PRACTICAL GUIDELINES FOR MANAGING PATIENTS WITH 22Q11.2 DELETION SYNDROME

Thu, 4/23: 2:30 PM - 4:00 PM

Room: Celebrity B

Abstract No: 60

Format: Workshop

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Background/Purpose:

Chromosome 22q11.2 deletions have been identified in the majority of patients with DiGeorge syndrome, velocardiofacial syndrome and conotruncal anomaly face syndrome; and in a subset of patients with Opitz G/BBB syndrome and Cayler Cardiofacial syndrome. Although clinically under-recognized, the 22q11.2 deletion syndrome (22q11.2DS) is the most common microdeletion with an estimated prevalence of 1 in 1000 – 1 in 4000 live births. Furthermore, 22q11.2DS is the second most common cause of developmental delay and congenital heart disease after Down syndrome, and importantly, it is the most common cause of syndromic palatal anomalies.

Methods/Description:

Despite these facts and figures, broad recognition of the condition and systematic anticipatory guidance for clinical management remains limited. That said, the International 22q11.2 Deletion Syndrome Consortium established practical guidelines, developed in multiple stages, including three international consensus meetings where participants with broad expertise including 18 subspecialties representing >15 countries determined best practice based on experiences, data and review of 239 relevant publications with a goal of transcending nationalities, health care systems, and subspecialty biases. These recommendations were synthesized by the lead authors of this Study Session; published in the Journal of Pediatrics in 2011; and are currently being followed routinely by the multidisciplinary panel members. Therefore, their comprehensive experiences will be shared with the audience during this interactive workshop. Specific topics to be discussed will include: assessment and treatment of palatal abnormalities; speech and language challenges; intellectual and behavioral deficits; genetic counseling and psychosocial concerns; and a coordinated approach to medical and surgical care including important considerations prior to scheduling operative procedures and across the perioperative period. Finally, as these recommendations appear applicable to patients presenting to the Cleft Palate Clinic with atypical nested 22q11.2 deletions, as well as, 22q11.2 duplications, these conditions will also be defined in the setting of this collaborative workshop.

Primary Subject Area:

Genetics

Objectives/Additional Information

Please list the MAIN LEARNING OBJECTIVES of the presentation. SAMPLE OBJECTIVE: "Each learner will be able to identify and write a definition for the following terms: self-directed learning, certification, and CEUs." (For additional information about Learner Objectives, please refer to the American Nurses Association Guidelines For Writing Instructional Objectives appended to the Abstract Submission Guidelines - you will need to return to the previous page to access the Guidelines).

Attendees will recognize the complexities of the 22q11.2DS; understand the concept of nested deletions and duplications; appreciate the utility of healthcare surveillance and coordinated multidisciplinary care in particular for palatal anomalies, speech/language, developmental and behavioral issues.

Has this abstract been previously presented?

Yes
If yes, when and where?
ACPA 2014 and 2012

If this abstract is being submitted in the Junior Investigator category, for the presenting author to qualify as a Junior Investigator the following statement must be true…

This abstract is not in the Junior Investigator category.

Financial / Non-Financial Disclosure

Other (including honoraria)

I have been a speaker for Natera.