Saturday, August 3, 2019

Breakout Session A2 • 10:45-11:45am • Grand Ballroom C

Gastrointestinal Issues in CHARGE Syndrome Does Your Gut Talk to You?

Dr. Kim Blake, IWK Health Centre

Presenter Information

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 explored post-operative airway events, sleep apnea, bone health, cranial nerve abnormalities, and gastrointestinal issues. In the last 10 years Dr. Blake has partnered with Dr. Jason Berman and they have developed a zebra fish model of CHARGE syndrome to answer further research questions. With this model we have been able to understand about the abnormalities of the vagus nerve and gut mobility in CHARGE syndrome which has influenced our knowledge of gut motility. Anesthesia and sedation risk has also been researched in our zebra fish model. This supports the clinical findings that individuals with CHARGE syndrome have increased risk following anesthesia and should have combined procedures where possible in one anesthesia. Kim is very proud of the CHARGE syndrome checklist which has been developed both for families, individuals, and professionals to use as a guide and a teaching tool for anybody dealing with CHARGE syndrome.

Presentation Abstract

Review of Gastrointestinal (GI) motility and the connection to the vagus nerve and microbiome in CHARGE syndrome. Gastrointestinal (GI) dysfunction including feeding, and digestion difficulties are highly prevalent and represent a serious challenge for many individuals with CHARGE syndrome. We are much further along the journey in understanding the GI tract which is the largest organ of the body and deserves more attention. In this presentation we will summarize in an easy digestible format the knowledge to date; this will help you understand and advocate for the gut in CHARGE. We are excited in sharing with you the research undertaken by the Atlantic Canadian CHARGE syndrome research group. We have been studying the type of bacteria found in the gut called the “Gut microbiome.” We are continuing to recruit for this research at the conference, pop by and see us to learn more.

Learning Objectives

- To understand more about the gastrointestinal (GI) issues faced by individuals with CHARGE syndrome.
- To be an advocated for the CHARGE ‘gut’ and have the up to date literature at your fingertips to share with your specialist.
- To be knowledgeable about microbiome and how it may be impacted in CHARGE syndrome.

Gastrointestinal issues in CHARGE syndrome: Does your gut talk to you?

DEEP IN THE ♥ OF CHARGE
14th International CHARGE Syndrome Conference
Dallas, Texas • August 2-5, 2019

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DALHOUSIE UNIVERSITY

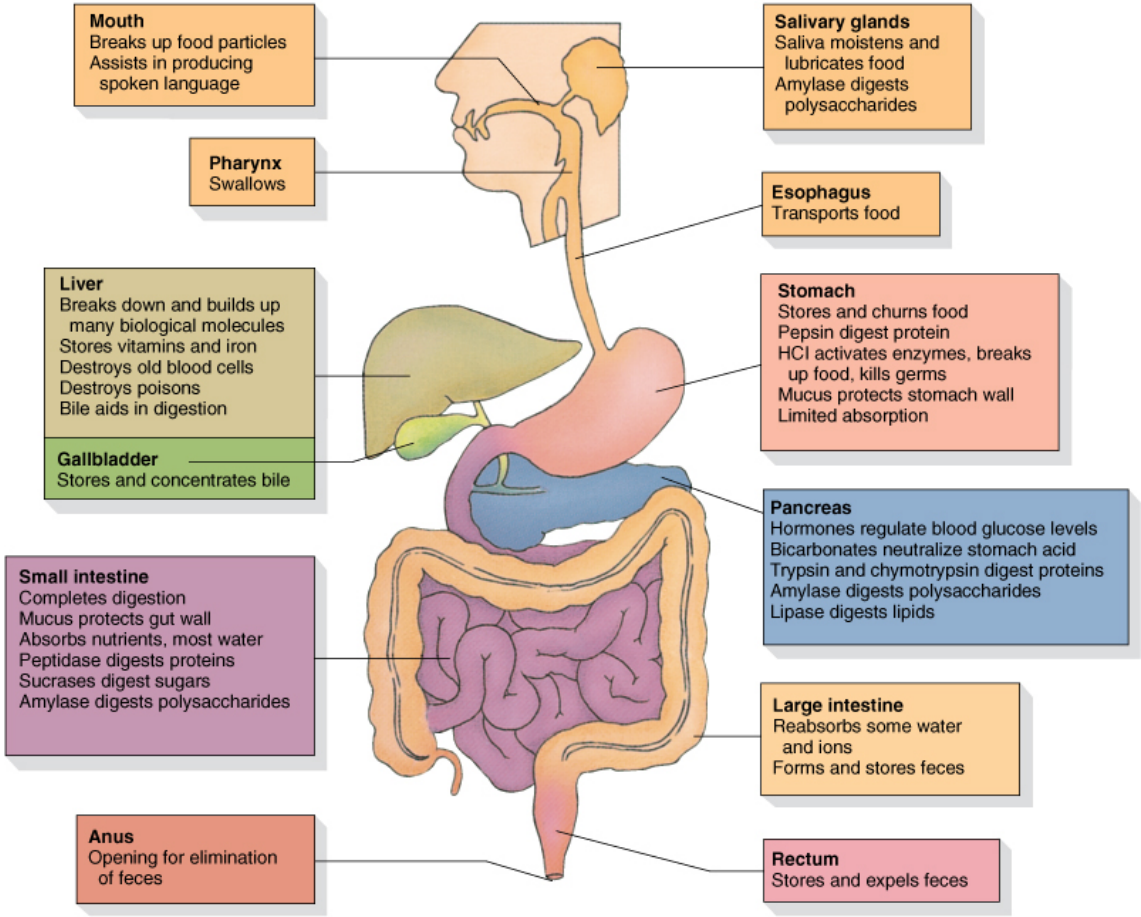
Case History

Objectives

After this presentation you will have a greater understanding of:

1. The breath of gastrointestinal issues in CHARGE syndrome
2. The clinical and basic science research, including our zebra fish model.
3. The provisional results of our microbiome studies

The GI tract goes from mouth to anus



The upper GI tract

Cranial nerve innervations and structural abnormalities are key issues

- Cranial facial abnormalities can interfere with feeding particularly in infancy.
- Children with choanal atresia/ stenosis have significantly more GI symptoms then those without.#
- Excessive salivation secretion can be a problem
- Mouth over stuffing and pocketing is prevalent.*

#Macdonald 2016 AJMG
*Hudson 2016 AJMG

Gastroesophageal Reflux (GER) and tube feeding

- Gastroesophageal Reflux is often severe and difficult to treat.
- Tube feeding is highly prevalent and can be protracted
- Tube feeds vs. oral feeders have more
  - Stomach pain
  - Discomfort when eating
  - Food and drink limits
  - Trouble swallowing
  - Nausea and vomiting
  - Constipation

"Motility issues" are a key problem.

Macdonald 2016 AJMG

Abdominal Pain

- Prevalent and difficult to assess and the underlining diagnosis is often missed.
- Digestion issues are clinically present. There has been very little research in this area.

"The gut is different in CHARGE syndrome"

Hartshorne and Stratton,
Research on pain scale

Constipation

How many of you have problems with this?

Prevention:

- Fluids
- Exercise
- Behavioral therapy
- Diet
- Massage

Treatment:

- Polyethylene glycol / MiraLAX
- PEG
- Senocot
- Behavioral techniques

Risk factors for poor bone health in adolescents and adults with CHARGE syndrome.

Key Findings

- 87% of individuals are not getting enough vitamin D
- 41% not getting enough calcium

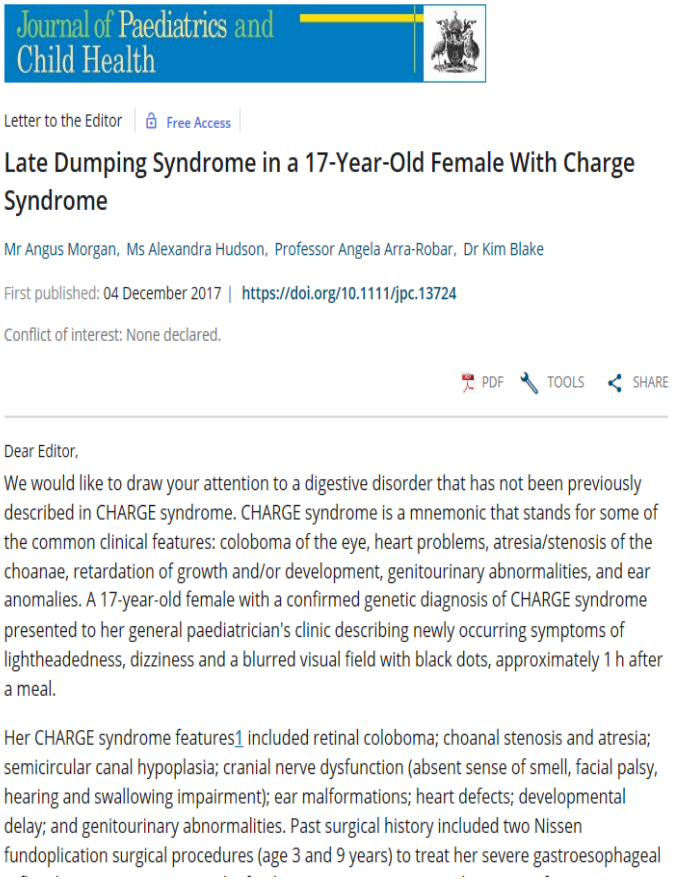
Recommendations:

- Increase in the amount of calcium and vitamin D
- Replace sex hormones.
- Increase in weight bearing activity
- # 100 iu Vit D

Forward 2007 AJMG

Conditions that are missed and need to be on the differential diagnosis

- Abdominal colic
- Pocketing/Overstuffing
- Gall stones
- Dumping syndrome



Journal of Paediatrics and Child Health

Letter to the Editor | Free Access

Late Dumping Syndrome in a 17-Year-Old Female With Charge Syndrome

Mr Angus Morgan, Ms Alexandra Hudson, Professor Angela Arra-Robar, Dr Kim Blake

First published: 04 December 2017 | https://doi.org/10.1111/jpc.13724

Conflict of interest: None declared.

PDF TOOLS SHARE

Dear Editor,

We would like to draw your attention to a digestive disorder that has not been previously described in CHARGE syndrome. CHARGE syndrome is a mnemonic that stands for some of the common clinical features: coloboma of the eye, heart problems, atresia/stenosis of the choanae, retardation of growth and/or development, genitourinary abnormalities, and ear anomalies. A 17-year-old female with a confirmed genetic diagnosis of CHARGE syndrome presented to her general paediatrician's clinic describing newly occurring symptoms of lightheadedness, dizziness and a blurred visual field with black dots, approximately 1 h after a meal.

Her CHARGE syndrome features[1] included retinal coloboma; choanal stenosis and atresia; semicircular canal hypoplasia; cranial nerve dysfunction (absent sense of smell, facial palsy, hearing and swallowing impairment); ear malformations; heart defects; developmental delay; and genitourinary abnormalities. Past surgical history included two Nissen fundoplication surgical procedures (age 3 and 9 years) to treat her severe gastroesophageal

Foods suitable on a low-fodmap diet

fruit	vegetables	grain foods	milk products	other
fruit banana, blueberry, boysenberry, canteloupe, cranberry, durian, grape, grapefruit, honeydew melon, kiwifruit, lemon, lime, mandarin, orange, passionfruit, pawpaw, raspberry, rhubarb, rockmelon, star anise, strawberry, tangelo Note: if fruit is dried, eat in small quantities	**vegetables** alfalfa, artichoke, bamboo shoots, bean shoots, bok choy, carrot, celery, choko, choy sum, endive, ginger, green beans, lettuce, olives, parsnip, potato, pumpkin, red capsicum (bell pepper), silver beet, spinach, summer squash (yellow), swede, sweet potato, taro, tomato, turnip, yam, zucchini **herbs** basil, chili, coriander, ginger, lemongrass, marjoram, mint, oregano, parsley, rosemary, thyme	**cereals** gluten-free bread or cereal products **bread** 100% spelt bread **rice** **oats** **polenta** **other** arrowroot, millet, psyllium, quinoa, sorgum, tapioca	**milk** lactose-free milk, oat milk*, rice milk, soy milk* *check for additives **cheeses** hard cheeses, and brie and camembert **yoghurt** lactose-free varieties **ice-cream substitutes** gelati, sorbet **butter substitutes** olive oil	**sweeteners** sugar* (sucrose), glucose, artificial sweeteners not ending in '-ol' **honey substitutes** golden syrup*, maple syrup*, molasses, treacle *small quantities

Dietary advice that may help digestion issues.

- Reduce simple carbohydrates (Bread, pasta)
- Small amounts of food regularly.
- Exercise and weight control.
- Low FODMAPP diets.

Etiology and functional validation of Gastrointestinal motility dysfunction in a zebra fish model of CHARGE syndrome

Loss of chd7 in zebrafish results in:

- Smaller stomachs and GI tracts with normal epithelial and muscular histology.
- Decrease and disorganized vagal nerve projections particularly in the fore gut.
- Less ability to empty their GI tract only minimally improved by pro kinetic agents.

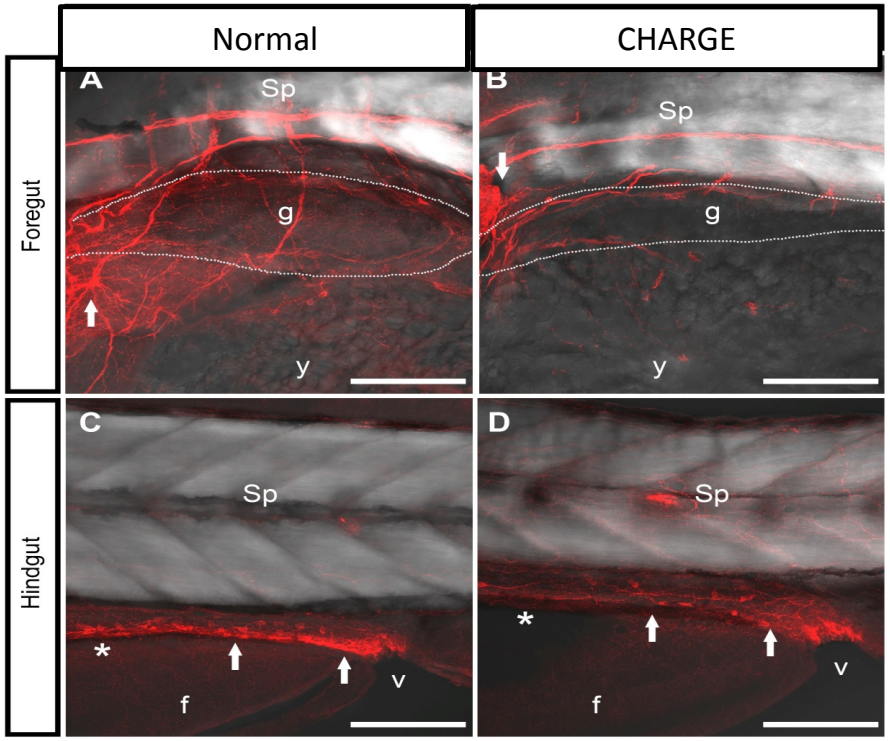
Future

Zebrafish are an excellent model for studying compounds that improve GI motility in individuals with CHARGE syndrome.

International Journal of Pediatric Otorhinolaryngology V82, March 2016, pgs. 107-115

Clooney et al FEEBS 285,11, 2018

Innervation of the CHARGE Zebrafish (chd7) and normal controls in the gut



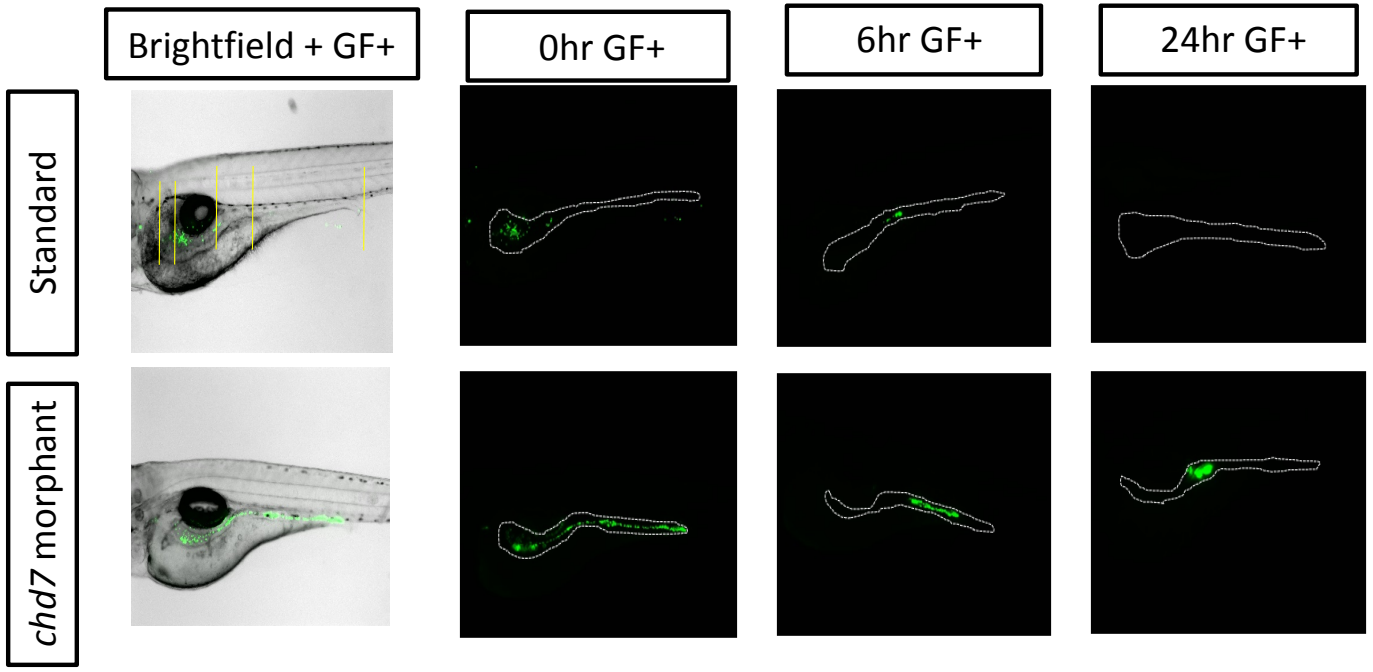
- Decreased enteric nerve branches around the fore gut (Compare A & B)
- Difference in size and shape of the gut in the CHARGE fish.

Sp = spine, F = ventral fin, V = vent, G = gut (outlined in hashed line), arrow = vagal nerve plexus, y = yolk

Clooney et al FEEBS 285,11, 2018

International Journal of Pediatric Otorhinolaryngology V82, March 2016, pgs. 107-115

Decreased motility shown in CHARGE zebrafish by delayed emptying of GI tract



*Florescent green = tagged food travel. GI tract of zebra fish over time

A feeding scale for CHARGE syndrome

Date: _____

Name of Individual: _____

Age:_____ Gender (Circle one): Male Female Not Disclosed

Completed By (Circle one): Mother Father Feeding Therapist Nurse/Physician Other:_____

What percentage of your child/adult's daily fluid/nutrition intake is by G/J tube feeding? (Circle one percentage):

0%	25%	50%	75%	95%

	Circle one number on the scale:	Never	A Little	Sometimes	A lot	Always
1	He/she will refuse food when eating orally.	0	1	2	3	4
2	He/she takes longer than 45 minutes to eat orally.	0	1	2	3	4
3	He/she takes less than 15 minutes to eat orally.	0	1	2	3	4
4	He/she needs **close supervision** when eating orally.	0	1	2	3	4
5	He/she needs **someone in the room** when eating orally.	0	1	2	3	4
6	He/she has problems cutting food when eating orally.	0	1	2	3	4
7	He/she has problems feeding him/herself when eating orally.	0	1	2	3	4
8	He/she chokes or coughs when eating orally.	0	1	2	3	4
9	He/she has **trouble** chewing food.	0	1	2	3	4
10	He/she has **trouble** swallowing food.	0	1	2	3	4
11	He/she has to be **told or reminded** to chew.	0	1	2	3	4
12	He/she has to be **told or reminded** to swallow.	0	1	2	3	4
13	He/she does not like to mix food textures when eating (e.g. mixing puree and solid food).	0	1	2	3	4
14	He/she accidentally loses food out of his/her mouth during eating.	0	1	2	3	4
15	He/she will over-stuff his/her mouth with food during eating.	0	1	2	3	4
16	He/she has difficulty moving food around with his/her tongue during eating.	0	1	2	3	4
17	He/she has a hard time feeling food or anything touching the inside of his/her mouth.	0	1	2	3	4
18	He/she dislikes oral eating.	0	1	2	3	4
19	He/she lets food sit in his/her **cheeks or palate** during eating (on purpose or not).	0	1	2	3	4
20	He/she will have food hidden in his/her **cheeks or palate** after the meal has ended (on purpose or not).	0	1	2	3	4
21	The **Parent/Caregiver** gets worried about their child/adult's ability to eat orally.	0	1	2	3	4
22	The **Parent/Caregiver** has difficulties feeding their child/adult. (e.g. preparing food the right way, getting enough information about helping them eat/drink)	0	1	2	3	4

	Does the child/adult have problems with:	No	Yes
23	Cold foods	0	1
24	Room temperature foods	0	1
25	Warm foods	0	1
26	Thin liquids (e.g. water)	0	1
27	Pureed foods (e.g. applesauce)	0	2
28	Mashed lumpy food (e.g. mashed potatoes or mashed vegetables)	0	2
29	Soft chewable foods (e.g. bread, crackers)	0	2
30	Tough chewable foods (e.g. meat)	0	1
31	Hard vegetables and fruit (e.g. raw apples)	0	1

Total Score (sum of all items)	**/100 total points**
Circle one:	**Feeding difficulties:** **Mild (0-25 points)** **Moderate (26-50 points)** **Severe (51-100 points)**

Circle one number on the scale:		Never	A Little	Sometimes	A lot	Always
16	He/she has difficulty moving food around with his/her tongue during eating.	0	1	2	3	4
17	He/she has a hard time feeling food or anything touching the inside of his/her mouth.	0	1	2	3	4
18	He/she dislikes oral eating.	0	1	2	3	4
19	He/she lets food sit in his/her **cheeks or palate** during eating (on purpose or not).	0	1	2	3	4
20	He/she will have food hidden in his/her **cheeks or palate** after the meal has ended (on purpose or not).	0	1	2	3	4

Subsection of Feeding Scale

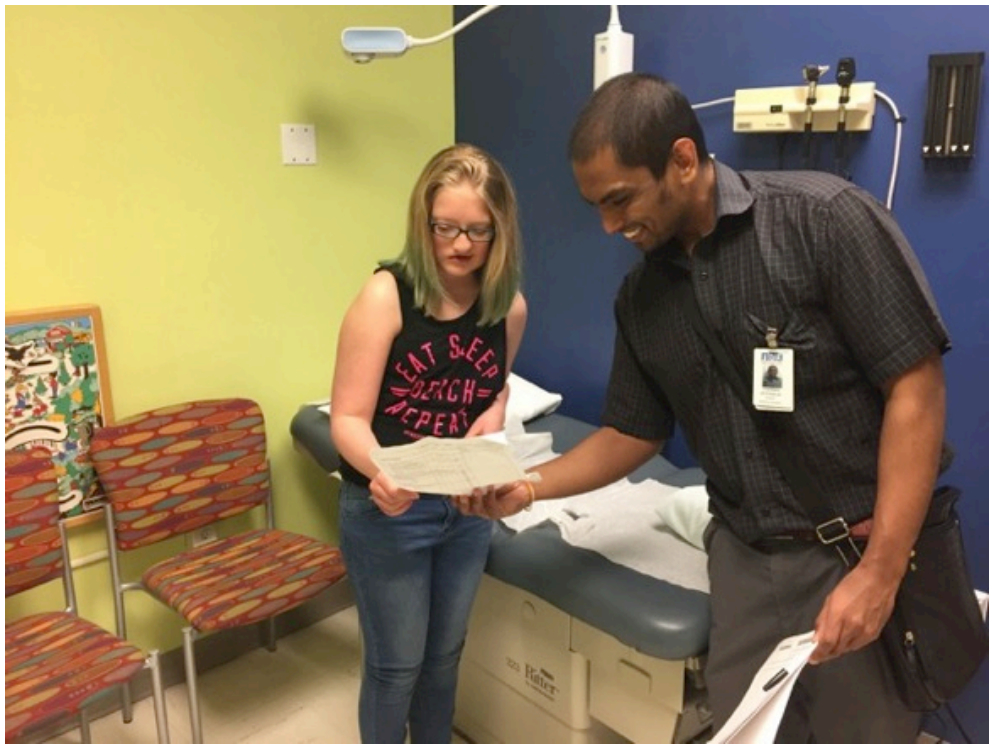
Scoring of feeding scale for CHARGE syndrome

29	Soft chewable foods (e.g. bread, crackers)	0	2
30	Tough chewable foods (e.g. meat)	0	1
31	Hard vegetables and fruit (e.g. raw apples)	0	1
Total Score (sum of all items)			**/100 total points**

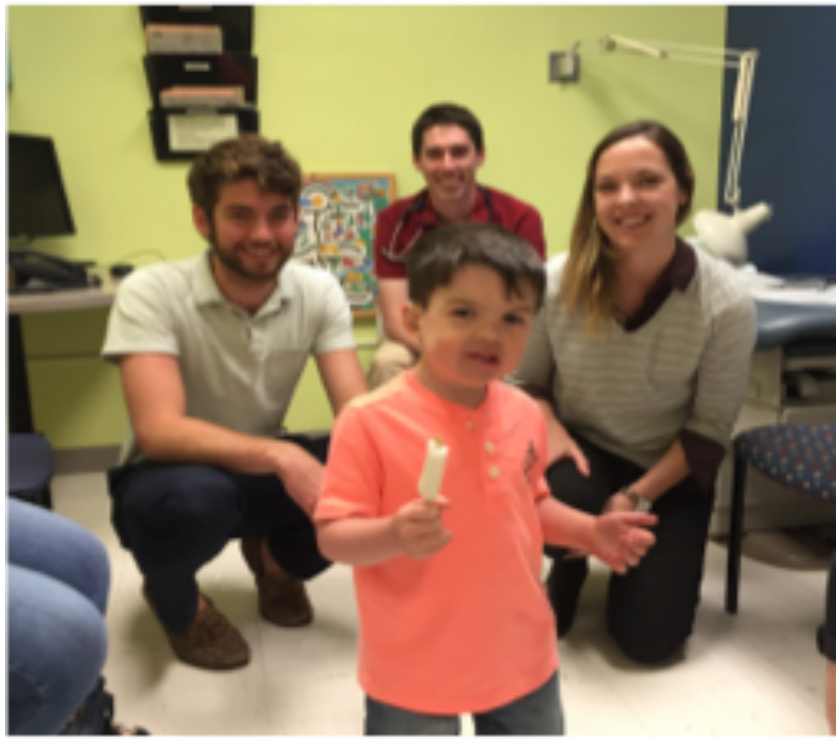
Out of 100 points
Higher score = worse feeding difficulties

Three uses for the feeding scale for CHARGE Syndrome

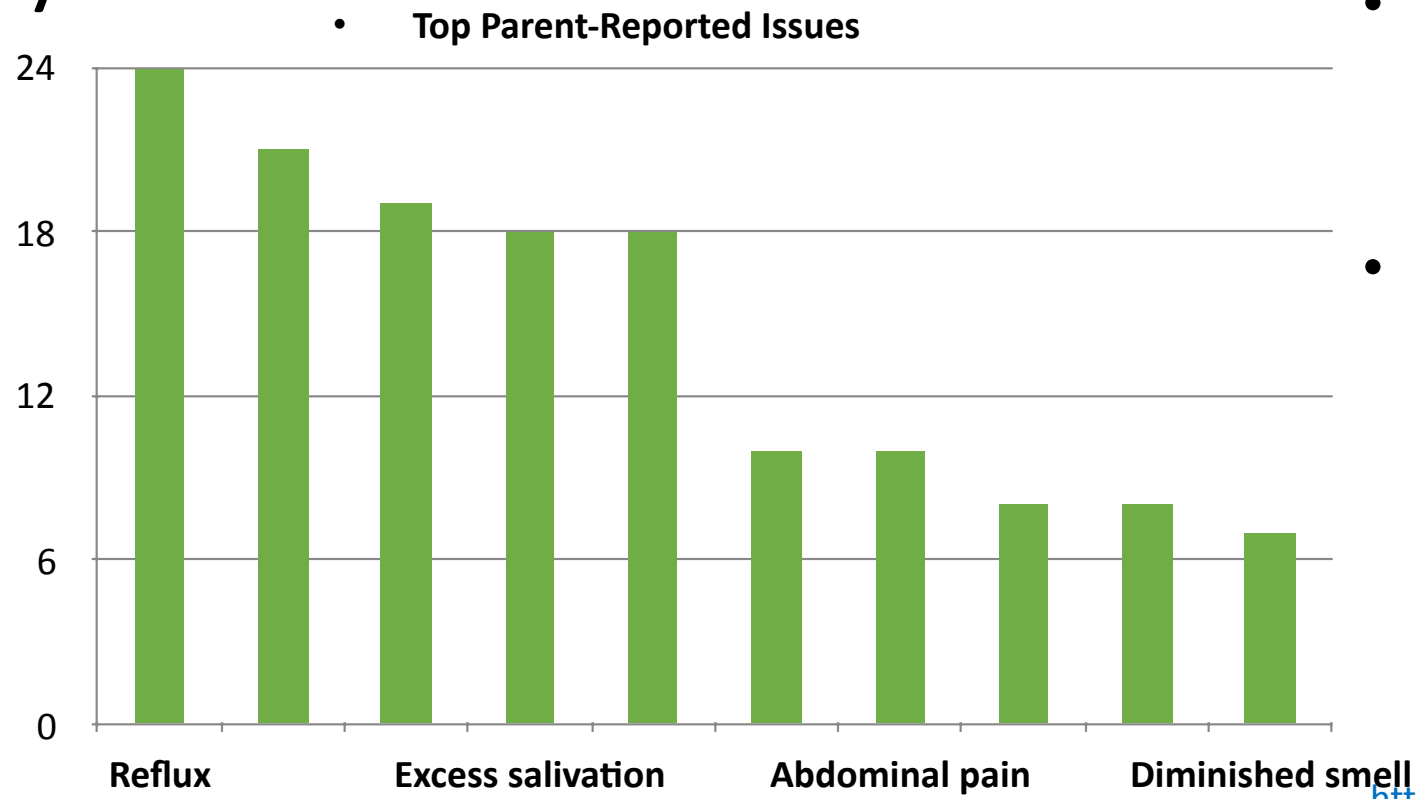
1. To assess the severity of feeding difficulties
2. To track oral feeding progress before and after interventions
3. To warn the clinician and feeding therapist of new concerns



GI and feeding difficulties in CHARGE syndrome: Treatments tried and parents' perceptions of their effectiveness



Parents' perceptions of treatments tried for gastrointestinal and feeding issues in CHARGE syndrome



- 18 males, 13 females with CHARGE syndrome
- 31 respondents (Australia, NZ, USA, UK, Sweden, Indonesia)

https://surveys.dal.ca/opinio/s?s=39536

Medications Tried

- **Excess saliva** – Glycopyrolate, Botox
- **Reflux** – Omeprazole, Esomeprazole, Ranitidine, Cisapride

- **Abdominal pain** – Acetaminophen
- **Constipation** – PEG, Osmolax, Coloxyl, Parachoc, Movicol Jr, Domperidone

Surgeries Undergone

- **Aspiration –** G-tube, tracheostomy, tracheoesophageal fistula (TEF) repair
- **Abnormal tongue movement** – frenulectomy
- **Abnormal chewing** – teeth extraction, orthodontics
- **Abnormal swallowing** – esophageal dilatation to treat esophageal stenosis
- **Reflux** – Nissen fundoplication
- **Abdominal pain** - laparoscopic investigations, emergency laparotomy for bowel obstructions

Behavioral Therapy Tried

- **Excess salivation** – Speech Language Therapy (SLT), suctioning, swallowing reminders, vital stim therapy
- **Aspiration** – limit certain foods/liquids, SLT, close supervision
- **Pocketing of food in cheeks, Overstuffing, Abnormal tongue movements, Abnormal swallow/chew** – Prompting while eating, SLT, dissolvable foods, eating with the family at mealtime
- **Reflux** – Tilt the head of the bed up, remain upright after eating
- **Abdominal pain** – Dairy-free diet, abdominal massage, warm baths, fibre
- **Constipation** – Exercise
- **Overweight –** more emphasis on whole foods and less processed foods

Treatments That Parents Think are LEAST Effective

- **Excess saliva** – Constant suctioning
- **Abdominal pain** – Acetaminophen
- **Constipation** – Movicol Jr, lactulose
- **Reflux** – Changing formula types

Treatments That Parents Think are MOST Effective

- **Excess saliva, Aspiration** – Being vertical, SLT, vitalstim therapy, botox
- **Pocketing of food, Over-stuffing, Abnormal sensation in mouth** – SLT, eating with family
- **Abnormal chewing** – Orthodontics, behavioral therapy
- **Reflux** – Fundoplication, medication
- **Abdominal pain** – Increased fibre, abdominal massage, warm baths
- **Constipation** – Less junk food, parachoc

Members of the Treatment Teams

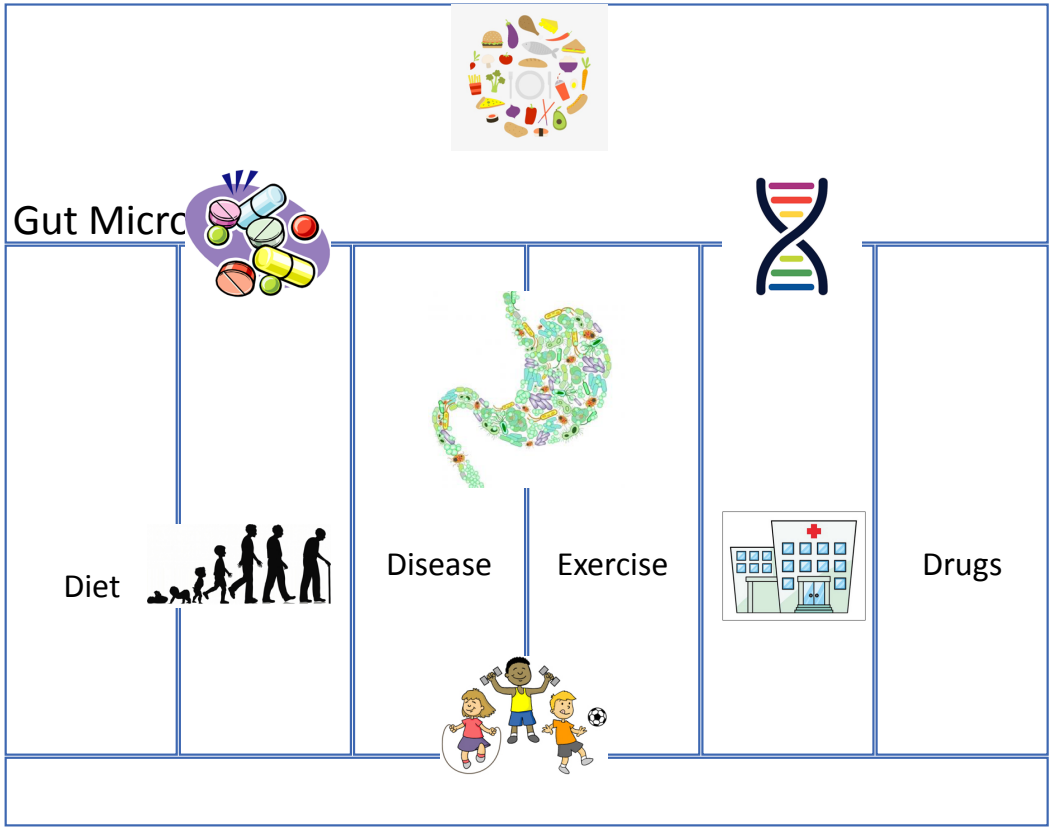
- Pediatrician
- Family physician
- Pulmonologist
- Gastroenterologist
- ENT surgeon
- General surgeon
- Physiatrist/Rehabilitation physician
- Speech language pathologist
- Occupational therapist
- Physical therapist
- Massage therapist
- Dentist
- Dietician

Microbiome

Background

- Food travels from mouth to anus through the *gastrointestinal tract (GI tract)*
- Food is digested and excreted along the way by chemicals and precise movements in the GI tract

BUT… there are also trillions of *bacteria* and other organisms that help keep our guts healthy = **GUT MICROBIOME**



Gut dysbiosis

Typical microbiome contains:

- Firmicutes
- Actinobacteria
- Bacteroidetes
- Proteobacteria

When these change in *type* or *number* and cause GI distress → **dysbiosis**

Gut dysbiosis is associated with GI disorders and extra-intestinal disorders:

- Crohn's/Colitis
- Irritable bowel syndrome
- Obesity
- Autism
- Etc….

Research Question

1) Does the gut microbiome differ in individuals with CHARGE syndrome compared to individuals who are not affected with CHARGE?

2) If so, does the change in gut microbiome correlate with the severity of GI symptoms?

3) And does the change in gut microbiome correlate with dietary factors?

Study Design

Participants: Individuals with CHARGE syndrome from the Canadian Maritimes and if possible, their sibling who is unaffected by CHARGE

- 7 individuals with CHARGE (proband)
- 4 sibling controls (subject)

Each participant provided:

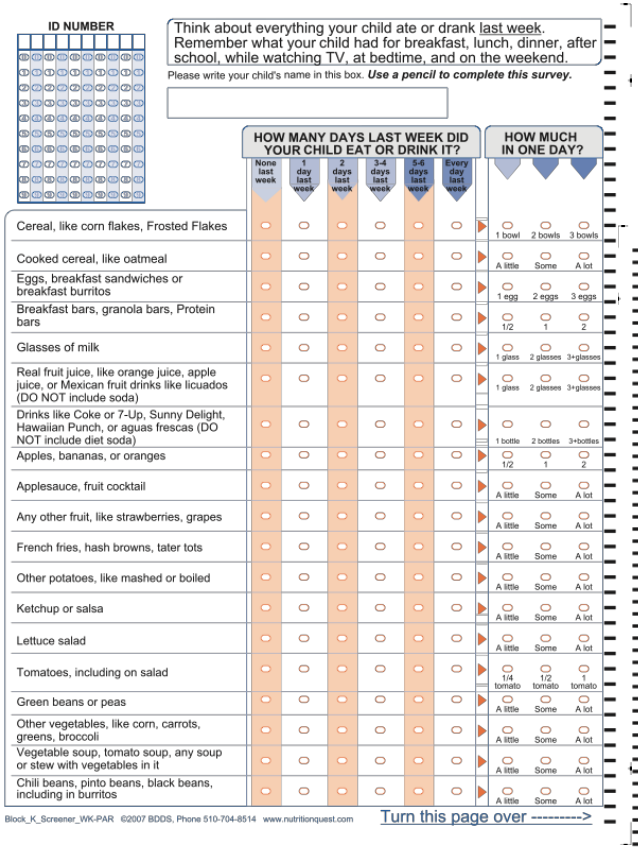
- a stool sample
- a Block Food Screener
- a PedsQL GI symptom severity questionnaire

PEDSQL GASTROINTESTIONAL SYMPTOM SCALE

Rate the following from 0 (never) – 4 (almost always)

- stomach pain
- stomach discomfort when eating
- food and drink limits
- trouble swallowing
- heartburn and reflux
- nausea and vomiting
- gas and bloating
- constipation
- diarrhea
- blood in bowel

Block Food Screener



ID NUMBER

Think about everything your child ate or drank last week. Remember what your child had for breakfast, lunch, dinner, after school, while watching TV, at bedtime, and on the weekend.

Please write your child's name in this box. ***Use a pencil to complete this survey.***

	HOW MANY DAYS LAST WEEK DID YOUR CHILD EAT OR DRINK IT? None last week	1 day last week	2 days last week	3-4 days last week	5-6 days last week	Every day last week	HOW MUCH IN ONE DAY?		
Cereal, like corn flakes, Frosted Flakes	○	○	○	○	○	○	1 bowl	2 bowls	3 bowls
Cooked cereal, like oatmeal	○	○	○	○	○	○	A little	Some	A lot
Eggs, breakfast sandwiches or breakfast burritos	○	○	○	○	○	○	1 egg	2 eggs	3 eggs
Breakfast bars, granola bars, Protein bars	○	○	○	○	○	○	1/2	1	2
Glasses of milk	○	○	○	○	○	○	1 glass	2 glasses	3+glasses
Real fruit juice, like orange juice, apple juice, or Mexican fruit drinks like licuados (DO NOT include soda)	○	○	○	○	○	○	1 glass	2 glasses	3+glasses
Drinks like Coke or 7-Up, Sunny Delight, Hawaiian Punch, or aguas frescas (DO NOT include diet soda)	○	○	○	○	○	○	1 bottle	2 bottles	3+bottles
Apples, bananas, or oranges	○	○	○	○	○	○	1/2	1	2
Applesauce, fruit cocktail	○	○	○	○	○	○	A little	Some	A lot
Any other fruit, like strawberries, grapes	○	○	○	○	○	○	A little	Some	A lot
French fries, hash browns, tater tots	○	○	○	○	○	○	A little	Some	A lot
Other potatoes, like mashed or boiled	○	○	○	○	○	○	A little	Some	A lot
Ketchup or salsa	○	○	○	○	○	○	A little	Some	A lot
Lettuce salad	○	○	○	○	○	○	A little	Some	A lot
Tomatoes, including on salad	○	○	○	○	○	○	1/4 tomato	1/2 tomato	1 tomato
Green beans or peas	○	○	○	○	○	○	A little	Some	A lot
Other vegetables, like corn, carrots, greens, broccoli	○	○	○	○	○	○	A little	Some	A lot
Vegetable soup, tomato soup, any soup or stew with vegetables in it	○	○	○	○	○	○	A little	Some	A lot
Chili beans, pinto beans, black beans, including in burritos	○	○	○	○	○	○	A little	Some	A lot

Block_K_Screener_WK-PAR ©2007 BDDS, Phone 510-704-8514 www.nutritionquest.com

Turn this page over ---------->

	HOW MANY DAYS LAST WEEK DID YOUR CHILD EAT OR DRINK IT? None last week	1 day last week	2 days last week	3-4 days last week	5-6 days last week	Every day last week	HOW MUCH IN ONE DAY?		
Refried beans	○	○	○	○	○	○	A little	Some	A lot
Hamburgers, cheeseburgers	○	○	○	○	○	○	1 small	1 large	2 large
Hot dogs, corn dogs, or sausage	○	○	○	○	○	○	1	2	3
Lunch meat like boloney, ham, Lunchables	○	○	○	○	○	○	1 slice	2 slices	3+ slices
Pizza or pizza pockets	○	○	○	○	○	○	A little	Some	A lot
Spaghetti or ravioli with tomato sauce	○	○	○	○	○	○	A little	Some	A lot
Macaroni and cheese	○	○	○	○	○	○	A little	Some	A lot
Chicken, including nuggets, wings, tenders, also in sandwiches or stew	○	○	○	○	○	○	A little	Some	A lot
Fish, fish sticks or sandwiches, tuna, shrimp	○	○	○	○	○	○	A little	Some	A lot
Burritos or tacos	○	○	○	○	○	○	1/2	1	2
Beef like roast, steak or in sandwiches	○	○	○	○	○	○	A little	Some	A lot
Meat balls, meat loaf, beef stew, Hamburger Helper	○	○	○	○	○	○	A little	Some	A lot
Pork, like chops, roast, ribs	○	○	○	○	○	○	A little	Some	A lot
Popcorn	○	○	○	○	○	○	A little	Some	A lot
Snack chips like potato chips, Doritos, Fritos, tortilla chips	○	○	○	○	○	○	A few	Small bag	Large bag
Ice cream	○	○	○	○	○	○	1 scoop	2 scoops	3 scoops
Candy, candy bars	○	○	○	○	○	○	Mini	Small	Large
Cookies, donuts, cakes like Ho-Hos	○	○	○	○	○	○	A little	Some	A lot
Cheese. Remember cheese in sandwiches or nachos with cheese or quesadillas	○	○	○	○	○	○	1 slice	2 slices	3+ slices
Whole wheat bread or rolls (NOT white bread)	○	○	○	○	○	○	1 slice	2 slices	3 slices

What kind of cereal did your child eat? (MARK ONLY ONE)

○ Plain Cheerios, Grape Nuts, Shredded Wheat, Wheaties, Wheat Chex, Kix
○ Honey Nut Cheerios, Cap'n Crunch, Lucky Charms, Life, Golden Grahams, Frosted Mini Wheats, Raisin Bran
○ Other sweet cereals, like Frosted Flakes, Froot Loops
○ Any other cereal, like Corn Flakes, Rice Krispies

What kind of milk did your child drink? (MARK ONLY ONE)
○ Whole milk ○ Reduced fat 2% milk ○ Low fat 1% milk ○ Nonfat milk ○ Chocolate milk ○ Soy milk ○ Lactaid milk ○ Don't know

Please tell us about your child

Are they ○ Male ○ Female

How old are they? ○ 2 ○ 3 ○ 4 ○ 5 ○ 6 ○ 7 ○ 8 ○ 9 ○ 10 ○ 11 ○ 12 ○ 13 ○ 14 ○ 15 ○ 16 ○ 17

Results and Discussion

In progress

What's next?

Limitations:	Small study size: hard to make inferences since the gut microbiome is highly variable
	Lack of control subjects for all participants with CHARGE
Future Study:	Repeat study with more participants
	Include parents to participate as matched control

INTERESTED IN HELPING?

Come visit us!

Study can be completed at the conference

OR

We provide you with mailing material

IWK CHARGE Clinic Students & Residents Using the CHARGE Checklist

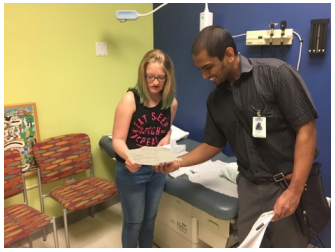
CHARGE Syndrome Checklist: Health Supervision Across The Lifespan (From Head To Toe)

*Shaded boxes indicate key assessment points

		INFANCY (0-2 years)	CHILDHOOD (3-11 years)	ADOLESCENCE (12-17 years)	ADULTHOOD (18+ years)
Genetics	Clinical diagnosis (Blake et al. or Verloes or Hale et al. criteria)				
	Genetic testing – Genetics consult (CMD/ 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 EEG				
	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 E Ophthalmology consult (dilated eye exam in infancy, vision assessments)				
	Corneal exposure – lubricating eye drops				
	Photophobia – tinted glasses, sunhat				
	Choanal atresia/cleft palate/tracheoesophageal fistula E ENT/Plastics consult				
	Audiometry and tympanometry, monitor for recurrent ear infections				
	Adaptive services for individuals with deafness/blindness				
	Cochlear implant assessment if applicable				
	Obstructive sleep apnea – monitor for tonsil/adenoid hypertrophy				
	Excessive secretions – consider Botox, medication				
	Dental issues – consider cleaning under anaesthetic				
Cardiology Respirology	Cardiac malformations common – major/minor defects, vascular ring or arrhythmias possible (echocardiogram, chest x-ray, ECG) E Cardiology consult				
	Sinusitis, pneumonia, asthma E monitor				
	Anesthesia risk (difficult intubations/postop airway obstruction/aspiration) – extensive preoperative assessment, combine surgical procedures				
Gastroenterology Genitourinary	Gastroesophageal reflux – Gastroenterology consult – consider motility agents with proton pump inhibitor				
	Poor suck/chew/swallow E feeding team assessment/intervention				
	Aspiration risk, tracheoesophageal fistula – swallowing studies				
	May need supplemental feeds – frequently requires gastrostomy tube or Gastro-jejunostomy tube				
	Constipation – consider Senna glycoside with polyethylene glycol				
	Renal anomalies – abdominal u/s +/E VCUG, blood pressure monitoring				
Endocrinology	Hypogonadotropic hypogonadism – LH, FSH by 3 months				
	Genital hypoplasia (if undescended testes E consider orchidoplexy)				
	Delayed puberty – Endocrinology consult E gonadotropin levels, MRI				
	Osteoporosis – DEXA scan				
	Poor growth – Endocrinology consult – GH stimulation test, GH therapy				
	Obesity E monitor				
	Fertility and contraception E 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) E evaluate				
Psychology Developmental	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				
	Toileting skills E support				
	Life skills/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

Tridar C, ArroßRobar A, van RavenswaaijßArts C, Blake K



Questions and Answers