

CHARGE 101 Hobart

The CHARGE Syndrome Checklist from head to toe.
Anesthesia risks, bone health and migraines.



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Objectives

- To use the CHARGE Syndrome checklist as a guide for medical surveillance.
- To help you understand the risks of anesthesia in CHARGE syndrome.
- To discuss bone health and delayed puberty, particularly around prevention.
- Migraines and the use of Botox.

There are Always Risks of Complications with Anaesthesia

- “...you sign a consent”
- Are you informed?
- Are Individuals with CHARGE Syndrome more at Risk?

If yes, what are the risks?
Who should know?



Friends at APSEA 2006

Growing up With CHARGE Syndrome

Kennedy 0-6 years



Age 0-2 years: 7 surgeries



Age 2-4 years: 3 surgeries



Age 4-6 years: 6 surgeries

Kennedy's Four ICU Admissions

- 2 weeks – open heart surgery
- 6 months – G-tube/fundoplication extubation attempted (x 3)
- 18 months – aspiration pneumonia
- 6 yrs – heart surgery – pneumonia after heart surgery

Postoperative Airway Events of Individuals with CHARGE Syndrome

Population n=9

Mean age 11.8 years (\pm 8.0)

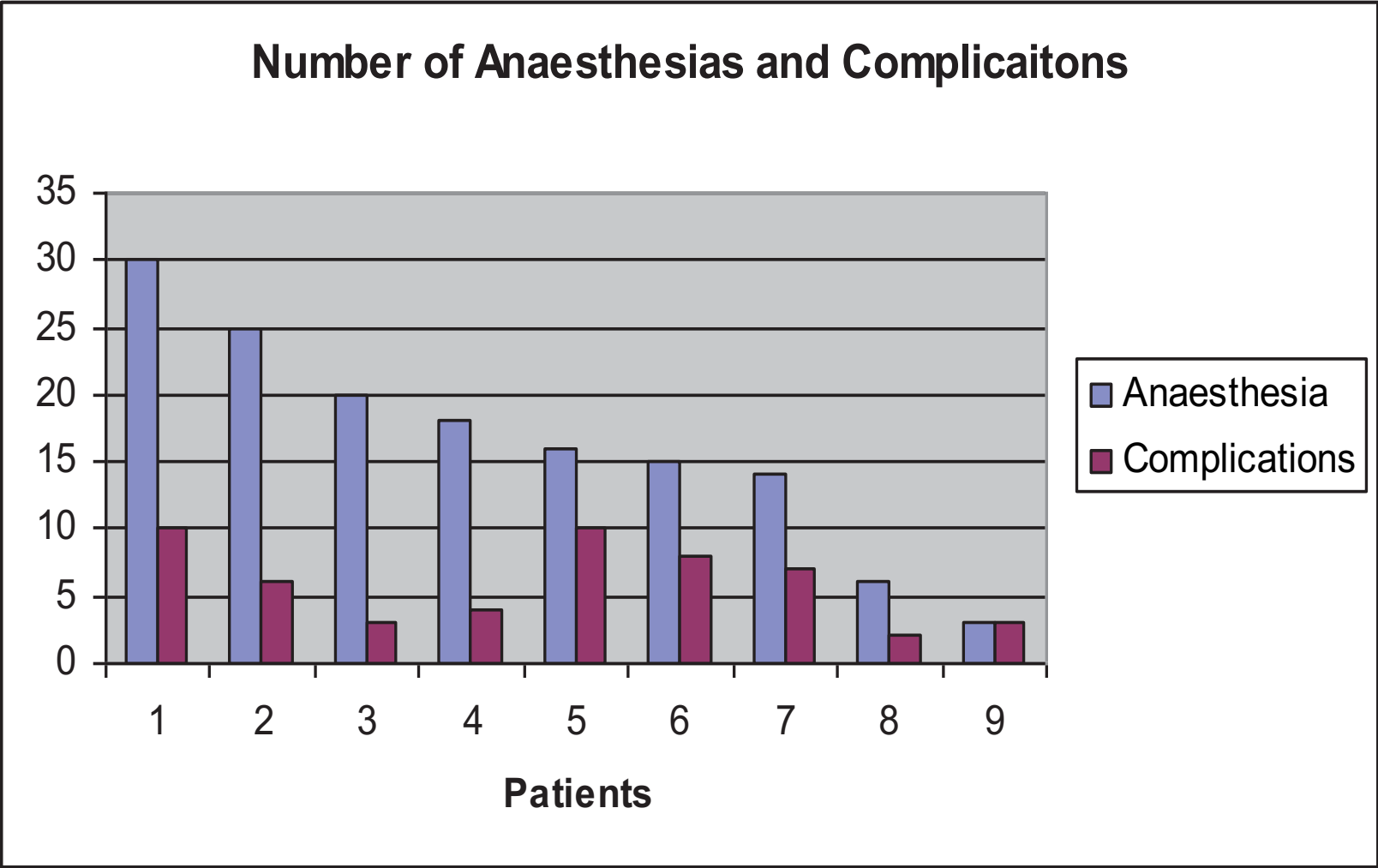
215 surgeries (mean 22 per child)

147 anesthesia's (mean 16 per child)



Postoperative events (reintubation for apneas and desaturations, airway obstruction due to excessive secretions)

Results



35% (51/147) of anaesthesias resulted in post-operative events (>60% were major)

Summary of findings

1. Anesthesia related events occurs most often with heart, diagnostic scopes and gastrointestinal procedures.
2. High risk of complications in individuals with Nissen fundoplication and/or gastrostomy/jejunostomy tubes
3. Combining multiple procedures under one anesthetic does not lead to a increase in post operative airway events.

What about individuals with CHD7 mutations who have mild clinical criteria?

- Will they be at risk in the future?
- Have they actually been challenged with surgeries?

Airway problems are common in CHARGE syndrome and are often due to cranial nerve abnormalities (IX, X).

A study by Morgan et al. (1993) examining 50 patients with CHARGE syndrome found that 86% had upper airway abnormalities, 56% had posterior choanal abnormalities and 42% had retrognathia leading to intubation difficulties.

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Anaesthesia issues in CHARGE syndrome – what are the risks?

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Introduction

Children with CHARGE syndrome often require multiple surgeries throughout their lifetime to manage structural and functional anatomical abnormalities. It is important therefore, to consider any risks caused by anaesthesia. These risks tend to vary based on the individual child's CHARGE syndrome characteristics.

Airway Obstruction

Airway problems are common in CHARGE syndrome. Figure 1 shows the normal anatomy of the airway, as well as associated cranial nerves. Most children with CHARGE syndrome have cranial nerve abnormalities. Pertinent problems are those with the glossopharyngeal and vagus nerves, which are responsible for innervating the pharynx and larynx and allowing swallowing to occur (Blake et al., 2008).

Problems during with oral intubations can be a result of cranial nerve problems (Blake et al., 2009a). A study by Morgan et al. (1993) examining 50 patients with CHARGE syndrome found that 86% had upper airway abnormalities, 56% had posterior choanal abnormalities and 42% had retrognathia leading to intubation difficulties.

A study done by Natto et al. (2007) looked at 10 consecutive patients with CHARGE syndrome who underwent fiberoptic laryngoscopy. All patients had upper airway obstruction at the larynx level, with a hypotonic larynx and hypertrophic arytenoids. They also had uncoordinated movement of the supraglottis, which made it difficult to maintain airway patency. Other problems included glossopexia, oesophageal fistula and clefts. See Figure 7, page 2.



Securing an Airway

There are reports of cancelled surgery and aborted anaesthesia from failure to secure airway control in children with CHARGE syndrome (Hara et al., 2009). An early study showed greater difficulty with tracheal intubation that increased with age (Stack and Wyse, 1991). Laryngomalacia may contribute to upper airway collapse during light anaesthesia and may lead to a requirement of CPAP during induction.

Since the pharynx and larynx of CHARGE syndrome patients may be much smaller than expected from their physique, a smaller sized laryngeal mask airway (LMA) should be used first (Hara et al., 2009). A wide range of equipment sizes should be on-hand, including that to perform a tracheostomy (Stack and Wyse, 1991). It is suggested that a paediatric

Take Home Messages

- Your children are at high risk of post-operative anaesthesia complications. Combining procedures during one anesthesia does not increase the risk of post-operative airway events.
- The anaesthesiologist needs to be aware that, even with simple procedures, the individual with CHARGE syndrome is at high risk of post-operative events.



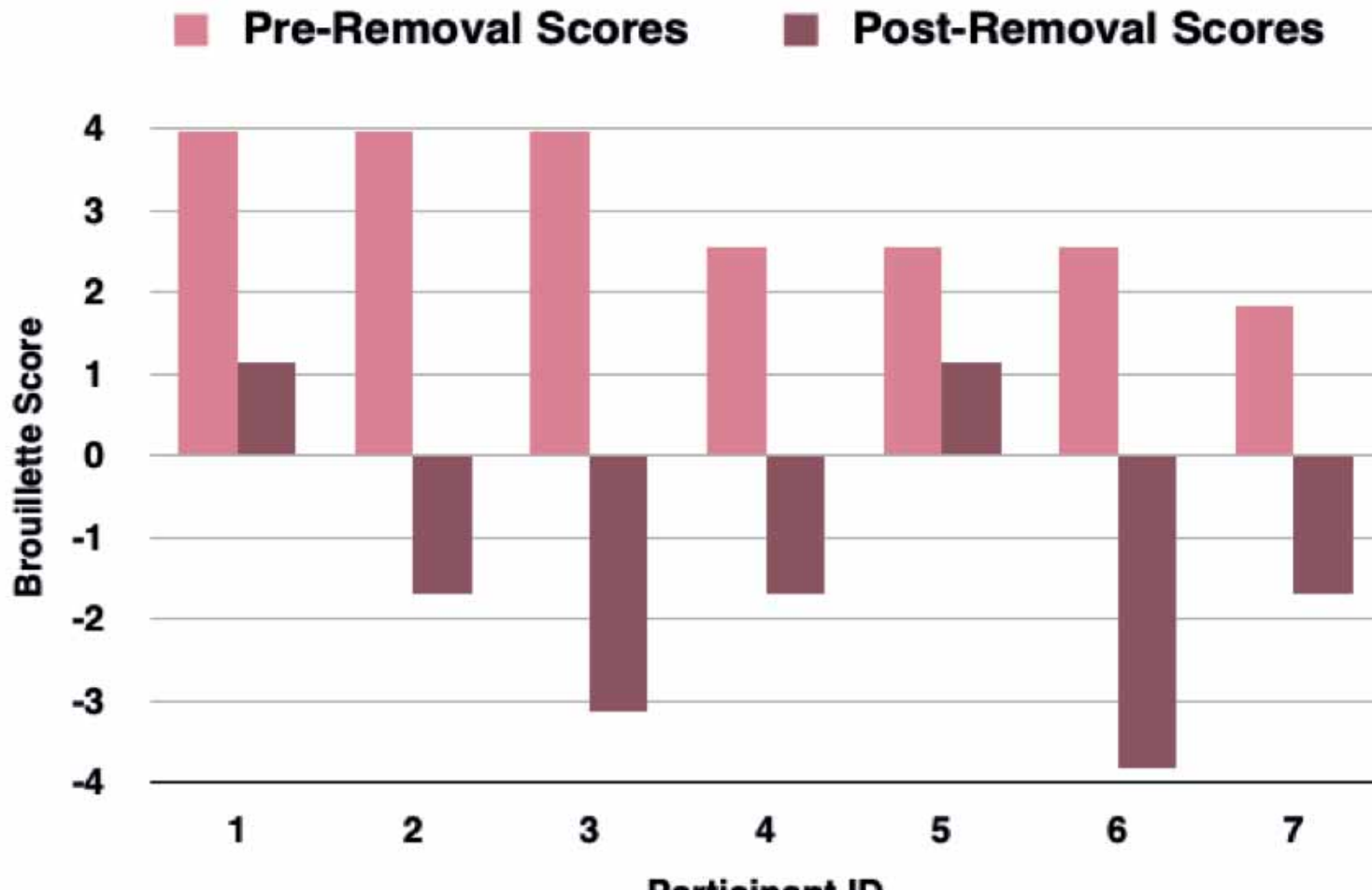
Mackenzie's Story 0-6 years

- 27 surgical procedures
- 18 anesthesia's
- 4 complications
- Multiple ICU admissions
- Removal of tonsils and adenoids reduced ICU admissions



Understanding Sleep Apnea in Children with CHARGE Syndrome

Removal of Tonsils & Adenoids



Brouillette
Scores >3.5 highly
predictive of OSA,
between -1 to 3.5
suggestive for OSA,
<-1 absence of OSA

Take Home Messages

- Obstructive sleep apnea is highly prevalent in the CHARGE Syndrome population
- Removal of tonsils and adenoids may be beneficial



Carrie-Lee & Freddy

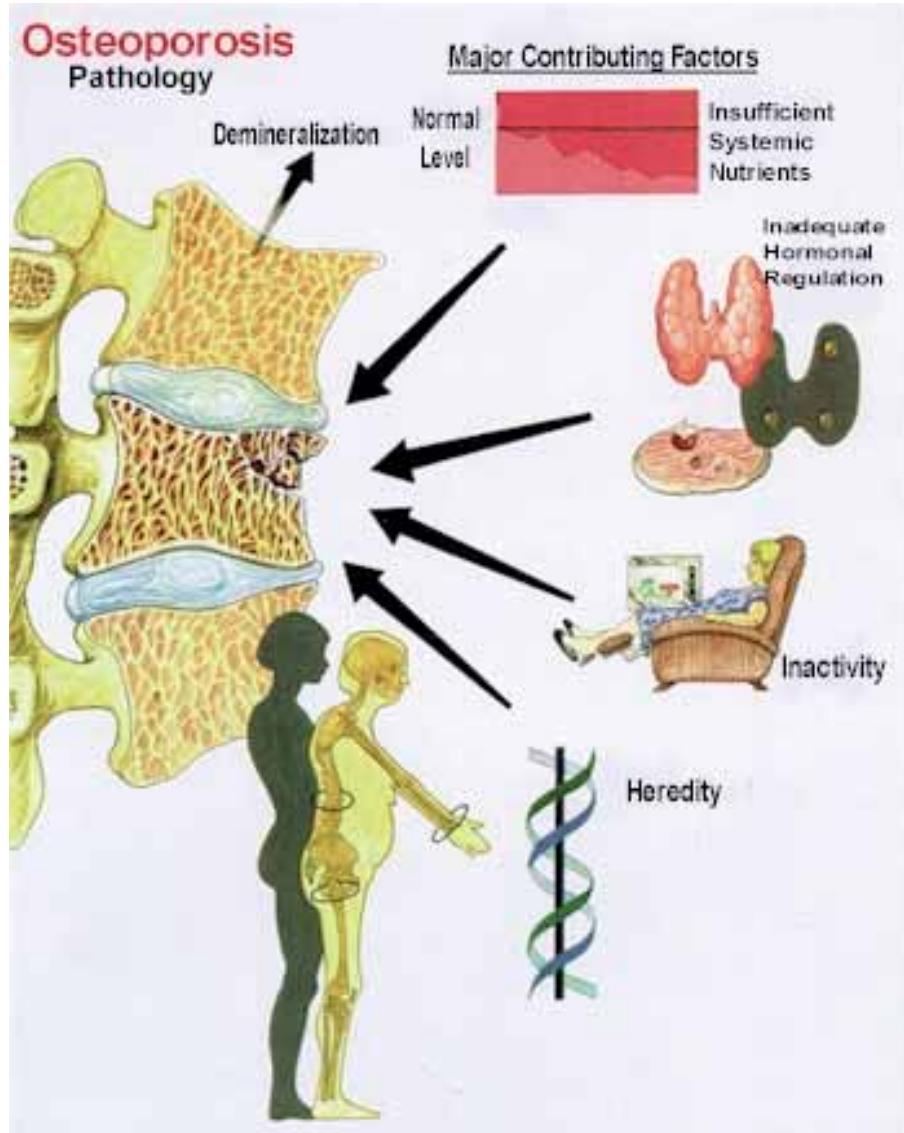
Osteoporosis

Why do I Need to Worry?



Two friends with CHARGE Syndrome

What is Osteoporosis?



Bone is a living tissue

Calcium and Phosphate
(CaPo_4) [Mineral]

Collagen [Protein]

Demineralization of bone and/or thinning of bone.

Results

- Approximately 50% of individuals with CHARGE Syndrome do not consume the Recommended Daily Allowed (RDA) for Calcium and most do not consume enough for Vitamin D.

Puberty

- Many CHARGE individuals do not undergo normal puberty (especially males).

Activity:

- Teens ages 13 – 18 are significantly less active than controls, particularly during weekends (when there is less 1:1 support).



Great article to share with your Endocrinologist

Osteoporosis Prevention Diet

Calcium*

Pre-pubertal (4-8 years) 800 mg/day

Adolescents (9-18 years) 1300 mg/day

Adults 1000 mg /day

* Call if renal issues

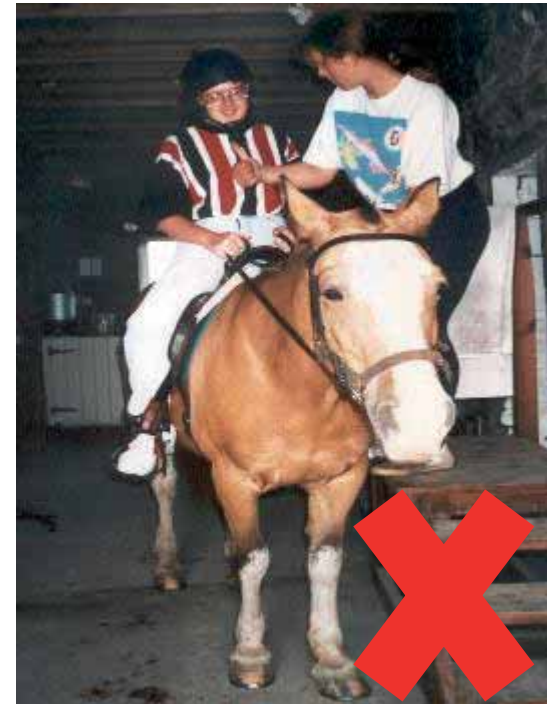
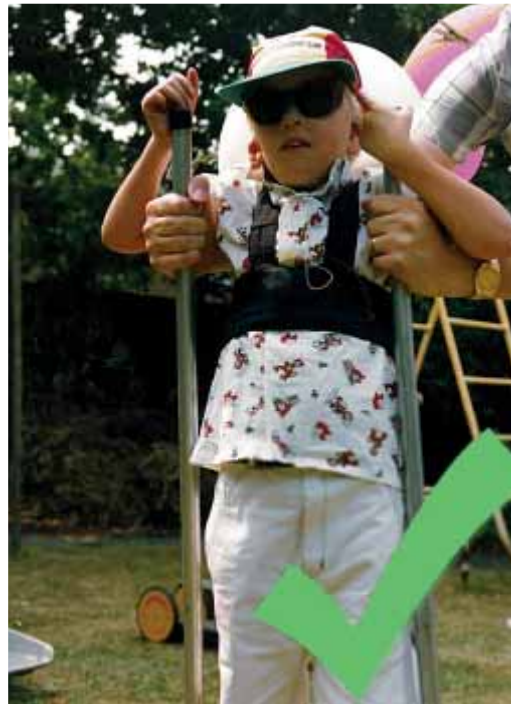
Vitamin D

1000 IU (International Units)

Fish Oils



Exercise – Non-weight bearing Great for Balance but not for Bone Mineral Density (BMD)



Physiotherapy & Recreational therapy

Prevention of Osteoporosis in CHARGE Syndrome

- Adequate diet and exercise*
- Regular follow up with an endocrinologist for height, weight and pubertal status
- Sex Hormone replacement therapy
 - Testosterone in boys start at low dosage
 - Low dosage estrogens in females

*Seek physiotherapy, recreational therapy



Scoliosis in CHARGE Syndrome



Incidence of Scoliosis Among Adolescents and Adults with CHARGE Syndrome.
C. Doyle, Medical Student Elective, Dalhousie University

CHARGE Syndrome from Birth to Adulthood: an individual reported on from 0 - 33 years.



Searle et al American Journal of Medical Genetics 2005:113A(3), 344-349.

New Medical Issues (N = 30)

Medical Issue	Number	%
Scoliosis	19	63
Sleep Apnea	13	43
Abdominal Colic	12	40
Retinal Detachment/Cataract	10	33
Migraines	8	27
Seizures/Epilepsy	5	17
Urinary Tract Infections	5	17
Hypoglycemia	1	3

Migraines – Are these missed in CHARGE syndrome?

- Abnormal migraines
- Pain Scale
- Feeding Scale



MT History

- Presented on general pediatric ward.
- Abnormal ears and facial palsy.
- “Sand not felt in the eyes”.
- CHD7 anomalies, diagnosed with CHARGE Syndrome with few features.



Showing Right facial palsey

A case of CHARGE Syndrome demonstrates the importance of Cranial nerves – in clinical recognition and exploring Hypoalgesia

- Botox treatment to normal facial nerve. Every 3-4 months. Tracking improvement by diary of migraines.



**CHARGE SYNDROME CHECKLIST: HEALTH SUPERVISION ACROSS THE LIFESPAN
(FROM HEAD TO TOE)**

*Shaded boxes indicate key assessment points

	INFANCY (0-2 years)	CHILDHOOD (3-11 years)	ADOLESCENCE (12-17 years)	ADULTHOOD (18+ years)
GENETICS	Clinical diagnosis (Éliale et al. or Verloes or Hale et al. criteria) Genetic testing – Genetics consult (CHU/ analysis, array CGH) Genetic counselling			
NEUROLOGY	CNS malformations/hypoplasia olfactory bulb/temporal bone (semicircular canal) malformations – requires MRI/CT Seizures – more common at older ages – consider tbc Cranial nerve problems – monitor for absent sense of smell, facial nerve palsy, sensorineural hearing loss, vertigo, swallowing problems			
EYES, EARS, NOSE AND THROAT	Coloboma, risk of retinal detachment & Ophthalmology consult (dilated eye exam in infancy, vision assessments) Lamellar exposure – lubricating eye drops Photophobia – tinted glasses, sunhat Choanal atresia/cleft palate/tracheoesophageal fistula & EHI/Plastics consult Audiometry and tympanometry, monitor for recurrent ear infections Adaptive services for individuals with deafness/blindness Lachlear implant assessment if applicable Obstructive sleep apnea – monitor for tonsil/adenoid hypertrophy Excessive secretions – consider botox, medication Dental issues – consider cleaning under anesthetic			
CARDIOLOGY CARDIOLOGY	Cardiac malformations common – major/minor defects, valvular (rinc or arrhythmias possible (echocardiogram, chest x-ray, ECG) & Cardiology consult Sinusitis, pneumonia, asthma & monitor Anesthesia risk (difficult intubations/postop airway obstruction/aspiration) – extensive preoperative assessment, combine surgical procedures			
GASTROENTEROLOGY GASTROENTEROLOGY	Gastroesophageal reflux – Gastroenterology consult – consider motility agents with proton pump inhibitor Poor suck/chew/swallow & feeding team assessment/intervention Aspiration risk, tracheoesophageal fistula – swallowing studies May need supplemental feeds – frequently requires gastrostomy tube or Gastro-jejunostomy tube Constipation – consider ben na glycoside with polyethylene glycol Renal anomalies – abdominal u/s +/- E VCUG, blood pressure monitoring			
ENDOCRINOLOGY	Hypoparathyroid hypocalcaemia – LH, PSH by 3 months Genital hypoplasia (if undescended testes & consider orchidopexy) Delayed puberty – Endocrinology consult & gonadotropin levels, MRI Osteoporosis – DEXA scan Poor growth – Endocrinology consult – GH stimulation test, GH therapy Obesity & monitor Fertility and contraception & discuss			
IMMUNE SYSTEM	Note presence of thymus at open heart surgery Routine immunizations/antibody titres to immunizations in adolescence Recurrent infections – Immunology consult			
MSK	Scoliosis/kyphosis monitor Mobility (affected by ataxia, hypotonia) & evaluate Assess gross and fine motor skills – Occupational Therapy, Physiotherapy Communication, language, writing abilities – Speech Language Therapy Consider deafblind consultant Prepare for transitions to school, situations, places, systems Psychoeducational assessment, individualized Education Plan Sleep disturbances – consider melatonin Behavior management – self regulation, impulse control, anxiety, obsessions, compulsions, anger Life skills & support Life shifts/adaptive behaviour/social skills/social play Address sexuality Family stress – offer supports and resources Medical self-management – work on managing medications, understanding conditions, seeing healthcare provider independently			

*Abbreviations listed on page 2

Trider C, ArraBRobar A, van RavenwaayBArts C, Blake K

IWK CHARGE Clinic Students & Residents Using the CHARGE Checklist



Questions and Answers



DR KIM BLAKE

HOME DR KIM BLAKE NEWS CONTACT

TEXAS 2019

CHARGE CHECK-LIST

CHARGE GUT

MEDICAL EDUCATION



DR KIM BLAKE

DR. Kim Blake is a professor of Pediatrics at Dalhousie University in Nova Scotia, Canada. She has been researching in CHARGE syndrome over the last 35 years and has published extensively. She has answered research questions concerning post-

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