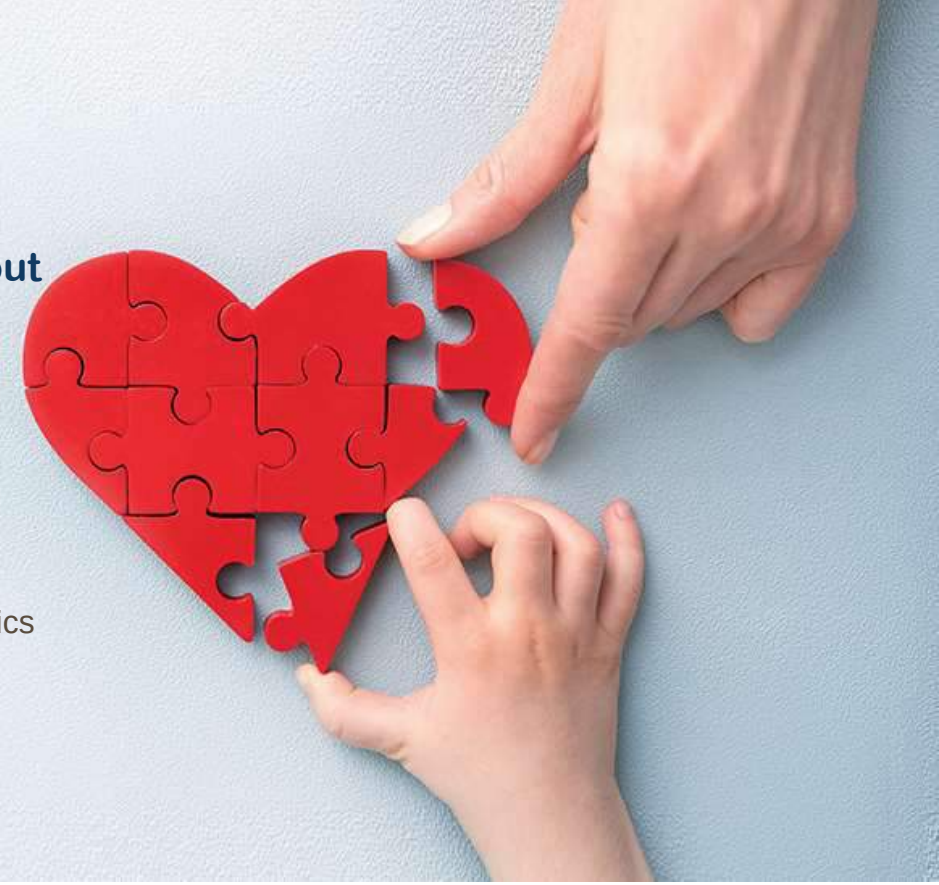
CARDIOLOGY 2024

What the Anesthesiologist Needs to Know about Genetic Syndromes Before Putting a Cardiac Patient to Sleep?



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Cardiac Center

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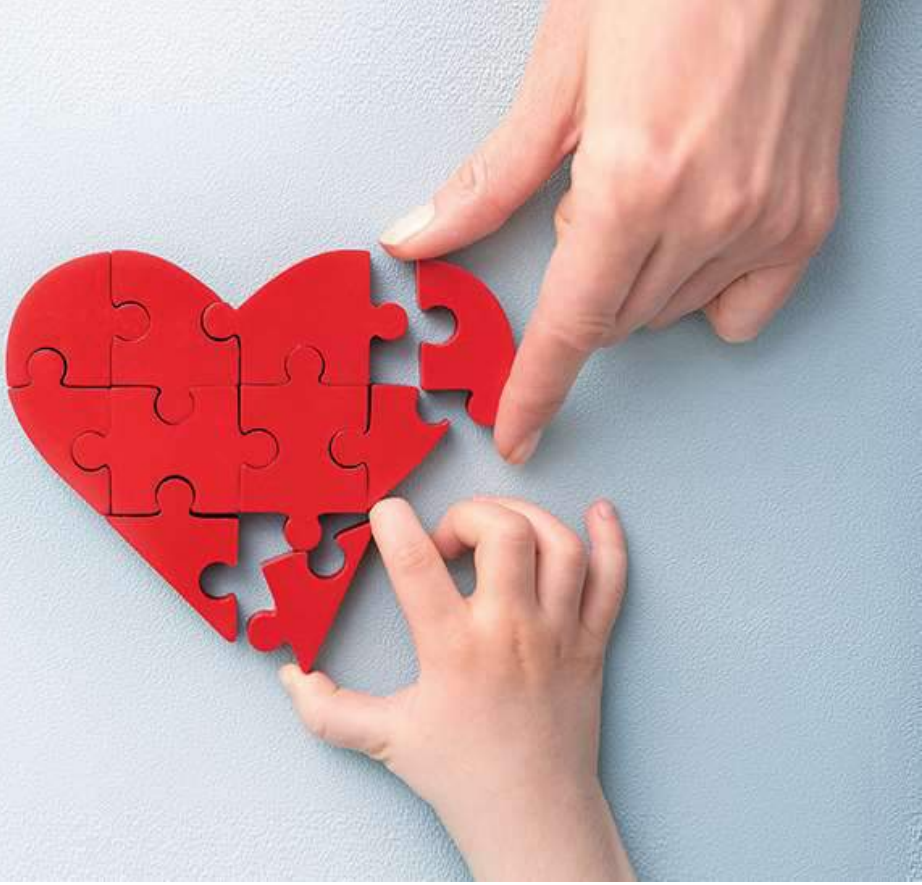
NO DISCLOSURES



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OBJECTIVES

- To review some of the common syndrome/genetic diseases encountered by the cardiac anesthesiologist
- To review preop anesthetic consideration for a child with heart disease and commonly occurring syndromes and genetic disorders
- To review some of the airway considerations/approaches for genetic syndromes

OUTLINE

Syndromes

- Trisomy 21
- Noonan
- 22q11
- Williams
- Turner

Connective Tissue Disorders

- Marfans
- Loeys-Dietz

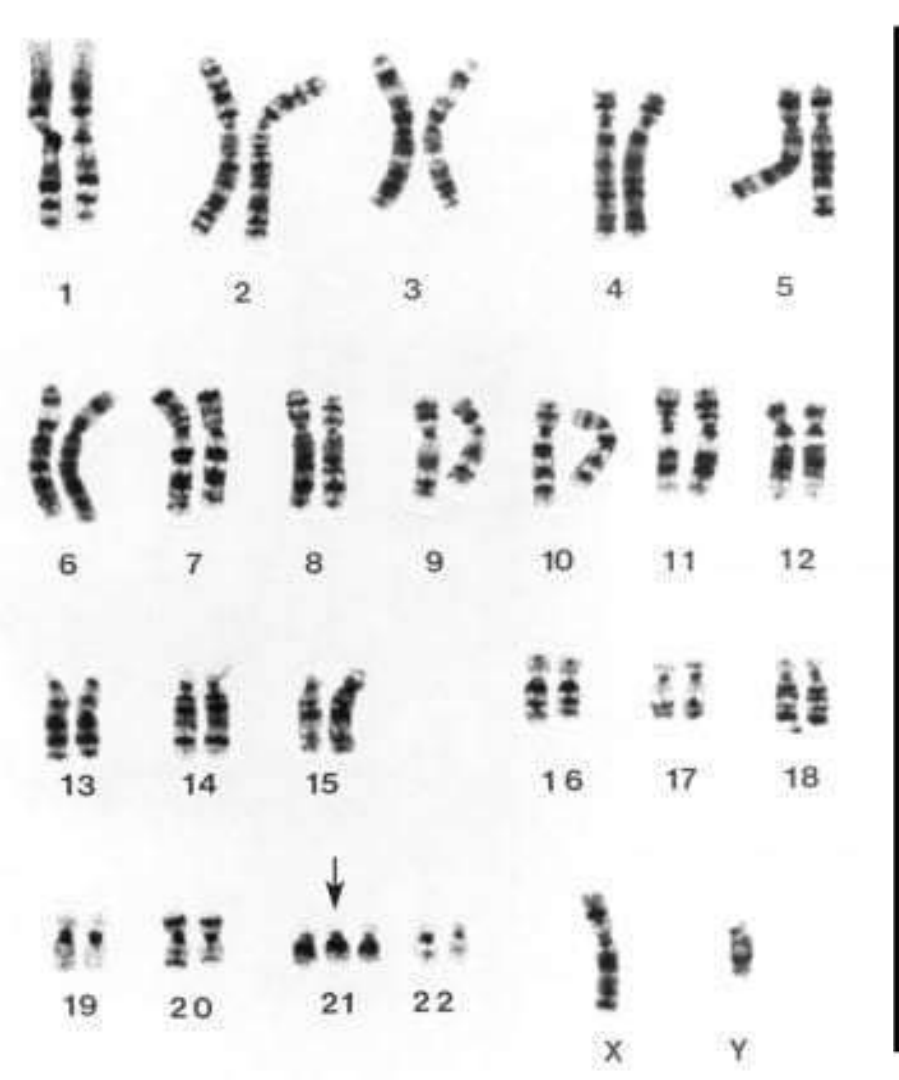
Channelopathies

- Long QT syndrome
- CPVT

SYNDROMES AND CHD

Genetic syndrome	Genetic abnormality	CHD %
Down syndrome	Trisomy 21	35–50%
Noonan syndrome	Mutations in genes that affect the RAS-MAPK pathway	81–90%
DiGeorge and velocardiofacial syndrome	22q11 deletion	75–83%
Turner syndrome (45,X)	XO	17 to 50%
Williams-Beuren syndrome	7q11.23 deletion	80%
Holt-Oram syndrome	Mutations in the TBX5 gene	79%

Duarte VE, Singh MNGenetic syndromes associated with congenital heart diseaseHeart Published Online First: 01 December 2023. doi: 10.1136/heartjnl-2023-323126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I'm The
HOMIE
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EXTRA
CHROMIE

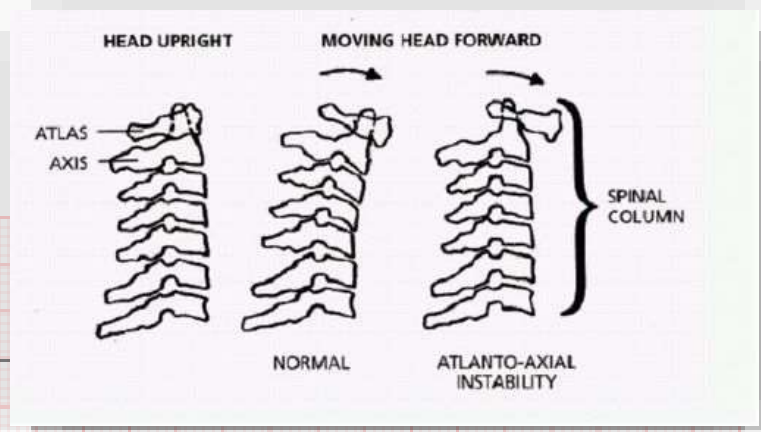
TRISOMY 21

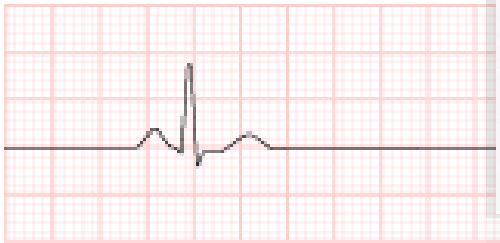
CARDIAC

- Specific to underlying CHD
- **Bradycardia** with induction
- Persistent pulmonary HTN

RESP

- Large tongue, pharyngeal hypotonia
- Undiagnosed OSA
- Subglottic stenosis
- Airway malacia
- **Possible difficult airway**
- Neck instability!!





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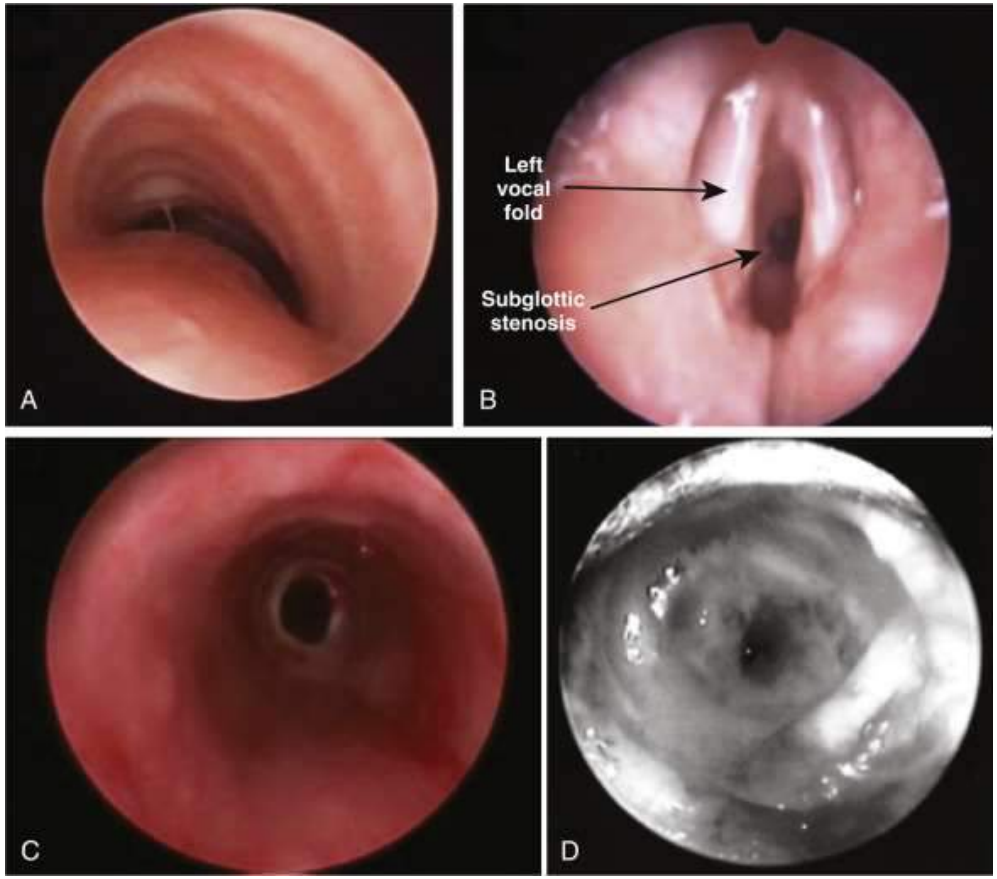
https://www.youtube.com/watch?v=oHPcuuKW984&t=90s

TRISOMY 21 & AIRWAY CONCERNS



From Hamilton J, Yaneza MM, Clement WA, et al. The prevalence of airway problems in children with Down's syndrome. Int J Pediatr Otorhinolaryngol. 2016;81:1-4.

Shapiro NL, Huang RY, Sangwan S, Willner A, Laks H. Tracheal stenosis and congenital heart disease in patients with Down syndrome: diagnostic approach and surgical options. Int J Pediatr Otorhinolaryngol. 2000;54(2-3):137-142

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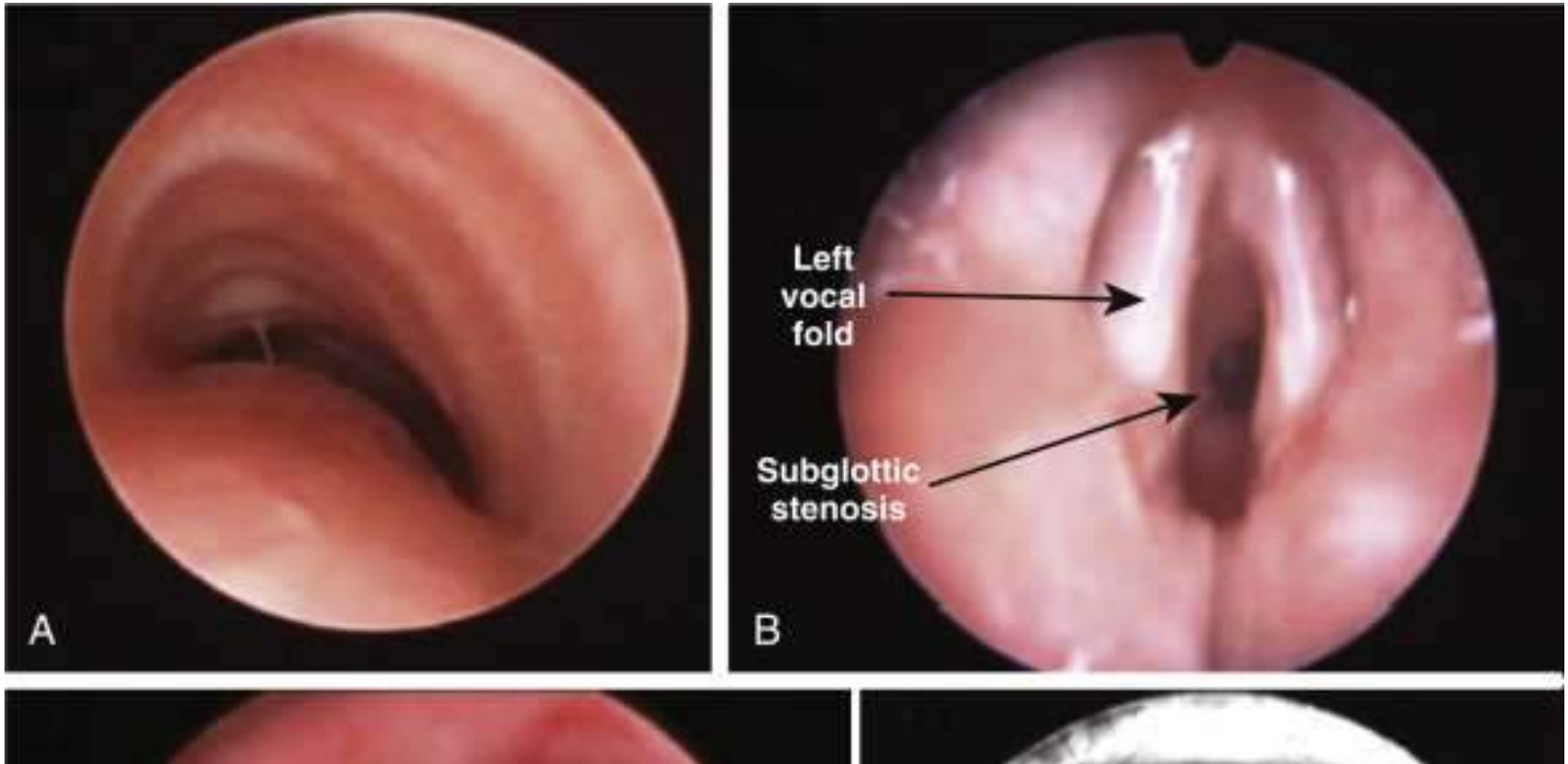

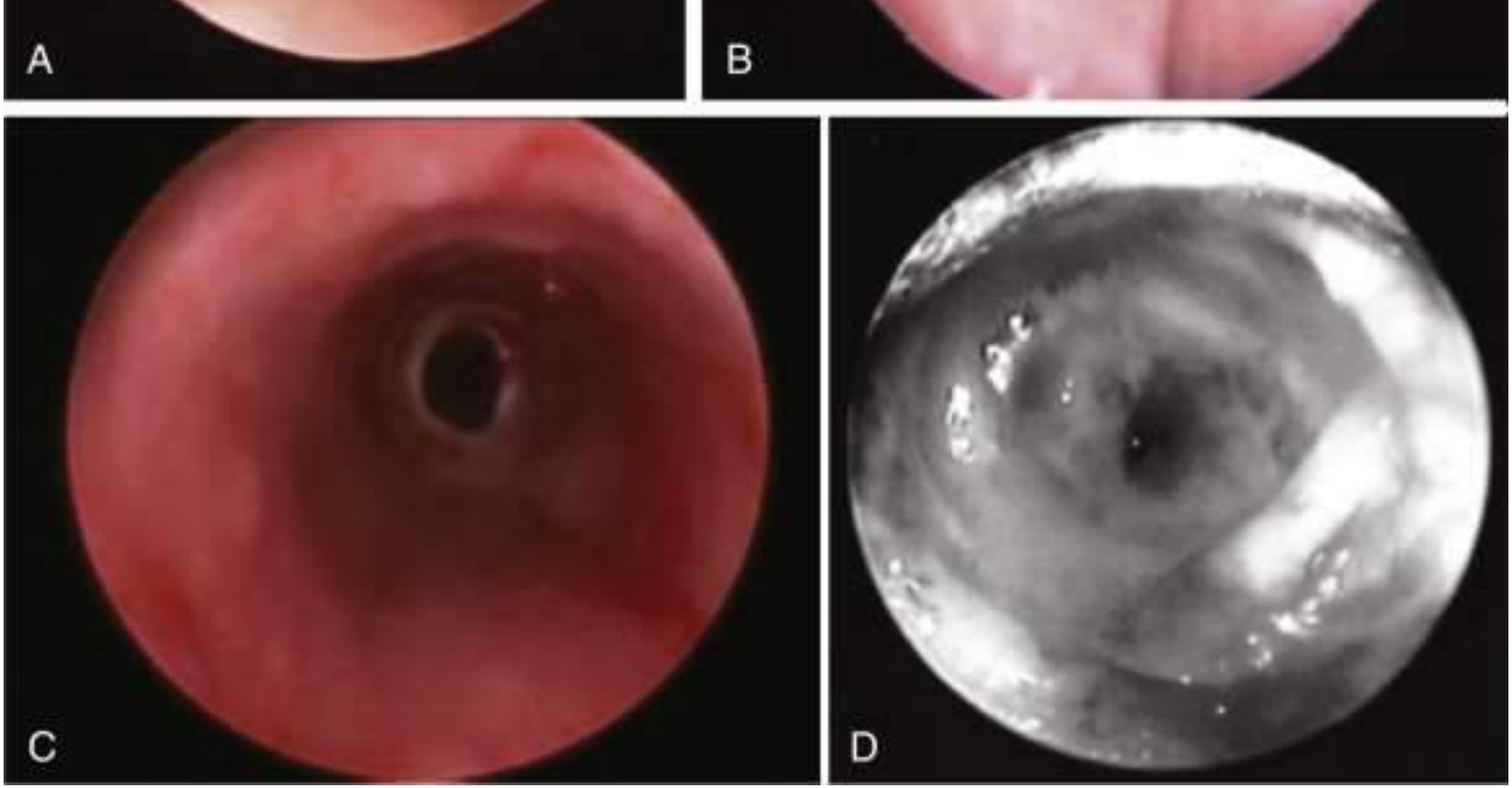

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TRISOMY 21

CARDIAC	RESP	GI/RENAL	NEURO	ENDO	SKELETAL	OTHER
• Specific to underlying CHD • **Bradycardia** with induction • Persistent pulmonary HTN	• Large tongue, pharyngeal hypotonia • Undiagnosed OSA • Subglottic stenosis • Airway malacia • **Possible difficult airway** • Neck instability!!	• Hirschsprung disease • Imperforate anus • Duodenal atresia • Growth delay • Obesity	• Developmental delay	• Hypothyroidism	• C1-C2 instability→ challenging airway	• Hematologic malignancies

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https://www.youtube.com/watch?v=oHPcuuKW984&t=90s

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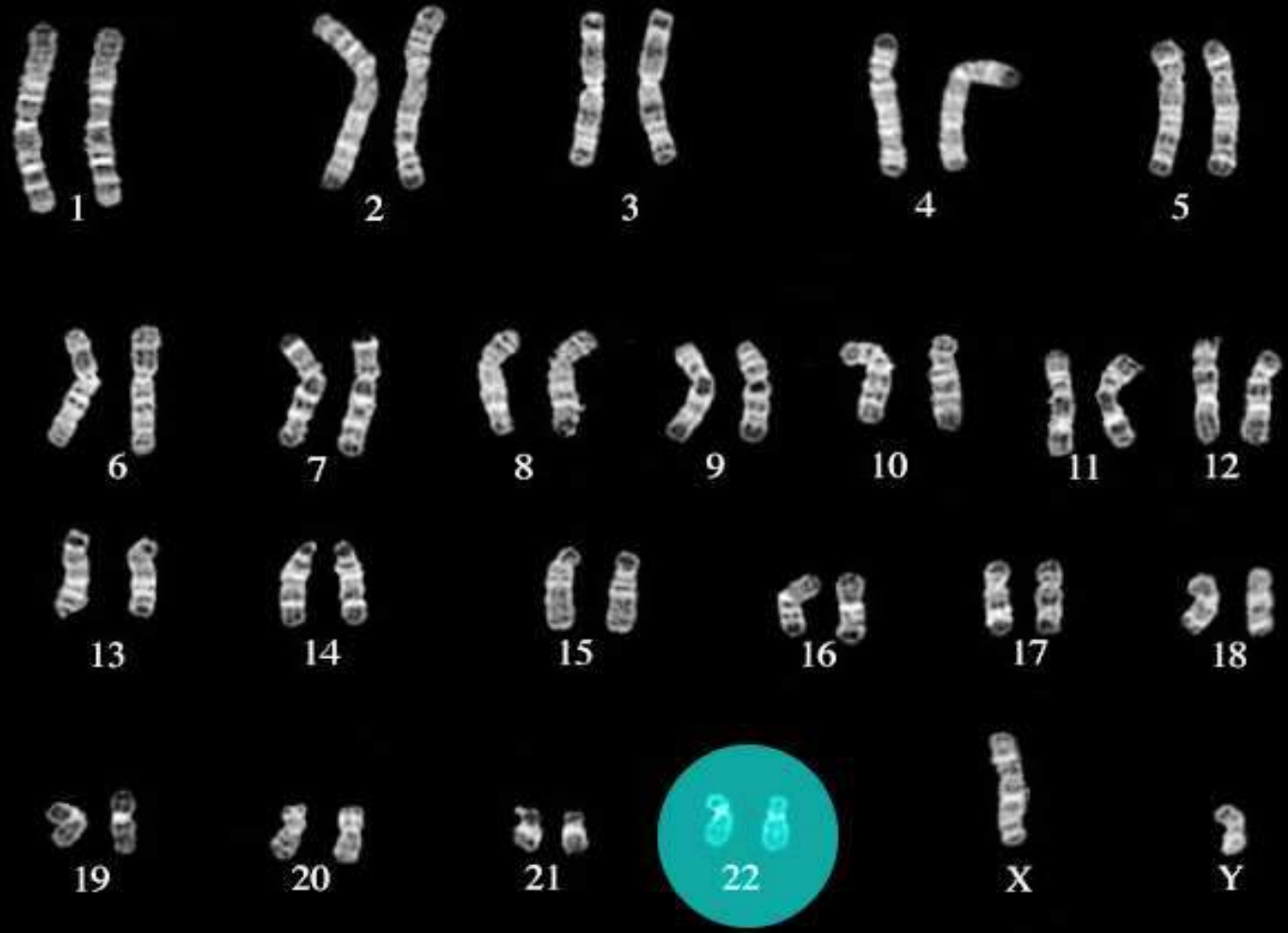
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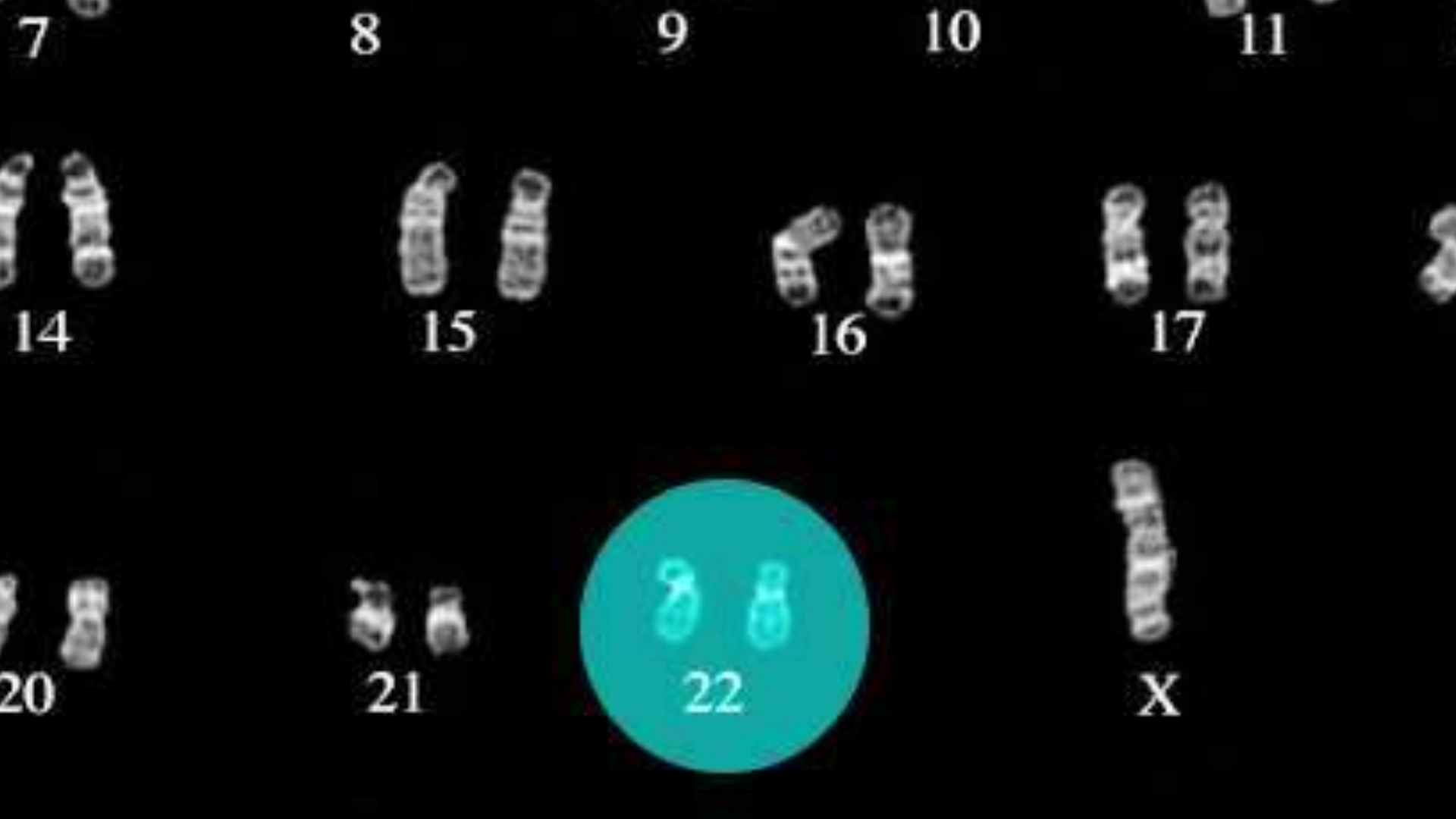
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22q11 deletion

CARDIAC	RESP
• Specific to underlying CHD • Conotruncal anomalies	• 11-17% association with Pierre Robin sequence→ **Possible difficult airway** • Micrognathia • Small mouth opening

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Children's Hospital of Philadelphia Cardiac Center

http://www.midwestsinus.com/todd-files/systemic-diseases/pierre-robin-sequence/

COMBINED TECHNIQUE: ANGULATED VIDEOSCOPE AND FIBEROPTIC INTUBATION

Children's Hospital of Philadelphia

Combined Technique

Scan the QR code to learn more about the Combined Technique

CHOP ACCM difficult airway program (AirEquip)

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22q11 deletion

CARDIAC	RESP	GI/RENAL	NEURO	ENDO	HEME	OTHER
• Specific to underlying CHD	• 11-17% association with Pierre Robin sequence→ **Possible difficult airway** • Micrognathia • Small mouth opening	• Association with TEF	• Developmental delay	• Hypocalcemia	• Thymic hypoplasia→ Irradiation of blood products	

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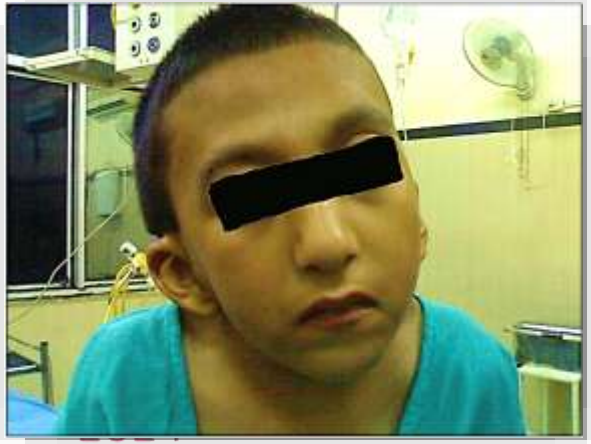
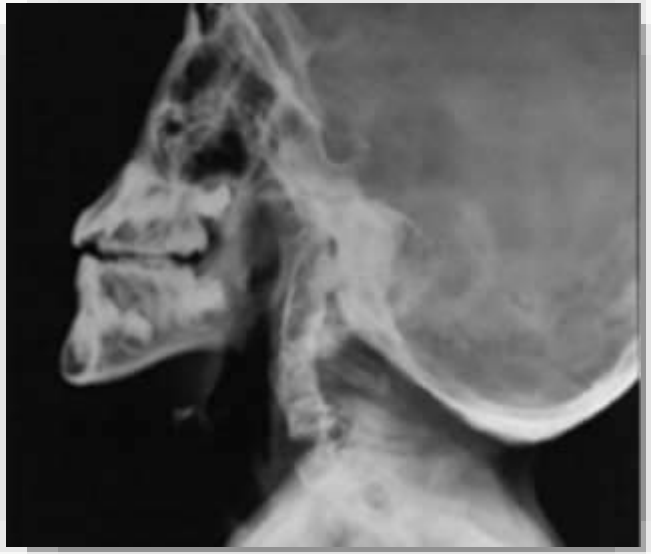
http://www.midwestsinus.com/todd-files/systemic-diseases/pierre-robin-sequence/

NOONAN SYNDROME

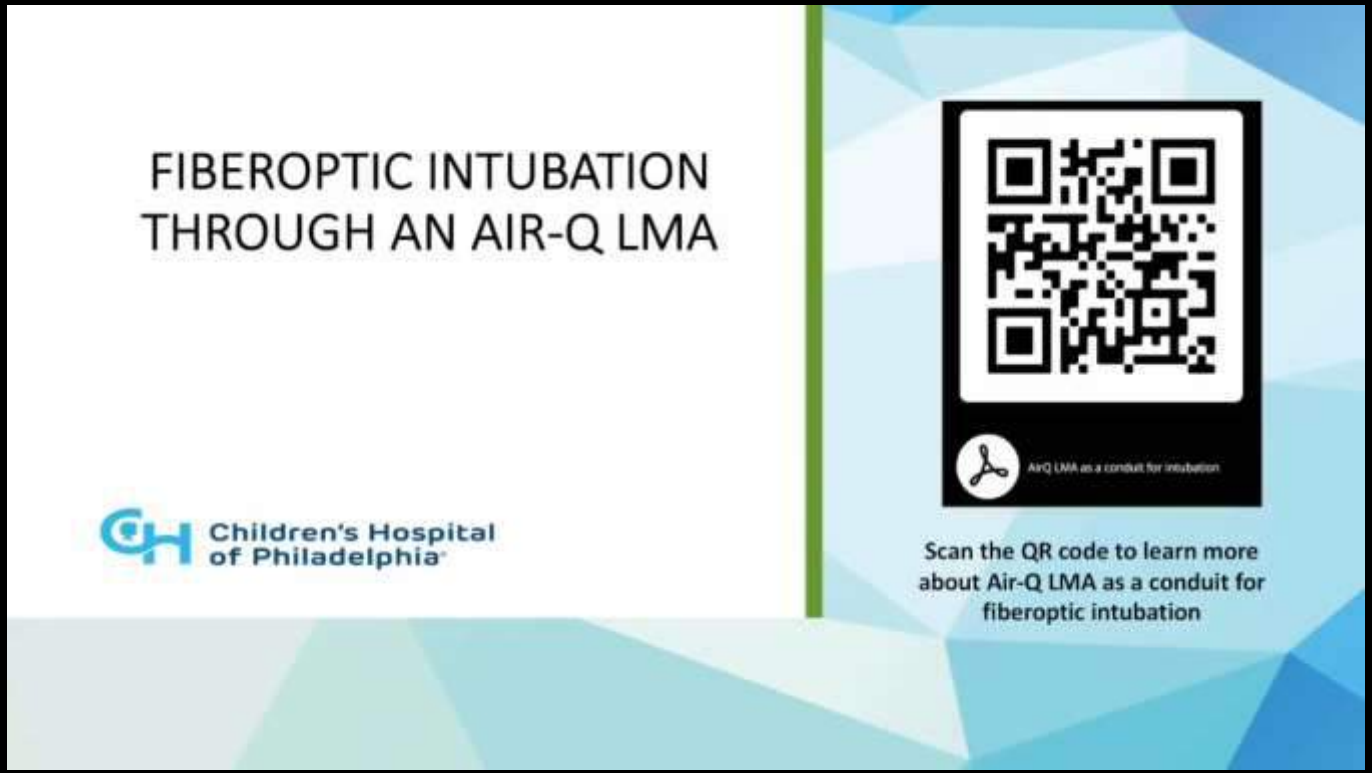
CARDIAC	RESP	GI/RENAL	NEURO	HEME	SKELETAL	OTHER
• Specific to underlying CHD • **PS, HCM, ASD, partial AV canal**	• Webbed neck • Short neck • Micrognathia • High arched palate • **Possible difficult airway!!**	• Renal anomalies		• Bleeding diathesis • Platelet dysfunction	• Spinal anomalies • Vertebral/rib malformation • Short stature • **Cervical spine fusion**	





Bajwa SJ, Gupta S, Kaur J, Panda A, Bajwa SK, Singh A, Parmar SS, Prasad S. Anesthetic considerations and difficult airway management in a case of Noonan syndrome. Saudi J Anaesth. 2011 Jul;5(3):345-7. doi: 10.4103/1658-354X.84121. PMID: 21957424

TRANS-LMA FIBEROPTIC INTUBATION



WILLIAMS SYNDROME

CARDIAC	RESP	GI/RENAL	NEURO	HEME	SKELETAL	OTHER

- Supravalve AS/PS
- Diffuse branch PS
- QTc prolongation
- **Coronary ostial stenosis/insufficiency→ ARREST at induction!**

- Midfacial flattening
- Mandibular hypoplasia
- Dental malocclusion,

- Pre-op nutritional status
- Hypercalcemia?
- Hypercalcuria?
- Renal stones?

Low risk	Moderate risk	High risk
Normal ECG	Mild stenosis of a branch of the pulmonary artery	Severe SVAS (>40 mmHg)
Normal Echocardiogram	Hypertension	Symptoms or ECG signs consistent with ischemia
Minimal cardiac anomalies	Hypoplastic SVAS (<40 mmHg)	Coronary disease demonstrated in imaging
	Other mild cardiac anomalies (e.g., ventricular septal defect)	Severe left ventricular hypertrophy
	Repaired SVAS or SVPS without residual gradients	Biventicular outflow tract disease
	Mild left venticular hypertrophy	Prolonged QTc on ECG
	Mild to moderate SVPS in isolation	
	Significant extracardiac disease such as difficult airway or severe gastroesophageal reflux	

STRATEGIES TO LOWER THE RISK OF CARDIAC ARREST WITH INDUCTION

- Pre-op hydration (PO and IV if present)
- Maintaining SVR during induction
- Avoid excessive tachycardia
- Exclusive IV induction
- Avoid inhalational induction

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Schmidt AR, Collins RT 2nd, Adusumelli Y, Ramamoorthy C, Anesthesia Considerations and Use of Modified Anesthesia Management for Pediatric Patients With Williams Syndrome. J Cardiothorac Vasc Anesth. 2021 Dec;35(12):3667-3674. doi: 10.1053/j.jvca.2021.04.019. Epub 2021 Apr 20. PMID: 34049787.

Matisoff AJ, Olivieri L, Schwartz JM, Deutsch N. Risk assessment and anesthetic management of patients with Williams syndrome: a comprehensive review. Paediatr Anaesth. 2015 Dec;25(12):1207-15. doi: 10.1111/pan.12775. Epub 2015 Oct 12. PMID: 26456018

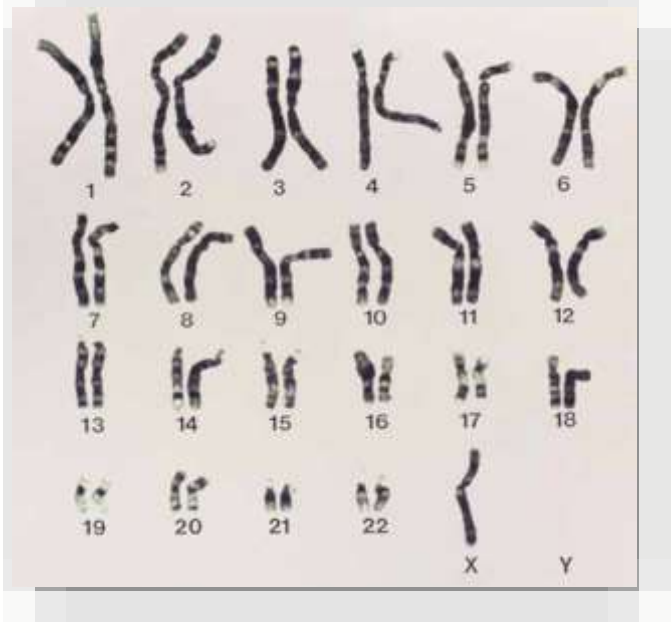
TURNERS SYNDROME

CARDIAC	RESP	GI/RENAL	NEURO	ENDO	SKELETAL	OTHER

- Aortic coarctation is the most common lesion
- 17%-45% can have cardiac involvement

- High arched palate
- Micrognathia
- Short/webbed neck→ limited mobility

- Structural renal malformations up to 40%



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MARFAN SYNDROME

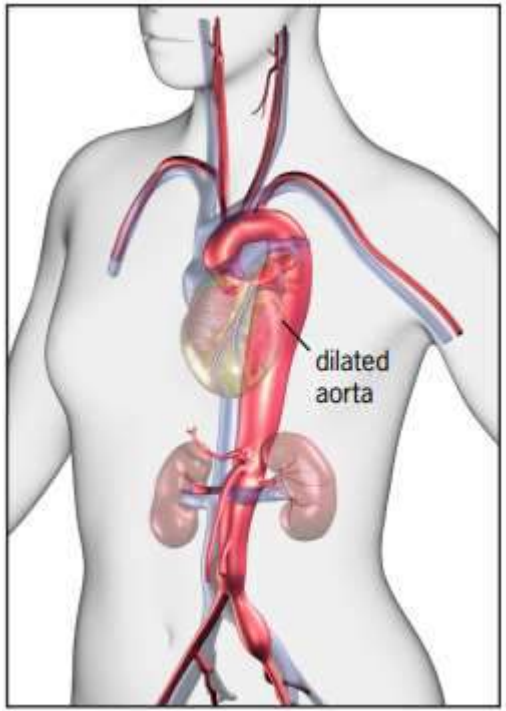
CARDIAC	RESP	GI/RENAL	NEURO	HEME	SKELETAL	OTHER

- Aortic dilation
- **Mitral valve involvement**

- Chest deformity→ Restrictive lung disease
- High arched palate
- TMJ laxity→ dislocation with DL
- C1/C2 ligament laxity

- **USE Indirect laryngoscope or fiberoptic technique to limit neck movement**

- Dural ectasia might be present→ cautious with neuraxial techniques



https://my.clevelandclinic.org/departments/heart/patient-education/-/scassets/23bd097f89a9400081e4543226aef4d2.ashx

LOEYS-DIETZ SYNDROME

CARDIAC	RESP	GI/RENAL	NEURO	HEME	SKELETAL	OTHER
• Earlier dissection compared to Marfan syndrome	• Cervical spine instability • Indirect laryngoscopy or Fiberoptic as primary modality for airway placement • Asthma more prevalent compared to Marfan syndrome	• GERD more common compared to Marfan syndrome		• Easy bruising		

Berger JA, Huh DD, Lee T, Wadia RS, Bembea MM, Goswami DK. Perioperative management and considerations in pediatric patients with connective tissue disorders undergoing cardiac surgery. Paediatr Anaesth. 2021 Jul;31(7):820-826. doi: 10.1111/pan.14196. Epub 2021 May 13. PMID: 33884693.

OUTLINE

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LONG QTC SYNDROME

Journal of Cardiothoracic and Vascular Anesthesia 33 (2019) 2030–2038

Contents lists available at ScienceDirect

Journal of Cardiothoracic and Vascular Anesthesia

journal homepage: www.jcvaonline.com

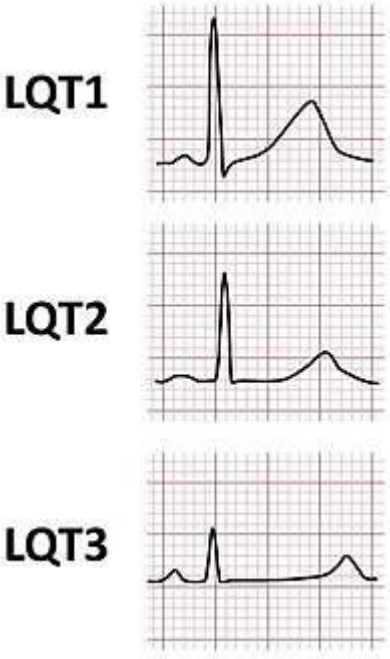
Review Article

Anesthetic Considerations for Pediatric Patients With Congenital Long QT Syndrome

Genevieve E. Staudt, MD*,†, Scott C. Watkins, MD†

*Department of Anesthesiology, Vanderbilt University School of Medicine, Monroe Carell Jr. Children's Hospital at Vanderbilt, Nashville, TN

†Department of Anesthesiology and Critical Care Medicine, Johns Hopkins University School of Medicine, Johns Hopkins All Children's Hospital, St. Petersburg, FL



LONG QT SYNDROME

- Identify patient's LQTS genotype
- Continue maintenance antiarrhythmics on the day of surgery.
- Interrogate PPM/ICD and have a plan for perioperative ICD management
- Establish a calm and quiet environment for the patient.
- Avoid QT-prolonging drugs.
- Ensure adequate depth of anesthesia prior to noxious stimuli
- Plan for an extended period of observation and consider overnight admission.
- Aggressively treat PONV and postoperative pain.
- Consider telemetry monitoring postoperatively

CPVT

British Journal of Anaesthesia 108 (5): 730–44 (2012)
doi:10.1093/bja/aes105

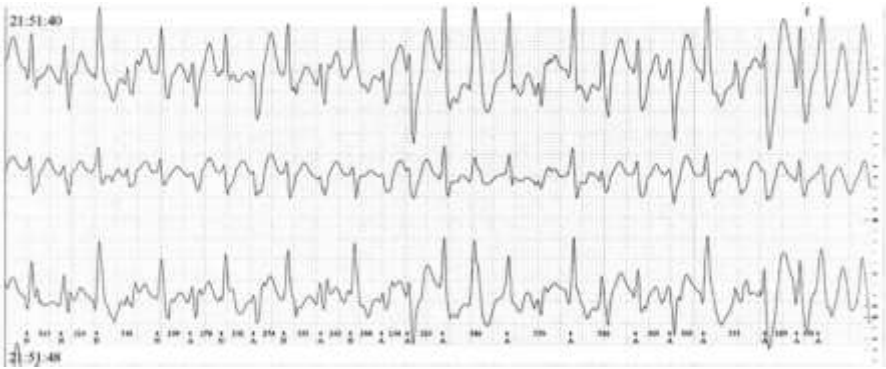
BJA

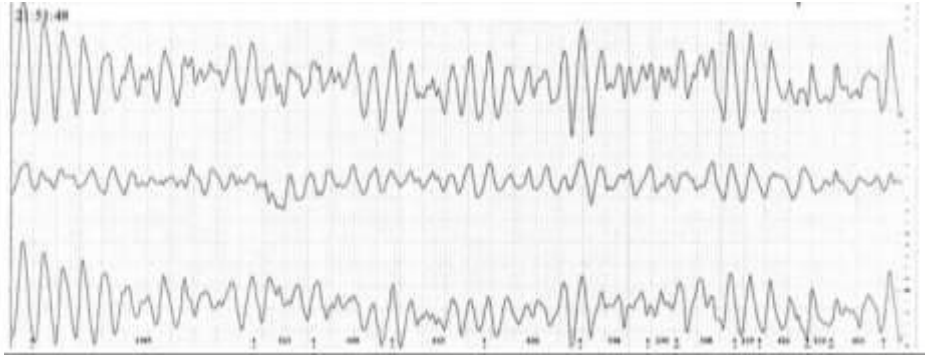
REVIEW ARTICLE

Perioperative management of hereditary arrhythmogenic syndromes

C. Staikou*, K. Chondrogiannis and A. Mani

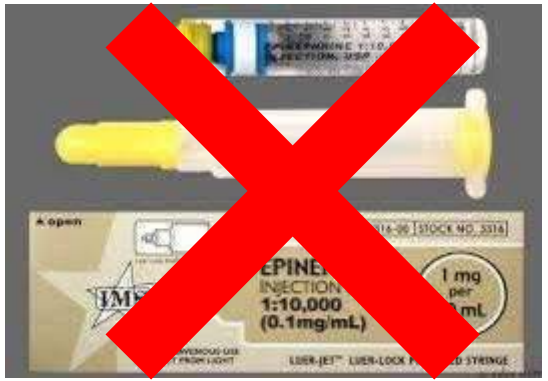
- CPVT -- characterized by adrenergic-dependent, potentially lethal tachyarrhythmias, structurally normal hearts
- Autosomal-dominant and mutations of cardiac RYR gene-2
- Often presents as syncope with exertion





CPVT

- Continue maintenance Beta blockers on the day of surgery.
- Establish a calm and quiet environment for the patient.
- Generous anxiolytic use preoperatively
- Ensure adequate depth of anesthesia prior to noxious stimuli (endotracheal intubation)
- Aggressively treat PONV and postoperative pain.
- Telemetry monitoring postoperatively
- AVOID EPINEPHRINE in a code situation

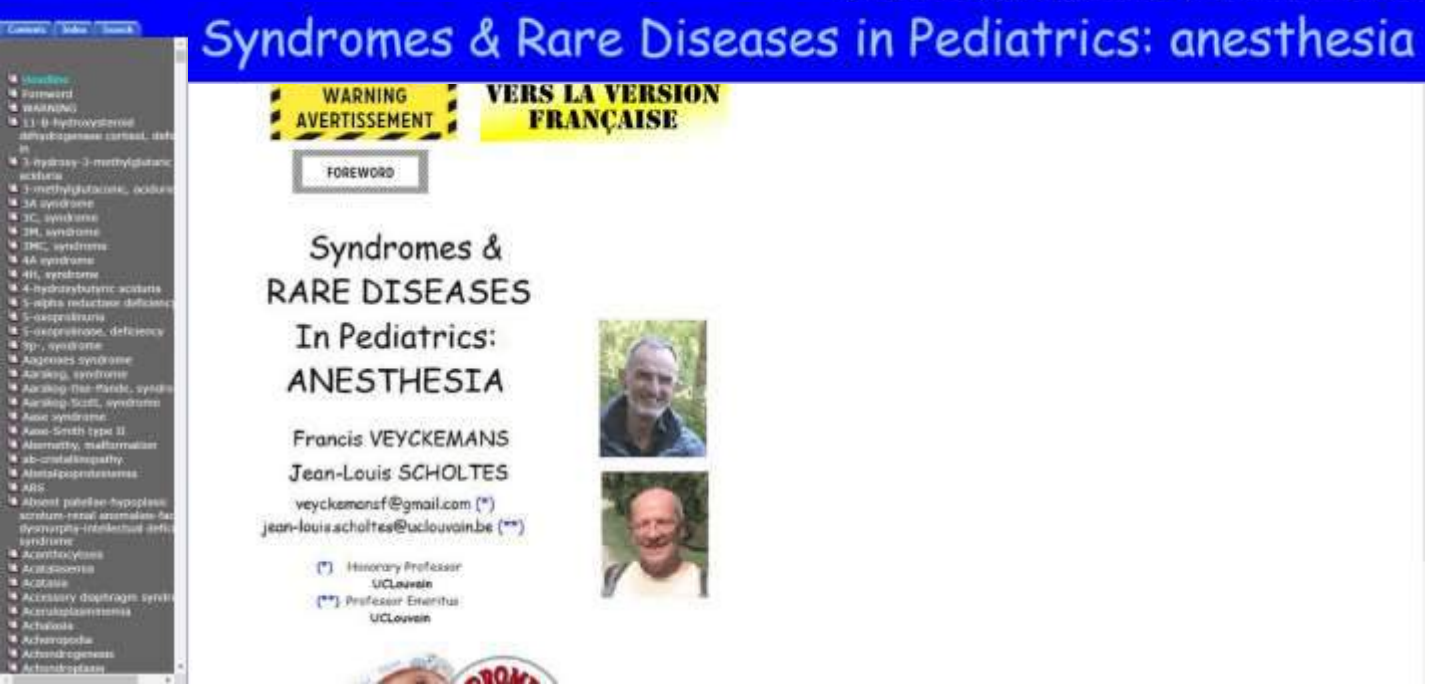


Bellamy D, Nuthall G, Dalziel S, Skinner J.R. Catecholaminergic polymorphic ventricular tachycardia: the cardiac arrest where epinephrine is contraindicated. Pediatric Crit Care Med. 2019; 20: 262-268

SUMMARY

- Thorough preoperative evaluation for all syndromic patient, system based
- Cardiology colleagues as a valuable resource
- Coordinated multidisciplinary care
- Using advanced airway technique is NOT a sign of weakness with syndromic patients!

Syndromes & Rare Diseases in Pediatrics: anesthesia



https://sites.uclouvain.be/anesthweekly/MRP_ENG/index.html

Society for Pediatric Anesthesia

education • research • patient safety

ESPA

European Society for Paediatric Anaesthesiology

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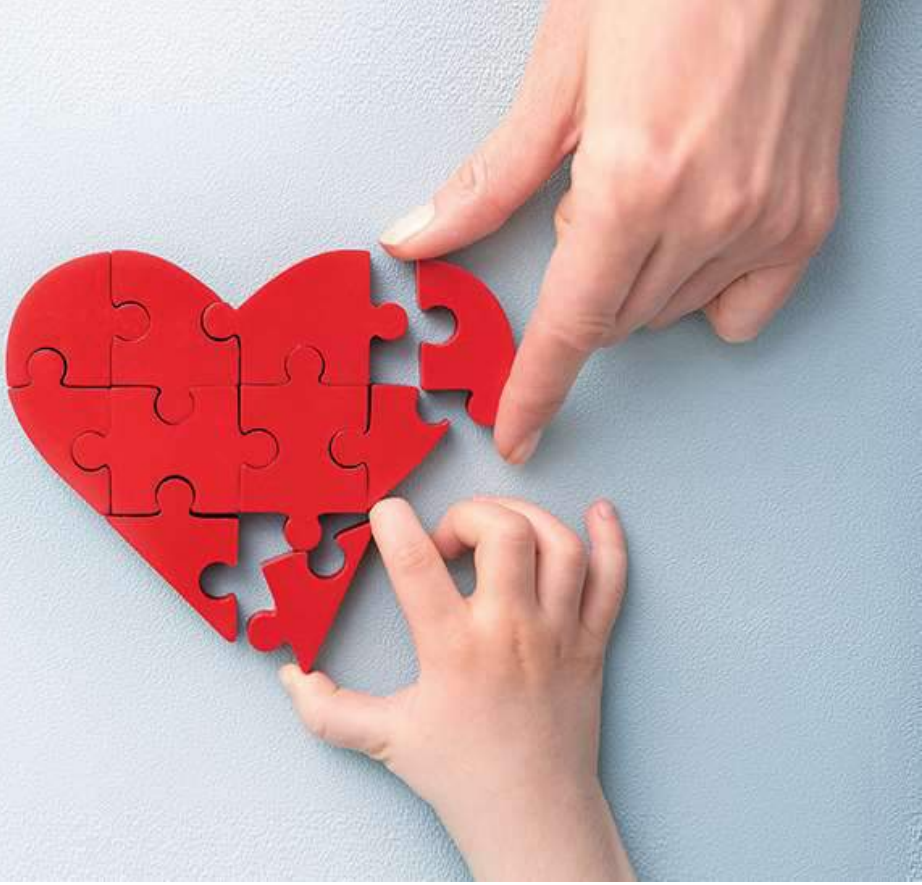
THANK YOU



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What the anesthesiologist needs to know about genetic syndromes before putting a cardiac patient to sleep?

EVERYTHING ABOUT THE PATIENT?