Pre-anesthetic Physical Examination Leading To A New Diagnosis of Velocardiofacial Syndrome

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Introduction

Our Patient:
4 year old male child

Diagnosis:
Ingested coin, 2 hours earlier

Procedure:
Removal of foreign body.

PMH:
• Mild cognitive delay
• URIs
• "Heart Problem," reportedly resolved

Medications / Surgeries / Allergies:
None

Physical Examination

Pertinent Positives:

HEENT: Syndromic face, poor dentition.

Heart: Grade 2/6 systolic murmur at right upper sternal border. Regular rhythm, Normal rate.

Lungs, MSK, Skin, Psych:
Normal
Intra-operative & Post-operative Care

Intra-operative Care
- Uneventful general anesthetic
- Successful retrieval of coin

Post-operative Care
- Echocardiogram post-op
- 3 large ASDs (unrestricted flow)
- Severely dilated RA & RV
- Persistent left superior vena cava with coronary sinus connection
- Normal function of chambers and valves
- Velocardiofacial Syndrome suspected
- Cardiology & Genetics Consulted

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Diagnosis: 1,2
Previously: Clinical constellation
Now: Genetic (FISH) testing

Pathogenesis: 1,2
Microdeletion of Chromosome 22 at the q11.2 band
- TBX1, PRODH, COMT
- > 40 genes effected

References:

Human 22q11.2 Genes
- Clinical
  - males (75%)
  - females
- Cardiac anomalies / delay
- Otic abnormalities
- Syndrome (10%)
- Dna (25x)
- General
- Chr. 22
- 10 features
- Trisome expression

Prevalence: 1:2000 in the US

Our Patient:
- Cardiology: Repair of ASDs necessary
- Dental disease must be addressed first
- Anticipate recurrent URTIs
- Genetic counseling for definitive diagnosis
- Need for follow-up emphasized to family
- Social and geographic difficulties in obtaining follow-up care

General Guidelines:
- Tailor care to the specific constellation of symptoms1

References:

Treatment and Care
Preanesthetic evaluation is essential to appropriate perioperative care. It should be:

- Thorough
- Attentive
- Address new or unexpected findings
- Ensure follow-up of new diagnoses

The anesthesiologist may be the only medical personnel to thoroughly evaluate the patient during their hospitalization.

- Every encounter is a new opportunity to improve a patient's care

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