, and the funding sources listed earlier...

generous private donors who keep us going...

Dan Comins, “video producer extraordinaire”...

...meet some of the most important people!