

Feeding and GI Issues in Individuals with Noonan Syndrome

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Disclaimers

- Not a gastroenterologist 😊
....but I work with an excellent one: Dr. Ajay Kaul
- Still not much published
 - Several new manuscripts since 2018

Overview

- Feeding and Nutrition
- Growth
- Functional issues
 - Reflux
 - Dysmotility
 - Oral-motor skills
- Abnormal anatomy
 - Less common but important to consider

Feeding and nutrition

- At least 50% of infants and children with NS experience “feeding problems”
- 30-50% of these use a feeding tube
- Feeding usually improves with time (by age 2-4)
- Late-onset feeding problems are associated with infection/illness, post-operative status, and behavioral concerns

What are Feeding problems?

- Feeding skill dysfunction:
 - Oral/motor coordination
 - Oral aversion
 - Texture sensitivity
- Medical dysfunction:
 - Aspiration
 - Dysmotility, reflux
- Nutritional dysfunction
 - restricted intake of one or more nutrients
 - Reliance on enteral feeds or supplements
- Psychosocial

Metabolic demand

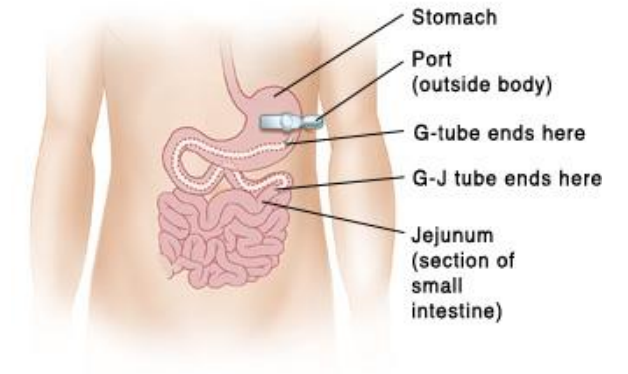
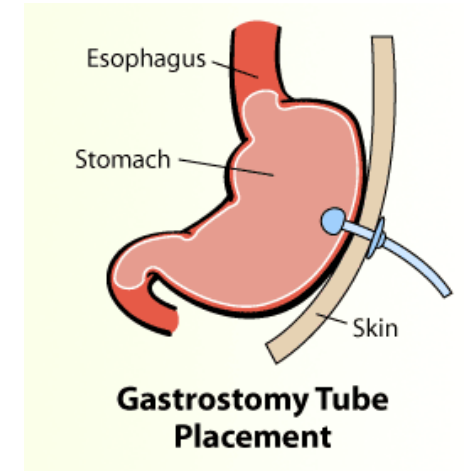
- Recommended daily allowance for caloric intake
 - For energy expenditure, and storage
 - Resting expenditure may be **INCREASED** in individuals with NS
- Other variables:
 - Illness
 - Congenital heart disease
 - Activity!
 - Other medical problems

How to manage feeding problems?

- Feeding route/content
 - Video swallow study
- Identify and treat oral motor weakness
- Identify and treat medical problems contributing
- Have realistic expectations

Feeding route and content

- Feeding route: oral, nasogastric, gastrostomy, gastro-jejunal
 - Oral (“PO”)
 - Tube: NG, GT, GJ
- Feeding schedule:
 - Bolus feed
 - Continuous
- Feeding content:
 - Concentrate formula/milk
 - Puree by g-tube (PBGT) diet (>8 months)
 - Reduces gagging, retching, and vomiting in patients with G-tubes
 - Improves oral intake



Feeding therapy

- Speech and occupational therapy
- Early Intervention programs (<3)
- Intensive outpatient or inpatient programs

Growth expectations

- Short stature is very common—separate growth curve
- Several studies have shown lower BMI in individuals with NS
- Gene-specific effects have been observed although studies have small numbers

Weight vs height

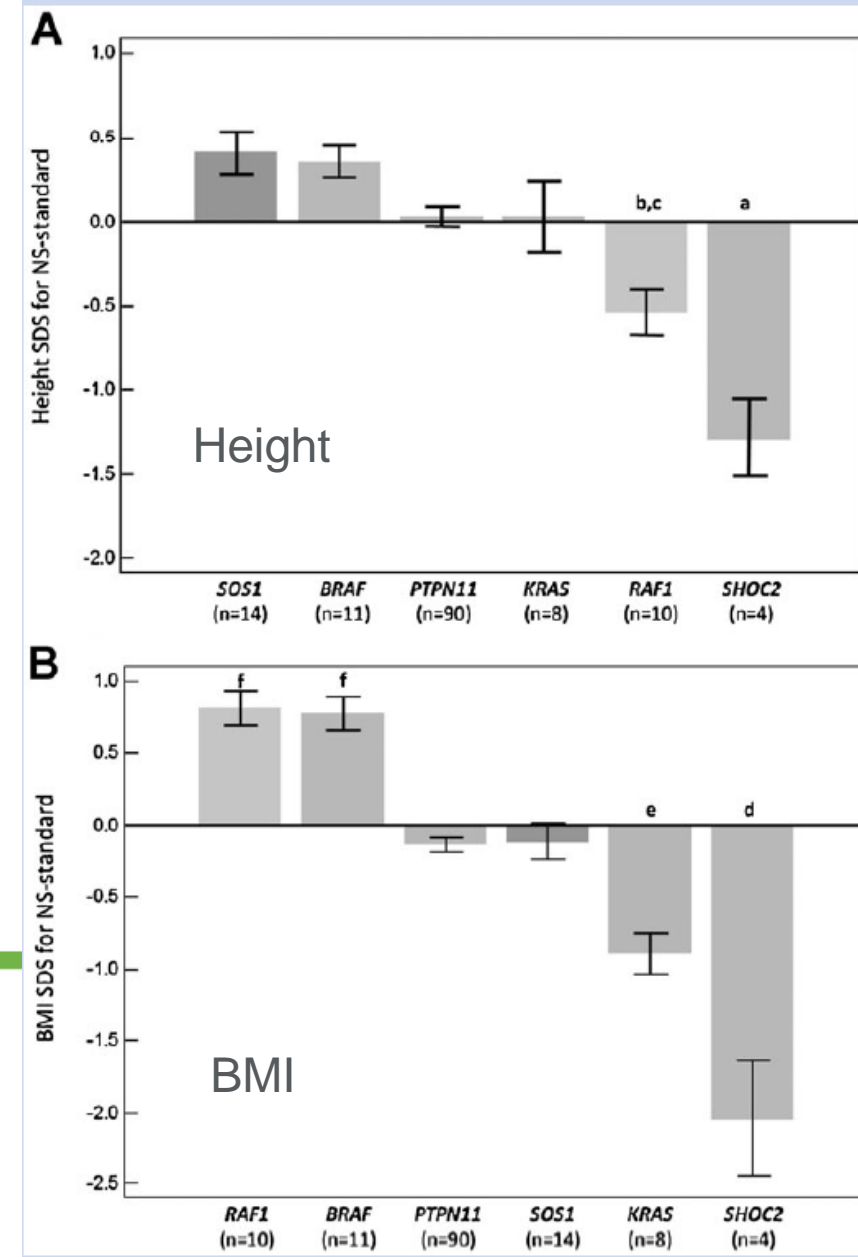
- < 2 y: “weight-for-height”
- > 2 y: body mass index
- Is weight appropriate for height?
- Is weight gain velocity appropriate?

Genotype, growth, and feeding

Table 1. Prevalence of feeding problems in the first year of life in patients with NS with the most prevalent gene mutations.

	<i>PTPN11</i>	<i>SOS1</i>	<i>RAF1</i>
Digilio et al., 2012 [9]	13/20 = 65%	0/8 = 0%	3/4 = 75%
Draaisma et al., 2020 [15]	36/63 = 57%	6/16 = 37.5%	2/4 = 50%

	<i>PTPN11</i> (n = 63)	<i>SOS1</i> (n = 16)	Other gene mutations (n = 29)
Oral diet without restrictions	23 (36%)	8 (50%)	6 (21%)
Oral diet with restrictions	17 (27%)	6 (38%)	8 (27%)
Tube-dependent feeding	23 (37%)	2 (12%)	15 (52%)



Other growth variables

- Weight Gain--Positive
 - Parent with NS
 - Higher birth weight
- Weight Gain—Negative
 - PTPN11
 - Higher gestational age
 - Feeding problems
- Length—Positive
 - Maternal height
 - Gestational age
- Length--negative
 - Cardiac surgery
 - Feeding problems

Registered Dietician consult

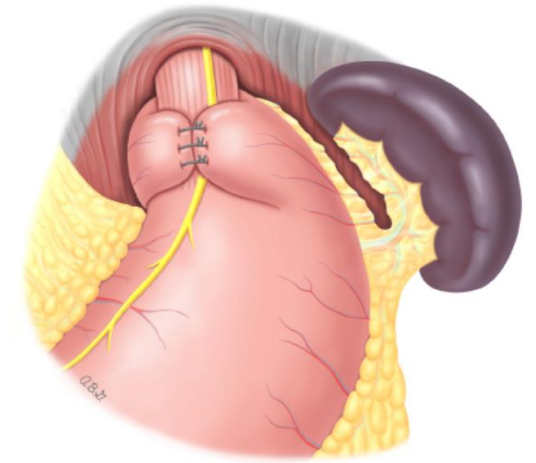
- Target goals for calories, nutrients, free water
- Reassess based on growth and age
- Right-size feeds
 - Overfeeding is also harmful

MEDICAL GI PROBLEMS

GERD

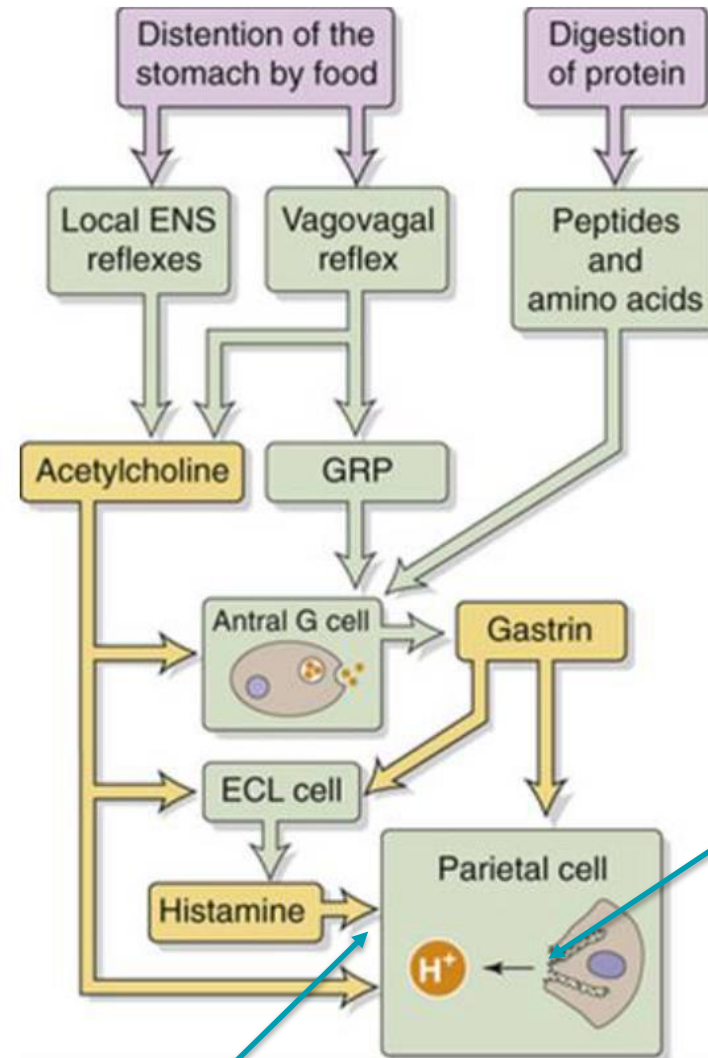
- Symptoms: vomiting, pain, poor weight gain, coughing
- Diagnosis: by symptoms, pH probe/manometry
- Upper GI study can rule-out anatomic abnormalities that might be contributing
- Medication
 - Famotidine (Histamine type 2 receptor inhibitor)
 - Omeprazole (proton pump inhibitor): block acid secretion
- (Surgical)
 - (Nissen fundoplication)

Nissen fundoplication



The Nissen fundoplication provides a 360° "wrap" or collar around the esophagus. Mobilization of the fundus is generally accomplished by dividing the short gastric vessels to the spleen.

GERD treatment



Proton pump inhibitor
(omeprazole)

H2 Histamine Receptor
Blocker (famotidine)

Dysmotility and NS: 1 (old) paper

- Shah et al 1999: 25 kids with clinical diagnosis of NS
- 5 of 25 had surface electrogastrography
 - Delayed gastric emptying
- 4 of 5 had antroduodenal manometry
 - Immature contractile activity

Dysmotility

- Gastroparesis: Delayed gastric emptying of solids *without* a mechanical obstruction
 - Symptoms: nausea, vomiting, early satiety (feeling full), pain
- Treatment: Medication, Feeding route/strategy

Diagnostic tests

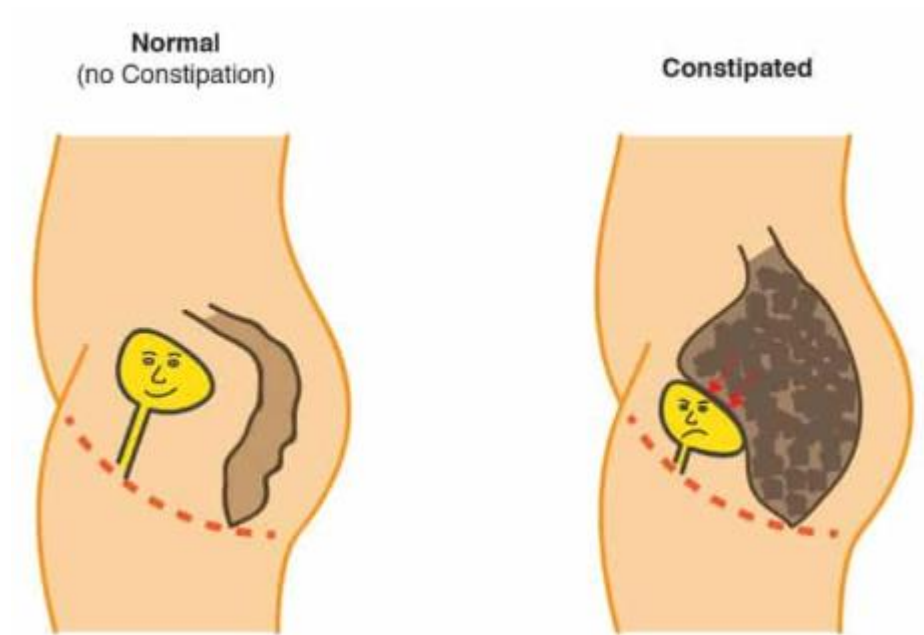
- Gastric emptying scan
 - How much food is left 1, 2, and 4 hours after eating
- Manometry
 - Measures pressure in intestines in order to quantify strength and pacing of contractions
 - Can identify small bowel dysmotility

Gastroparesis--treatment

- Diet:
 - GJ feeding (continuous)
 - Puree by g-tube
- Medication
 - treat reflux (delayed emptying exacerbates)
 - Pro motility agent (erythromycin, ciproheptadine)
- Pyloric botox and balloon dilation

Constipation

- Definition: <2 stools per week, or painful stooling, or excessive straining with hard stool
 - Chronic > 3 months
 - Can impact bladder continence
 - Can lead to overflow stool incontinence
- Interventions:
 - Laxatives: osmotic
 - Osmotic: Miralax, Lactulose
 - Stimulant: Senna, Bisacodyl
 - Suppository-Glycerin
- “Clean out”



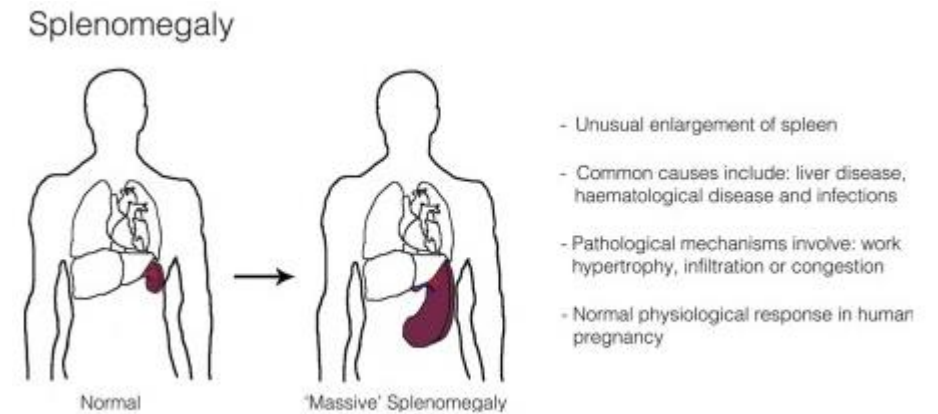
Prokinetics

- Prucalopride: serotonin receptor agonist
 - Enhances bowel motility
 - (off label in pediatrics)
 - Chronic constipation

Anatomical considerations

- Lymphangiectasia
- Malrotation
- Hiatal hernia
- Pyloric stenosis

- Splenomegaly (not feeding related, but a documented phenomena)



Conclusions

- GI problems are very common in individuals with NS
- Feeding issues are the most prevalent and often coexist with medical problems
- Generally they improve with age
- Multidisciplinary management is ideal
 - GI, speech/occupational therapy, RD, psychology (older kids)

References

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Feeding difficulties and foregut dysmotility in Noonan's syndrome

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RESEARCH ARTICLE

AMERICAN JOURNAL OF
medical genetics PART
A

Growth Standards of Patients With Noonan and Noonan-Like Syndromes With Mutations in the RAS/MAPK Pathway

Alexandra C. Malaquias,^{1,2*} Amanda S. Brasil,³ Alexandre C. Pereira,⁴ Ivo J.P. Arnhold,² Berenice B. Mendonca,² Debora R. Bertola,³ and Alexander A.L. Jorge^{1,2*}

2012

Nutritional Aspects of Noonan Syndrome and Noonan-Related Disorders

Fernanda Marchetto da Silva,^{1*} Alexander Augusto Jorge,² Alexandra Malaquias,² Alexandre da Costa Pereira,³ Guilherme Lopes Yamamoto,¹ Chong Ae Kim,¹ and Debora Bertola¹

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ORIGINAL ARTICLE

WILEY AMERICAN JOURNAL OF
medical genetics PART
A

First-year growth in children with Noonan syndrome: Associated with feeding problems?

Ellen A. Croonen¹ | Jos M. T. Draaisma¹ | Ineke van der Burgt² | Nel Roeleveld^{1,3} | Cees Noordam¹

European Journal of Pediatrics (2020) 179:1683–1688
<https://doi.org/10.1007/s00431-020-03664-x>

ORIGINAL ARTICLE



Young children with Noonan syndrome: evaluation of feeding problems

Jos M. T. Draaisma¹ | Joris Drossaers¹ | Lenie van den Engel-Hoek² | Erika Leenders³ | Joyce Geelen¹

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Journal of
Clinical Medicine

Review

Feeding Problems in Patients with Noonan Syndrome: A Narrative Review

Dagmar K. Tiemens^{1,2}, Leenke van Haaften³, Erika Leenders⁴, Annemiek M. J. van Wegberg⁵, Bregtje Gunther Moor⁶, Joyce Geelen¹ and Jos M. T. Draaisma^{1,*}

2020

