

## *Gut Problem and Noonan Syndrome*

Noonan Syndrome Seminar 2016  
For Genetics Alliance Australia  
Garvan Institute

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## *Disclaimer / Apologies*

- SMH article
  - Avoid all PowerPoint presentation
- Level 1 evidence in children especially with complex disease – not possible
- No financial conflict of interest

## *Noonan Syndrome*

- No common associated GI issues
  - Rarely lymphatic dysplasia
- Postnatal growth reduction
  - Short stature
  - Improvement in cases with GH

## *Noonan Syndrome Management*

- Maximize nutrition and growth
- Other more commonly inherited gut problem
  - GOR, constipation, Cow's Milk Protein Allergy
- Other common acquired conditions affecting feeding, sleeping and growth
  - OSA, Iron deficiency, unstable bladder

## *Genetics of Common Gut Problem*

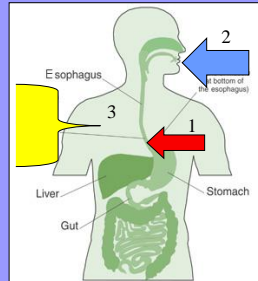
- “Slow motility” – common
- ~ 1:4
- Likely autosomal dominant
- Infant – GOR, CMP enterocolitis
- Children – GOR and chronic constipation tendency
- Adult – complications of chronic GOR, constipation
- Other inherited variables
  - size of hiatal hernia, ? redundant (long) colon

## *Slow Motility*

- *At birth – classically unsettled due to GOR and CMP/Soy protein allergy*
- *Often labelled as “colic”*
- *After 1 tendency to suffer from ongoing GOR and Constipation*

## Types of Reflux

1. Distal oesophagus
2. LPR
3. Mixed



## History

- Nothing to do with:-
  - Amount of vomiting
  - Failure to thrive
  - Unsettledness
  - Level of lack of response to Mylanta, “zantac” or “losec”

## History

- Reflux at night
  - frequent waking (worse in infants and older pts with OSA)
  - wakes and demands for drinks at night (milk)
  - hates breakfast (morning nausea - prefers only drinks in morning)
  - extreme “hunger pain” dyspepsia
  - chronic throat clearing coughs (without URTI / asthma / OSA)
- LPR reflux
  - *rattly wheezy chest*
  - *recurrent unexplained croup*
  - *hoarseness*
  - worsening of asthma (despite decent treatment)
  - halitosis
  - *enamel erosion*

## History

- Oesophagitis
  - Coffee-ground vomit (rare)
  - Prefers softer & mashed food (common)
  - Hates lumpy food (ongoing meat texture aversion, gagging)
  - Prefers liquid (milk or yogurt as “mylanta”)
  - Feels food getting caught (washes down with liquid)
    - > *TEXTURE & ORAL SENSORY AVERSION*
    - *Worse with development of large tonsils*

## History

- Oesophageal
  - Lower chest pain
  - sudden sharp debilitating pain
  - sudden feeling of dysphagia
  - “sliding hiatal hernia” pain
  - relates to physical activity too soon post-prandially

## Observations

- Waterbrash
- Enamel
- Restless legs & Periodic Limb Movement Disorder
  - From low iron store (ferritin < 50)
- Sleep disorder symptoms

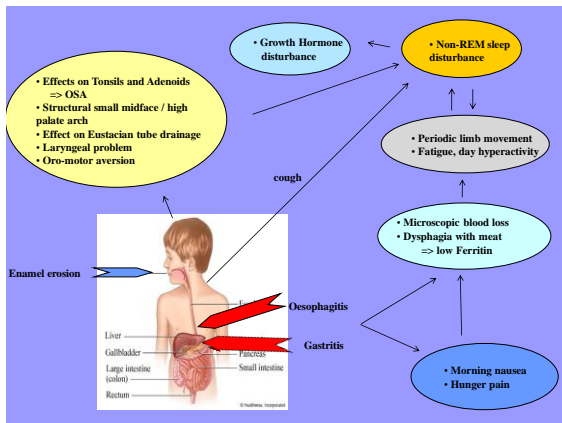
## Observations – ENT & Sleep

- Snoring
- Increased work of breathing
- Dark rings under eyes
- Unrefreshed sleep
- Mouth Breathing
  - Nasal blockage
    - Allergy
    - Adenoids
    - Turbinet
    - High arch palate

*OSA leads to increased secondary reflux at night*

## Associated Bile reflux

- Supine position allows bile reflux back into stomach and higher
- Pylorus is not a one-way valve
- Bile (alkaline) reflux worse than acid
- Bile gastritis creates more hunger pain and Iron problem
  - Gastritis



## Reasons for reflux

1. Oesophageal motility / clearance
2. LES tone
3. Hiatal Hernia
4. Gastric Motility
5. Downstream - motility/inflammation
6. ? Hypermobile Joint Syndrome



## AIM of Treatment

- Improve QOL (child & parent)
- Prevent complications
  - Anaemia
  - Fe deficiency (Concentration, learning, Restless leg, REM, growth)
  - ENT (T's, A's, Middle ear, vocal cord, airways)
  - enamel erosion
  - orthodontic issues
  - feeding aversion & speech development
  - sensory integration disorder
  - obesity

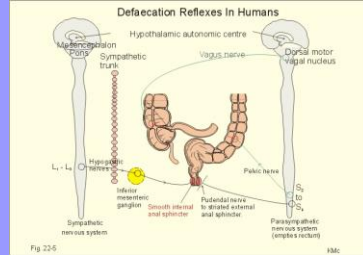
## AIM of Treatment

- Prevent complications (cont')
  - oesophageal complications
    - strictures, metaplasia
  - Development of allergic conditions ?
    - Eosinophilic Oesophagitis (EoE)
    - Coeliac disease ?
    - other protein allergy

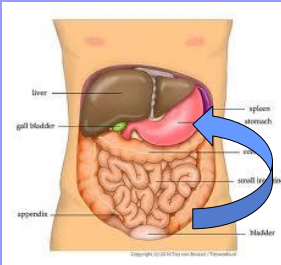


## Reflexes

- Gastro-colic reflex

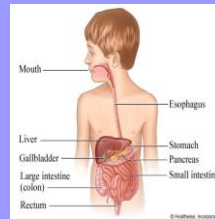


## Gastro-colic Brake



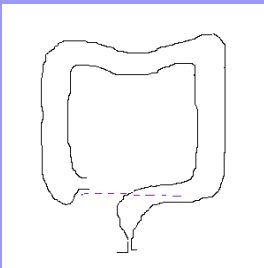
- Faecal loading leads to delay gastric emptying
- Compresses duodenum leading to more bile reflux (gastritis)

### Effects of faecal loading



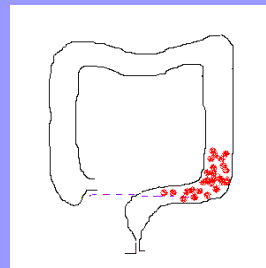
- Pain with peristalsis
- Soiling with associated psychological damage
- Urinary infections
- Unstable bladder
- Emuresis

## PHYSICAL EXAMINATION



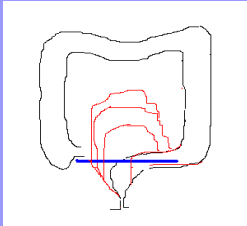
- Technique
- Light and deep palpation

## PHYSICAL EXAMINATION



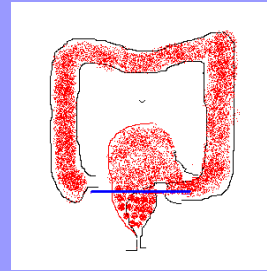
- Normal

*PHYSICAL EXAMINATION*

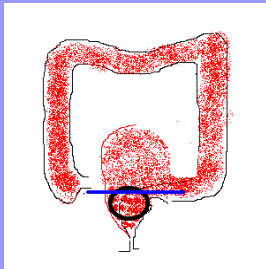


- "Obstetric examination"

*PHYSICAL EXAMINATION*

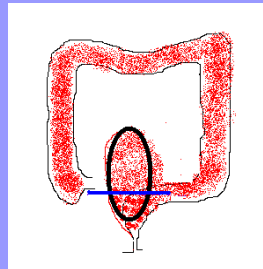


*PHYSICAL EXAMINATION*



- Variable sized stool
- pebbles

*PHYSICAL EXAMINATION*



- Variable sized stool
- Large diameter stool
- large flat pancake

*PHYSICAL EXAMINATION*



- Variable sized stool
- overflow diarrhoea

*Faecal Loading*

- Pain with peristalsis
- Fluffs
- Marks on pants (soiling)
- Funny posture
- Bloating from evening
- Poor behavior
- Poor appetite



- Disimpaction is paramount
- Home treatment with twice a day stool softener (PEG 3350)
  - Then long term daily maintenance after school

## *Constipation – poor definition*

- Defined by frequency (can be incorrect)
- Bristol chart confusing
- Faecal loading much better definition
  - Variable stooling
  - Pain / behavior change with peristalsis
  - Soiling
  - Difficulty toilet training
- Important to treat downstream to help upstream

## *Treatment of faecal overload*

- Posture / Positioning
- Timing of toileting (Signals)
- Stool softener
  - Lactulose (infants)
  - PEG 3350 (Movicol, Osmolax, ClearLax, VivaLax)
- Disimpaction and Maintenance
- Rectal treatment (Rare)
- Nasogastric washout (Rare)

## *Treatment of significant faecal overload*

- Avoid
  - high fibre (Dehydrated bulk)
  - Paraffin in aspiration risk
  - stimulant (Senekot) to avoid excessive pain & long term dependency
- Need
  - Soluble fibre
  - Fluid
  - No holding on (maturity)
- Prevention
  - Long term complications

## *Summary*

- Growth, Nutrition, Sleep and Quality of Life as priority
- Issues to manage
  - GIT
    - OSA
    - Iron
    - Orthodontic
    - Allergy
  - Bladder
- Always assume multiple pathology present

