Acid related problems in CdLS: medical and surgical management and feeding tubes

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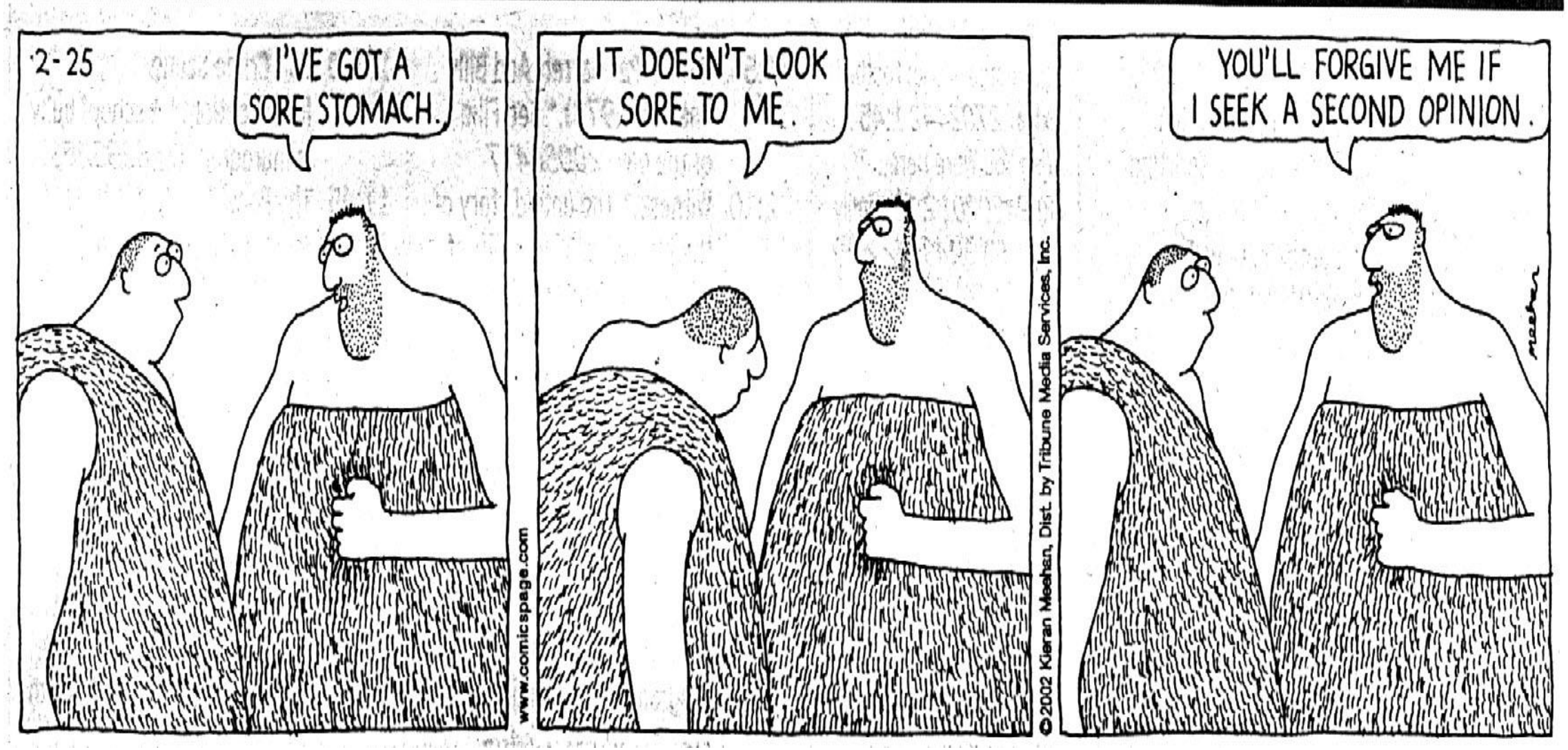
CdLS USA CONFERENCE 2020

Family Interaction with healthcare…..sometimes very frustrating !!

Meehan Streak by Kieran Meehan




I HAVE NO IDEA WHAT'S WRONG WITH YOU !
I DON'T UNDERSTAND.
3-21
I MAY BE A DOCTOR, JEFFREY, BUT I'M ONLY HUMAN...
I DON'T PRETEND TO KNOW ALL THE ANSWERS !
COULD I SEE A DOCTOR WHO DOES PRETEND TO KNOW ALL THE ANSWERS ?

Education – parents informing health care and vice versa

GI problems are a major issue !!

2018 Consensus Guidelines

nature REVIEWS GENETICS

SUPPLEMENTARY INFORMATION

In format as provided by the authors

Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement

Antonie D. Kline, Joanna F. Moss, Angelo Selicorni, Anne-Marie Bisgaard, Matthew A. Deardorff, Peter M. Gillett, Stacey L. Ishman, Lynne M. Kerr, Alex V. Levin, Paul A. Mulder, Feliciano J. Ramos, Jolanta Wierzba, Paola Francesca Ajmone, David Axtell, Natalie Blagowidow, Anna Cereda, Antonella Costantino, Valerie Cormier-Daire, David FitzPatrick, Marco Grados, Laura Groves, Whitney Guthrie, Sylvia Huisman, Frank J. Kaiser, Gerritjan Koekkoek, Mary Levis, Milena Mariani, Joseph P. McCleery, Leonie A. Menke, Amy Metrena, Julia O'Connor, Chris Oliver, Juan Pie, Sigrid Piening, Carol J. Potter, Ana L. Quaglio, Egbert Redeker, David Richman, Claudia Rigamonti, Angell Shi, Zeynep Tümer, Ingrid D. C. Van Balkom and Raoul C. Hennekam

https://doi.org/10.1038/s41576-018-0031-0

The Patient this card refers to has

Cornelia de Lange Syndrome

General information

Cornelia de Lange Syndrome (CdLS) is characterised by intellectual disability, typical facial features, upper limb anomalies, growth disturbances, and a large variety of other signs and symptoms. It can be caused by pathogenic variants in one of six genes, the most common one being NIPBL.

Cornelia de Lange Syndrome

Emergency Care Card

Fold 2

CdLS Emergency Care Card

Patient Details

Name_____

DoB_____ Gender_____

Address_____

_____

_____

Phone_____

Family Doctor Details

Name_____

Practice/Clinic Name_____

Address_____

_____

_____

Phone_____

Email_____

Last Updated (__/__/__)

Emergency Contacts

Name_____

Relation to Patient_____

Address (if different)_____

_____

_____

Phone_____

Email_____

Fold 3

Name_____

Relation to Patient_____

Address (if different)_____

_____

_____

Phone_____

Email_____

Health Care Professionals Information about Cornelia de Lange Syndrome (CdLS)

Main medical problems in CdLS

- Short stature (specific growth charts available)
- Microcephaly
- Long term feeding difficulties
- Developmental delay/Intellectual disability
- Speech problems
- Behavioural problems, especially self-injurious behaviour
- Severe recurrent gastro-oesophageal reflux
- Constipation
- Small hands, missing fingers to absent fingers
- Hearing loss
- Ptosis, recurrent blepharitis, myopia
- Cryptorchidism
- Cutis marmorata, hirsutism

Acute life-threatening complications in CdLS

- Bowel obstruction, volvulus
- Aspiration pneumonia (gastro-oesophageal reflux/swallowing difficulties)
- Seizures
- Cardiac problems
- Bladder infections
- Retinal detachment
- Small airways (anaesthesia risk)

Less frequent medical problems in CdLS

- Heart malformations (ventricular septal defect, pulmonary stenosis)
- Diaphragmatic hernia
- Seizures
- Intestinal malrotation, duodenal atresia, annular pancreas
- Perthes disease, hip dislocations
- Scoliosis
- Barrett esophagus
- Renal malformations
- Immunological problems
- Dental crowding, caries
- Nystagmus, strabismus
- Cleft palate

Further information on CdLS can be obtained from World Federation of CdLS Support Groups

www.cdlsworld.org

Using your CdLS Emergency Care Card

The CdLS Emergency Care Card enables the multi-disciplinary team necessary for best management of CdLS to be aware of the actions of other specialists. As well as filling out personal and medical details, carers should record details every time the patient sees a specialist. By listing the contact details and the date of visit, other specialists can get in touch. By listing medications, your doctors can be confident that there are no conflicting treatments being recommended. Listed specialists may include: geneticist, gastroenterologist, cardiologist, neurologist, dentist, optician, psychologist, occupational therapist, anaesthetist, paediatrician, social worker, surgeon and speech therapist. Carers should make sure they indicate the speciality of the professional.
When the card has been filled-up, extra copies can be downloaded from www.cdlsworld.org

My problems…..

- Gastro-oesophageal reflux and GERD*
- CMPA*
- Swallowing problems and choking and aspiration- chest issues
- Pain *
- CVS / motility problems*
- Surgical issues – fundoplication and feeding tubes
  - Overlap with nutritional issues – dietitians are key players

Cyclical Vomiting Syndrome

European Journal of Pediatrics (2018) 177:1435–1442
https://doi.org/10.1007/s00431-018-3218-7

REVIEW

Managing cyclic vomiting syndrome in children: beyond the guidelines

B U.K. Li[1,2,3]

Received: 11 May 2018 / Revised: 17 July 2018 / Accepted: 18 July 201
© The Author(s) 2018

Journal of Pediatric Gastroenterology and Nutrition
47:379–393 © 2008 by European Society for Pediatric Gastroenterology, Hepatology, and Nutrition and North American Society for Pediatric Gastroenterology, Hepatology, and Nutrition

Clinical Report

North American Society for Pediatric Gastroenterology, Hepatology, and Nutrition Consensus Statement on the Diagnosis and Management of Cyclic Vomiting Syndrome

*B U.K. Li, †Frank Lefevre, ‡Gisela G. Chelimsky, §Richard G. Boles, ||Susanne P. Nelson, ¶Donald W. Lewis, #Steven L. Linder, **Robert M. Issenman, and *Colin D. Rudolph

**Medical College of Wisconsin, Milwaukee, †Northwestern University, ‡Case Western Reserve University, Cleveland, OH, §Children's Hospital of Los Angeles, Los Angeles, CA, ||Children's Gastroenterology Specialists, Glenview, IL, ¶Children's Hospital of the King's Daughters, Norfolk, VA, #Dallas Pediatric Neurology Associates, Dallas, TX, and **McMaster University, Hamilton, ON, Canada*

CVSA
Cyclic Vomiting Syndrome Association

Hope Starts Here

Home | About | News & Events | For Patients | Health Providers | Resources | Research | International | Get Involved | Search

Chandra Wilson talks about her daughter's CVS

Watch the Full Episode

PEN People Entertainment Weekly NETWORK

CVS
Cyclical Vomiting Syndrome

Navigation

- Home
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- Symptoms
- Diagnosis
- Treatment
- Research
- Adult CVS
- Membership/CVSA
- Contacts
- Experiences
- Resources
- Purchase
- Thanks
- Family Day
- Survey

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Tweets by @CVSAUK

Welcome to CVSA UK

We are a registered charity providing information and support to sufferers of Cyclical Vomiting Syndrome and their families.

HON @ CODE CERTIFIED 01/2016

Please remember to update us if your email address changes.

Family Day 2018

Saturday 10th November

Birmingham Children's Hospital

Put it in the diary now - all welcome - free to attend - free creche

Supporting CVS

Grace Carmichael is raising funds for CVS by taking part in a Fun Run in Glasgow. Please support her and ask your friends and family to do the same

You can sponsor her here

Jo Hughes is walking the 46miles from Ediburgh to Glasgow to raise funds for CVSA.

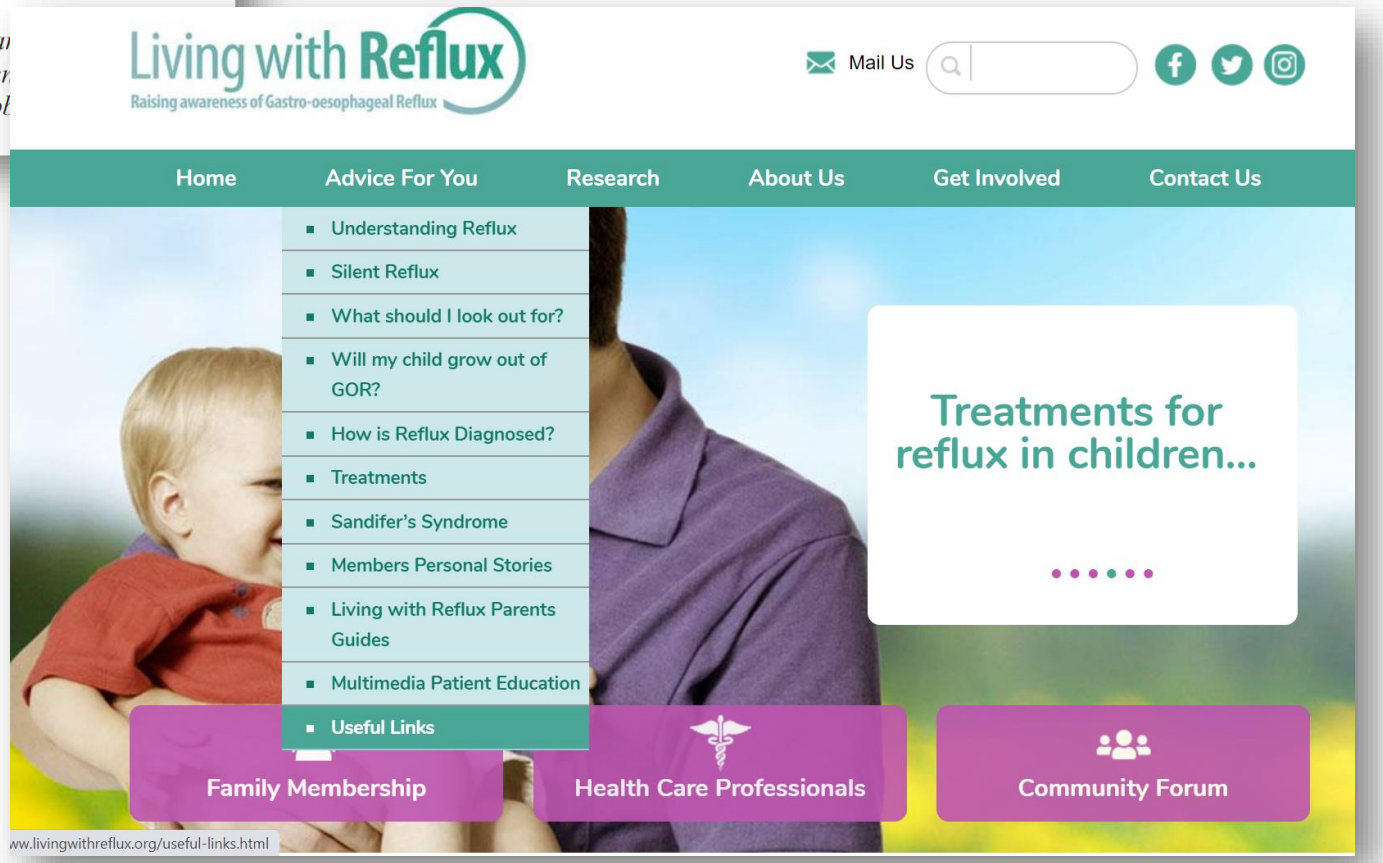
You can sponsor her here

Reflux in children….mostly physiological (normal)

Society Paper

Pediatric Gastroesophageal Reflux Clinical Practice Guidelines: Joint Recommendations of the North American Society for Pediatric Gastroenterology, Hepatology, and Nutrition and the European Society for Pediatric Gastroenterology, Hepatology, and Nutrition

*Rachel Rosen, †Yvan Vandenplas, ‡Maartje Singendonk, §Michael Caba
||Carlo DiLorenzo, ¶Frederic Gottrand, #Sandeep Gupta, **Miranda Langer
††Annamaria Staiano, ‡‡Nikhil Thapar, §§Neelesh Tipnis, and ‡Merit Tabl



Reflux in babies

Babies often bring up milk during or shortly after feeding – this is known as possetting or reflux.

It's different from vomiting in babies, where a baby's muscles forcefully contract.

Reflux is just your baby effortlessly spitting up whatever they've swallowed.

It's natural to worry something is wrong with your baby if they're bringing up their feeds. But reflux is very common and will usually pass by the time your baby is a year old.

This page covers:

Signs and symptoms of reflux in babies

Heartburn and gastro-oesophageal reflux disease (GORD)

Introduction

Gastro-oesophageal reflux disease (GORD) is a common condition, where acid from the stomach leaks up into the oesophagus (gullet).

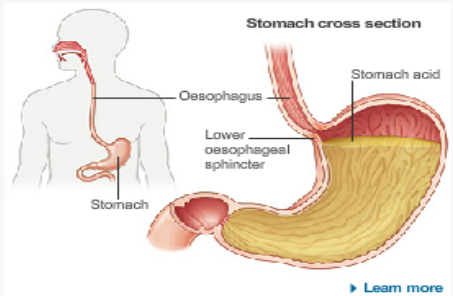
It usually occurs as a result of the ring of muscle at the bottom of the oesophagus becoming weakened. Read more about the causes of GORD.

GORD causes symptoms such as heartburn and an unpleasant taste in the back of the mouth. It may just be an occasional nuisance for some people, but for others it can be a severe, lifelong problem.

GORD can often be controlled with self-help measures and medication. Occasionally, surgery to correct the problem may be needed.

This topic focuses on GORD in adults. There are separate topics on reflux in babies and heartburn in pregnancy.

This page covers:



Media last reviewed:
Next review due:

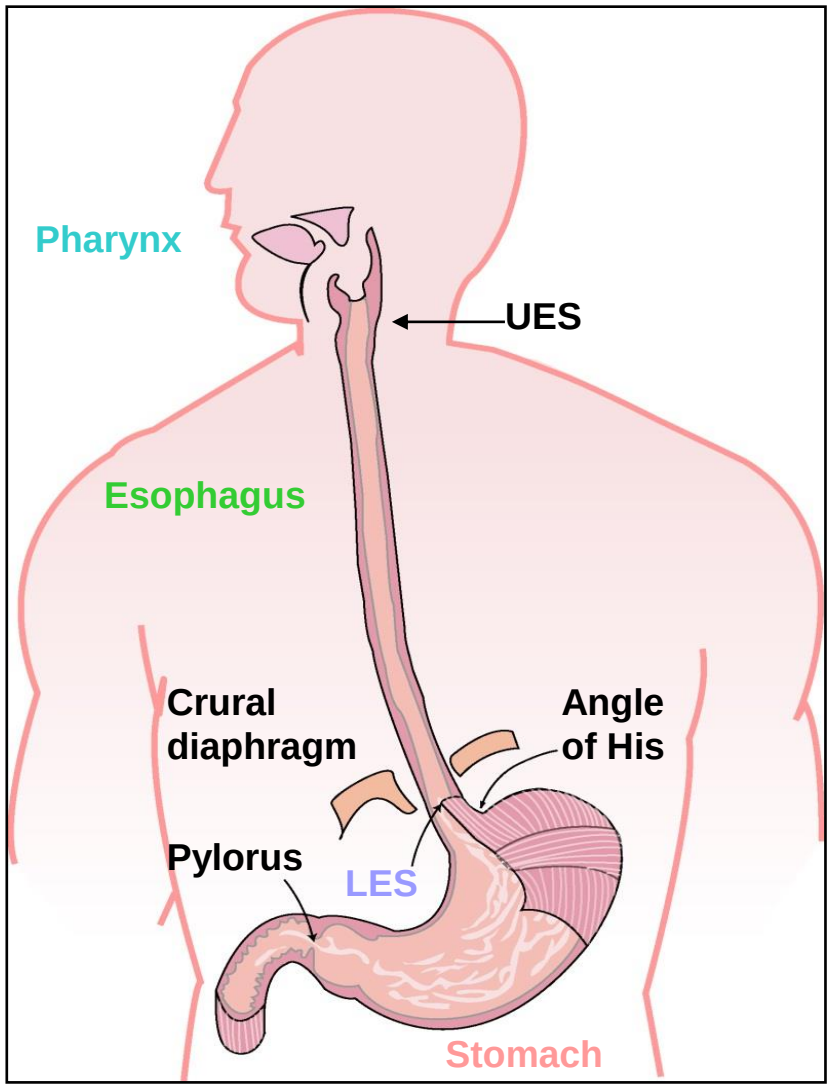
Useful links

NHS Choices links

Indigestion and heartburn in pregnancy
Reflux in babies
Indigestion

Bottle feeding advice

Pathogenic Factors in GORD (GERD)



Mechanisms of GER

- Transient LOS relaxation
- ↑ Intra-abdominal pressure
- Reduced esophageal capacitance
- ↓ Gastric compliance
- Delayed gastric emptying

Mechanisms of Esophageal Complications

- Impaired oesophageal clearance
- Defective tissue resistance
- Noxious composition of refluxate

Mechanisms of Airway Complications

- Vagal reflexes
- Impaired airway protection

Prevalence of GER(D) in the ‘regular’ population’

- GER 50% infants < 3/12

  5% of 10-12 month olds

Nelson SP , Arch Pediatr Adolesc Med 1997

- GERD - pathological
  - Heartburn, regurgitation, dysphagia

    GERD symptoms in 33% of 14 -18 year olds

    smoking and alcohol

Ramesh P, NASPGHAN Orlando 2001

- Obesity
- US adults at least 20% GORD prevalence

Presenting Symptoms and Signs of GERD

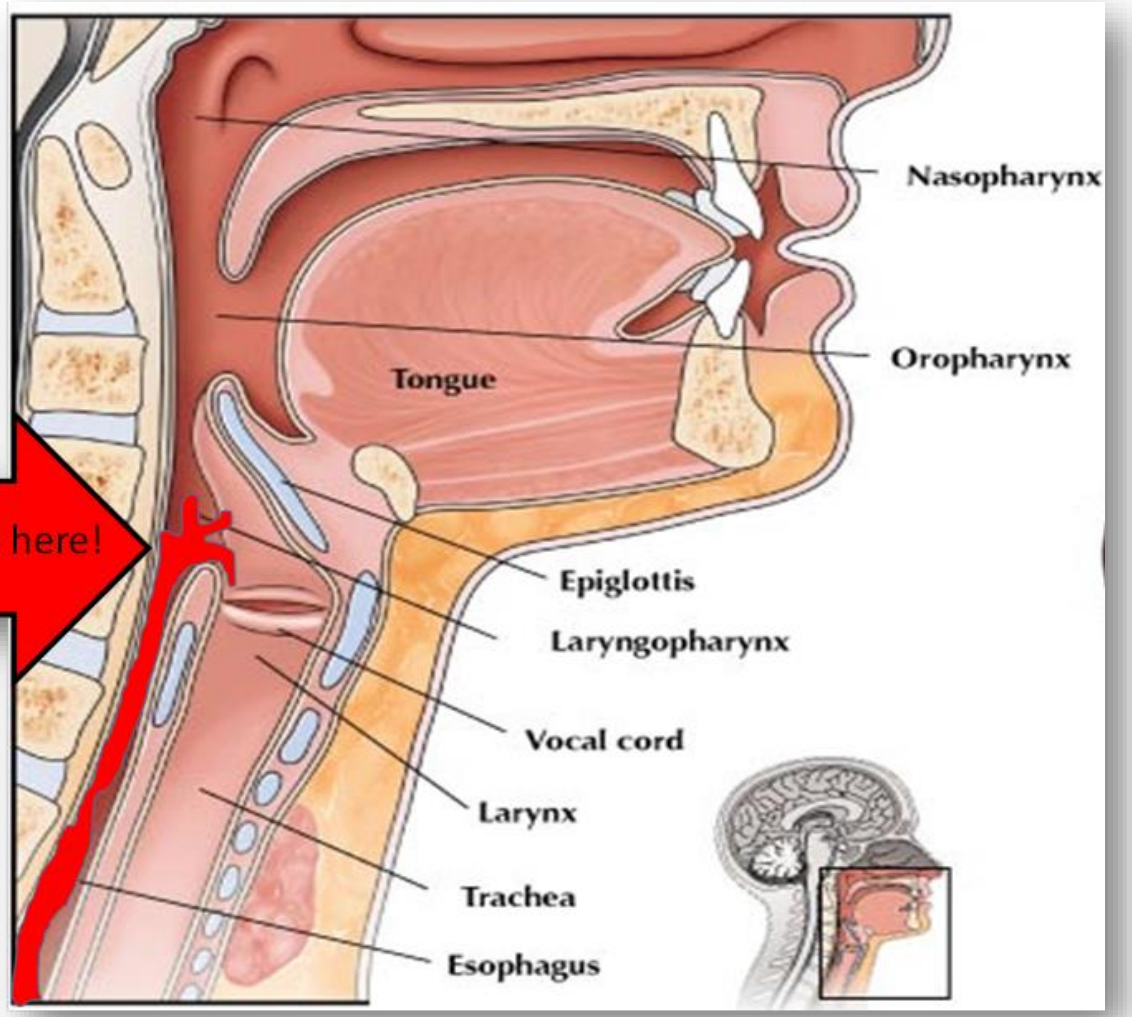
- Recurrent vomiting in infant
- Recurrent vomiting and poor weight gain in infant
- Recurrent vomiting and irritability in infant
- Feeding refusal
- Recurrent vomiting in older child

- Heartburn in child/adolescent
- Oesophagitis
- Dysphagia and choking
- Breathing problems or ALTE
- Asthma

- **Recurrent chest infections**
- **Upper airway symptoms / cough**
- **Abnormal posturing**
- **Abnormal behaviour / agitation**
- **Self Injury / harm / hitting out**
- **Teeth grinding**

The GI tract and Airway interface……cough/ choke

YIKES! Reflux all the way up here!



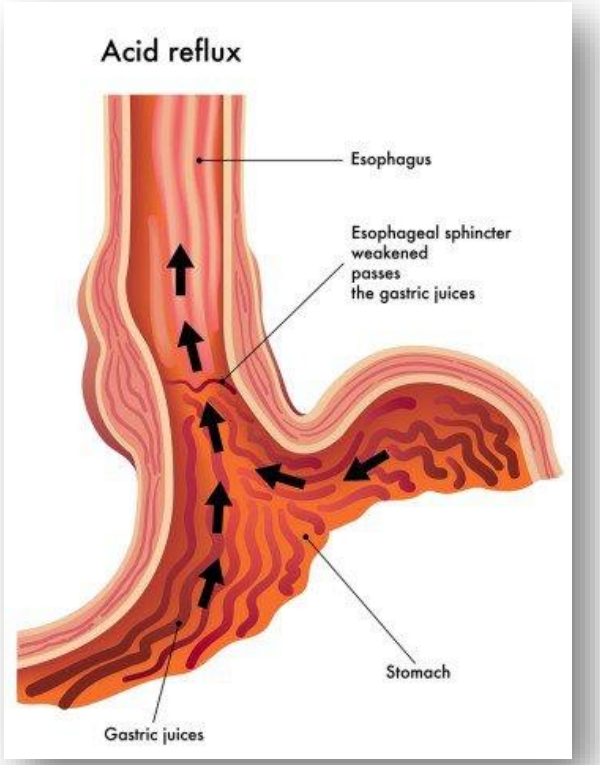
Airway and chest

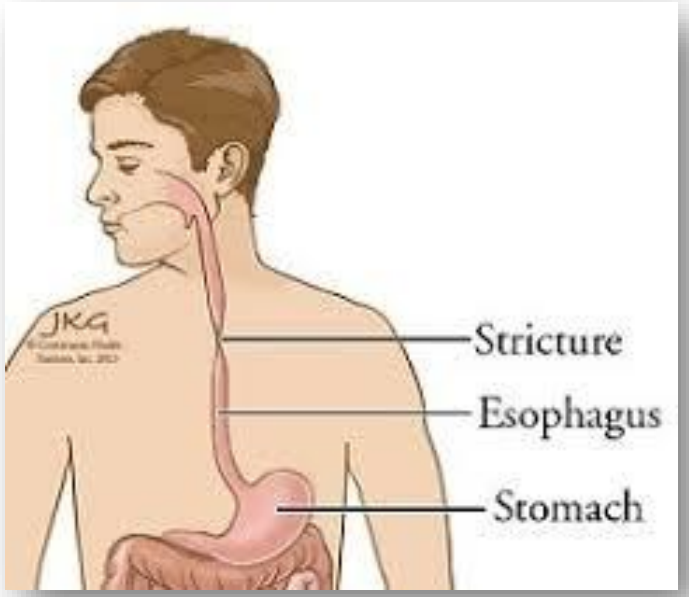
vomiting AND Reflux and pain

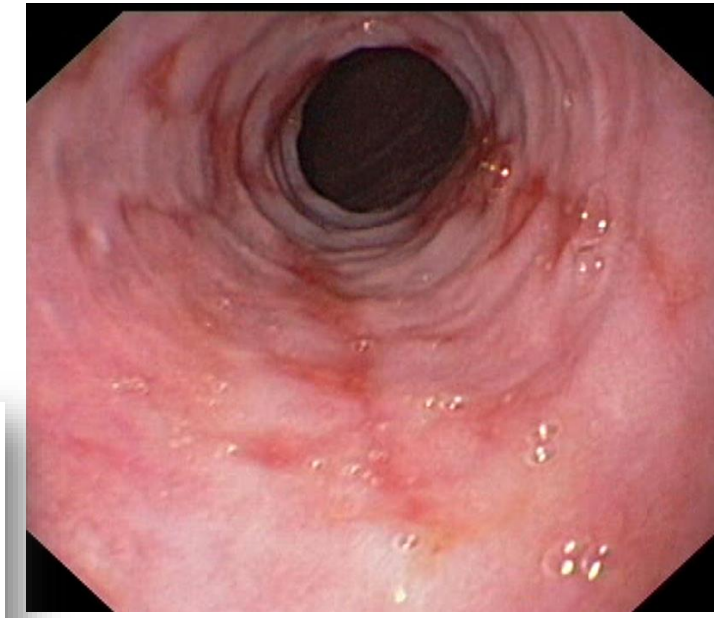
are my main worries !

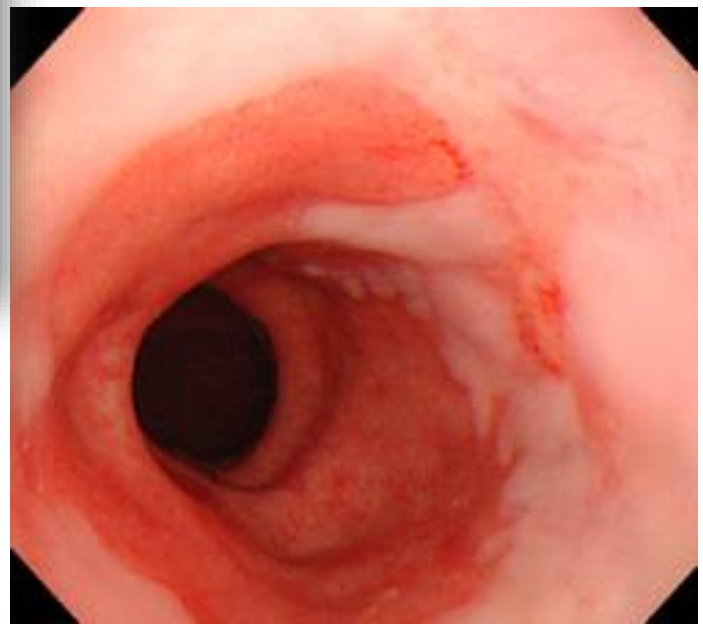
We are also trying to prevent complications….

Erosive esophagitis
Stricture
Barrett's









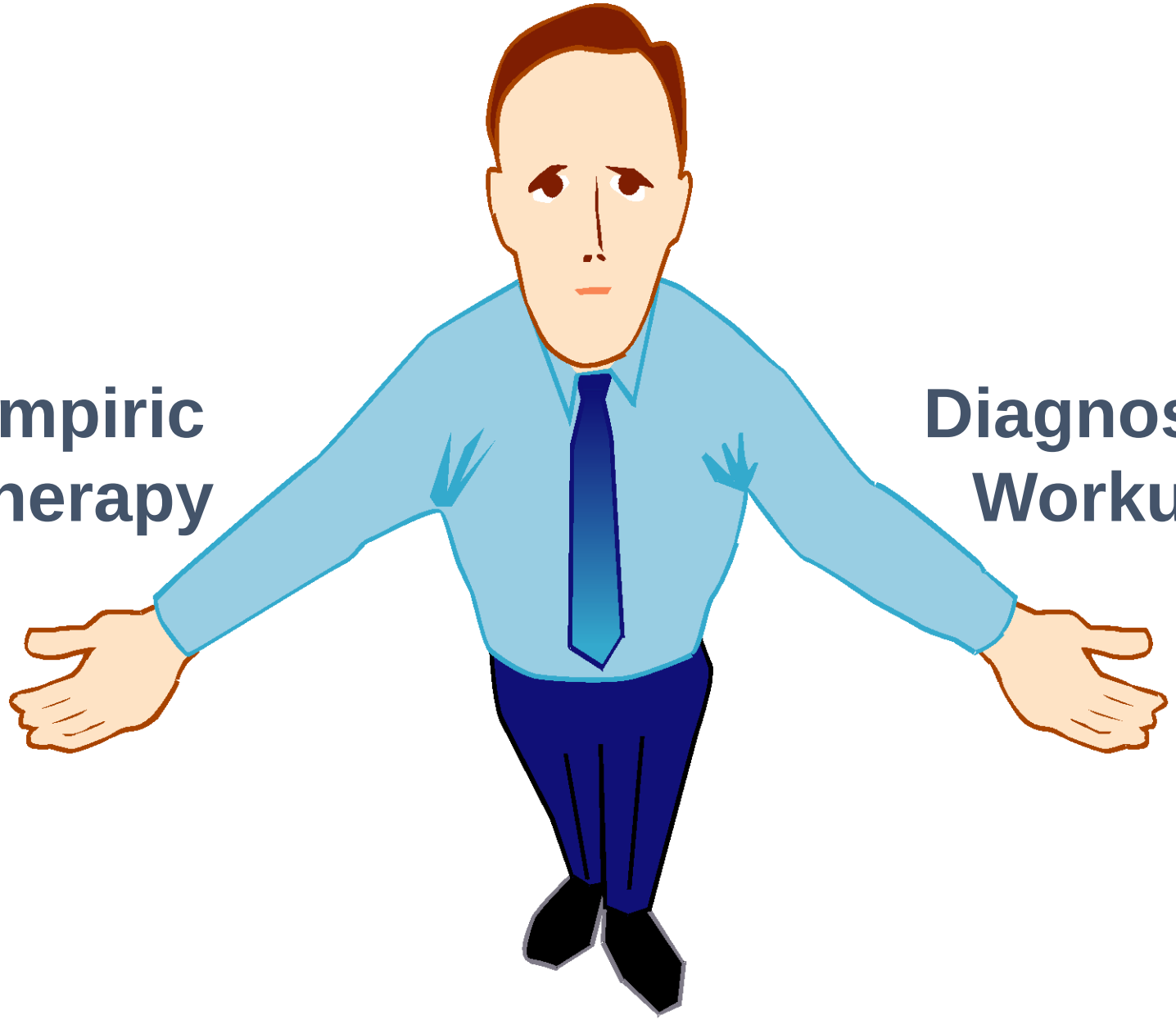
Are they NERDS?
Non erosive reflux disease !
No esophagitis but pain due to acid – a functional GI problem

GORD and Neurological conditions

Patient Population	GER Manifestation	Prevalence
Cerebral palsy [1]	**Moderately severe to severe GER**	**10/32 (32%)**
Severely Neuro-Developmentally affected [2]	**Recurrent vomiting** **Confirmed GER**	**20/136 (15%)** **15/20 (75%)**
Vomiting patients with neuro-developmental delay and/or cerebral palsy [3]	**Failure to thrive** **Respiratory symptoms** **Oropharyngeal incoordination**	**31/50 (62%)** **23/50 (62%)** **16/50 (32%)**

[1] Gustafsson & Tibbling, *Acta Paediatr* 1994;83:1081; [2] Sondheimer & Morris, *J Pediatr* 1979;94:710; [3] Ravelli & Milla, *J Pediatr Gastroenterol Nutr* 1998;26:56

Empiric Therapy



Diagnostic Workup

Diagnostic tests we can do

- Imaging*
  - Barium swallow and meal …. (follow through) – assess anatomy (HH etc)
  - Video-fluoroscopy (video swallow, VF) with Speech Therapist
- pH study and Impedance manometry*
- Upper endoscopy and biopsy

Goals of medical therapy

- Control symptoms
- Promote healing
- Prevent complications
- Improve health-related quality of life
- Avoid adverse effects of treatment
  - Clearly a balance to be had

What meds can we use?

- **Infant Gaviscon** (constipation)
- **Thickeners** and thickened feeds (Carobel, SMA staydown, Enfamil AR, Thick n' easy)
- **Anti-acid drug**s – dose, frequency**
  - ~~Ranitidine~~, **PPI** (omeprazole, lansoprazole, esomeprazole)
- **Motility agents (Prokinetics)**
  - Domperidone*, metoclopramide, bethanecol*
  - Baclofen, Erythromycin, Ondansetron, Pizotifen
- **Dietary changes-** CMP free, specialised milks (EHF, AA)

Approaches to Acid-Reducing Therapy

Step Down

- **Begin treatment with PPI**
- **Maintain with PPI**
- ~~**Switch to H2RA**~~

Step Up

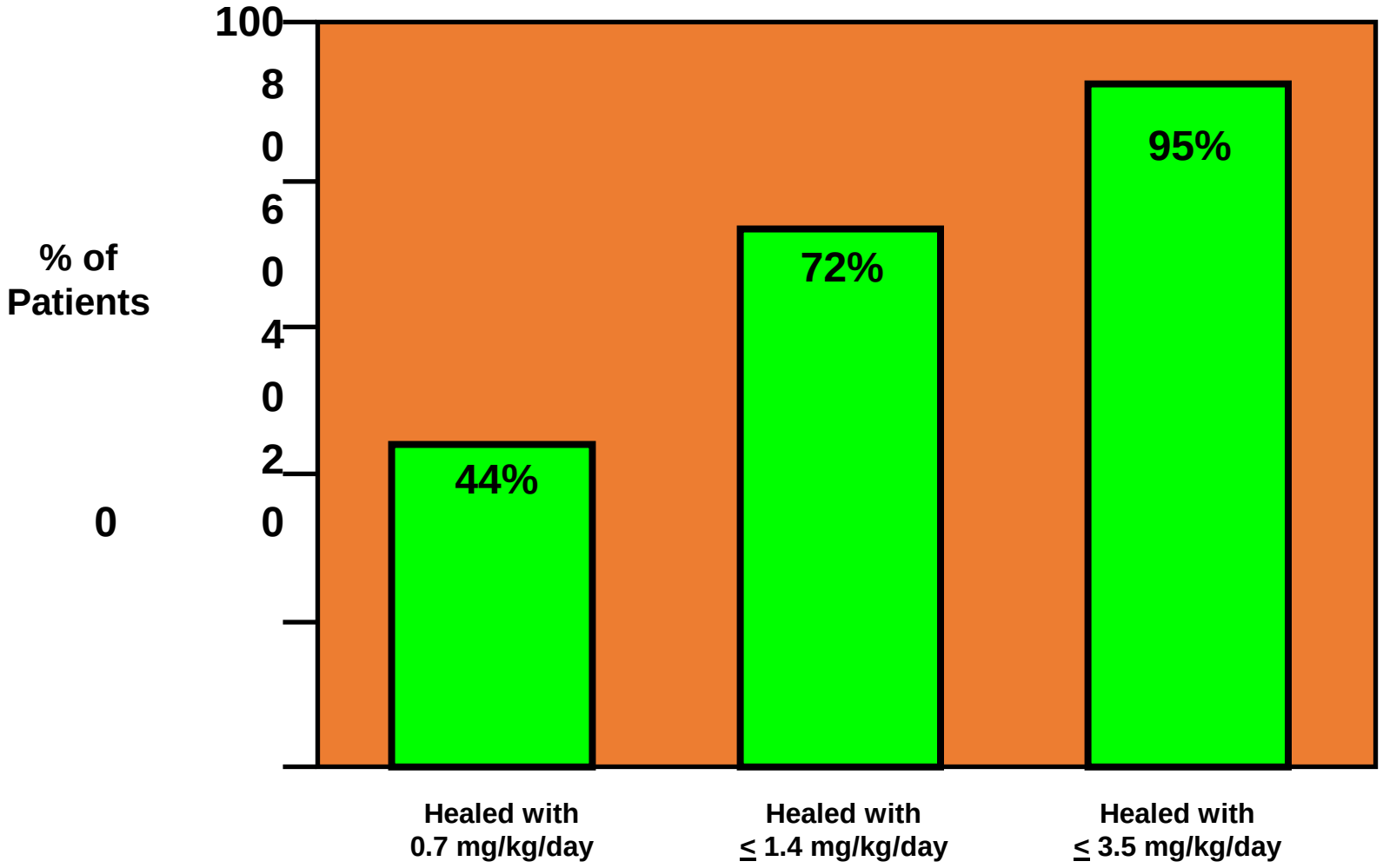
- ~~**Begin treatment with H2RA**~~
- **Inadequate response → PPI**
- **Inadequate response → ↑ PPI dose**

What's 'top-whack' dosing? And why might it be relevant?

3 is a magic number !

Extrapolated from dosing studies…

Omeprazole and Esophagitis



N = 65 children with erosive esophagitis,50% were neuro-disabled

Chart shows healing at 3 months

Hassall et al, *J Pediatr* 2000;137:800

Why bother with aggressive treatment early?

- **Many reasons**
  - **Self harm, aggressive behaviour well recognised**
  - **It hurts !**
    - **We would all want something done about it !**
  - **Complications must be avoided if possible**
    - **Distressing self harm and injury and QOL**
    - **Stricture***
    - **Barrett’s esophagus***

Who is a Candidate for Antireflux Surgery ?

As a medical doctor, In my opinion, a patient who:

- **Fails medical therapy due to GORD**
- **Is dependent on aggressive or prolonged medical therapy ** - all a balance in my view**
- **Airway safety !**
  - **Has persistent asthma or recurrent pneumonia due to GERD**
  - **Has ALTE (apneas, near-miss SIDS)**
- **Must take into account post op risks of fundo – retching especially – are there alternatives / options?**

If aggressive anti reflux treatment fails….there needs to be an MDT discussion about what next….

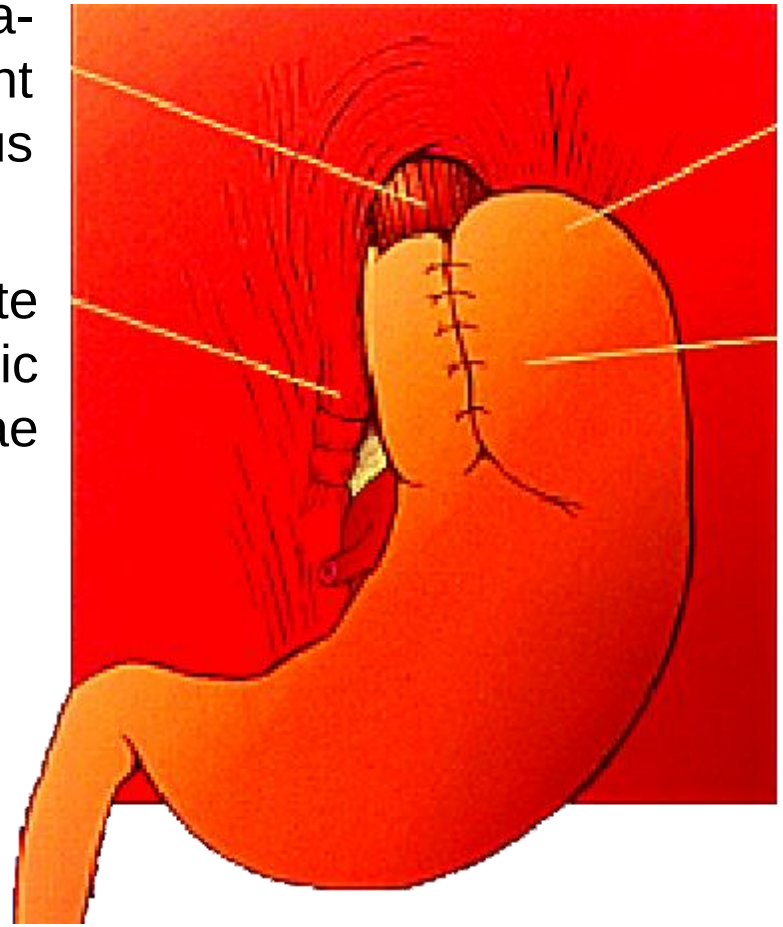
Surgical Options…..many…..

- Fundoplication
- Fundoplication and gastrostomy (G-tube)
- Gastrostomy only
- PEG-J
- G-J
- Needs to be bespoke shared decision….
- Ask your surgeon about their experience and outcomes !

Principles of Antireflux Surgery

Restore intra-abdominal segment of oesophagus

Approximate diaphagmatic crurae



Reduce hiatal hernia when present

Wrap fundus around LOS to reinforce antireflux barrier

Patient Education

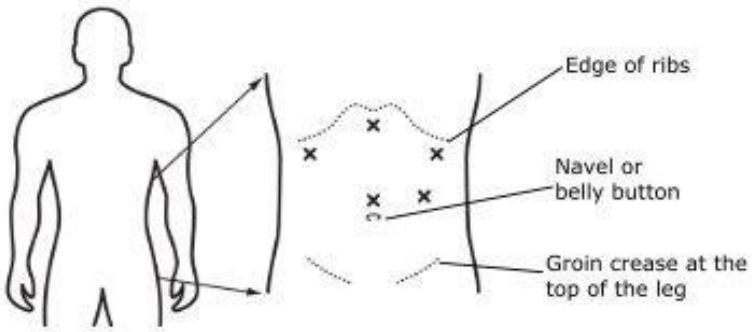
Laparoscopic Fundoplication

What is a laparoscopic fundoplication?

A fundoplication is surgery to correct an abnormal flow of acid, called reflux, from the stomach up into the esophagus. The surgery tightens the area where the esophagus and stomach join.

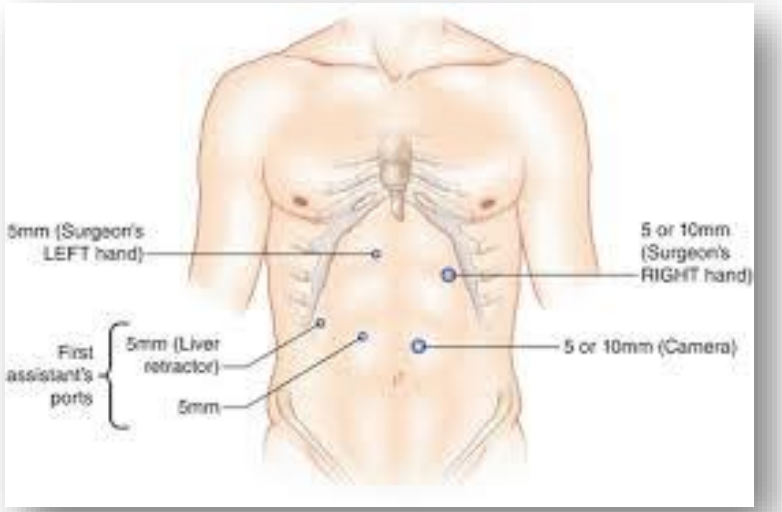
Laparoscopic means the surgery is done through small incisions and uses a small telescope. There are 5 incisions, each 5 to 10 mm long.

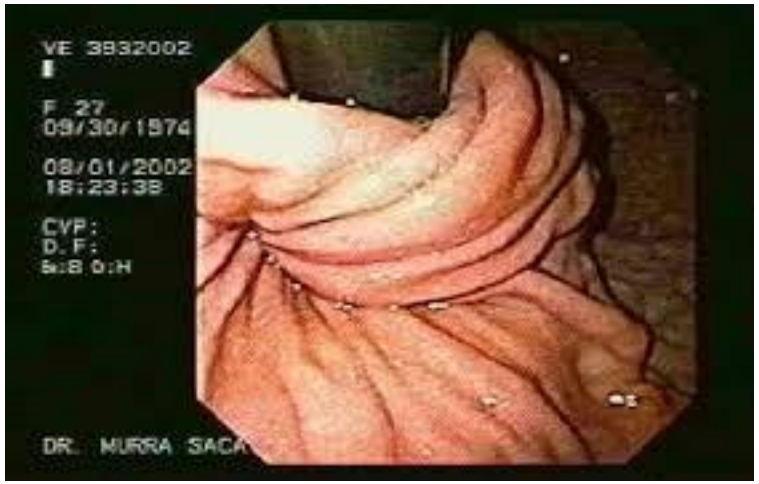
This picture shows where the incisions are usually made.

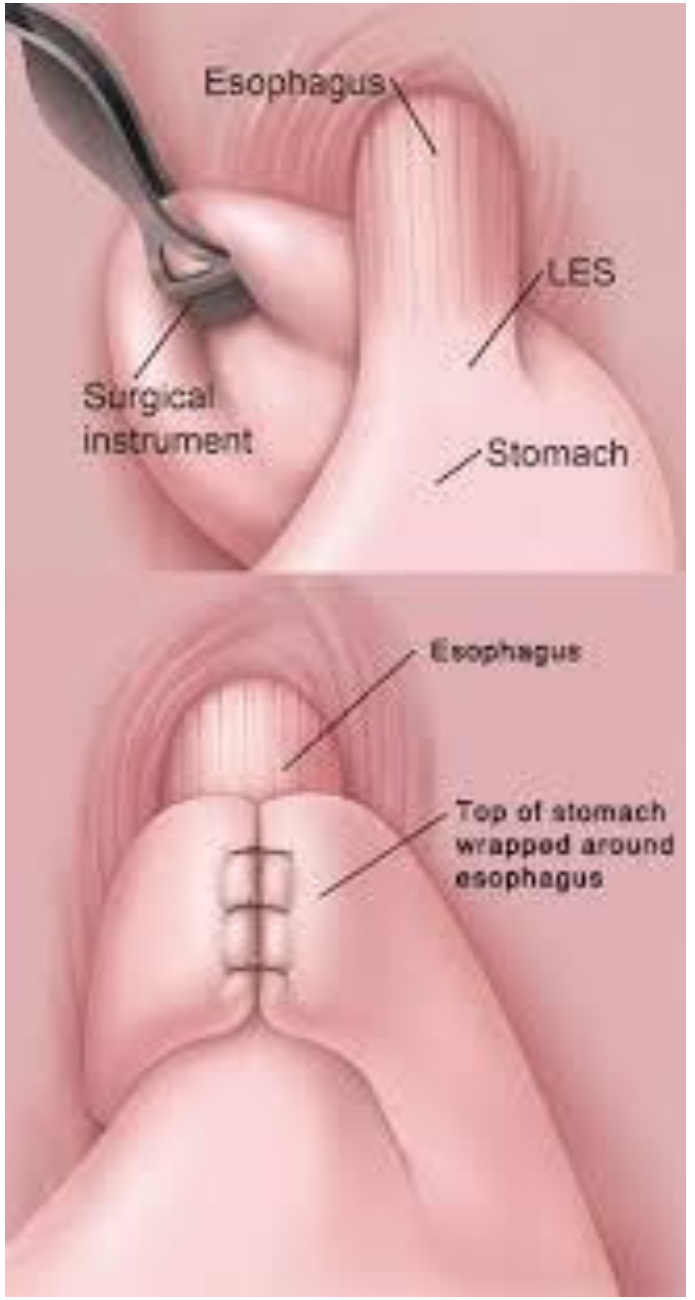


A small lighted telescope and 4 instruments are put into our abdomen through the incisions. The telescope also has a camera, which sends a picture to a screen. The surgeon watches this screen as he or she does the operation.

For a small number of people, the fundoplication cannot be done this way. For safety reasons, the surgeon may decide during the operation to change to an open procedure. An open procedure is a fundoplication done through a standard incision.

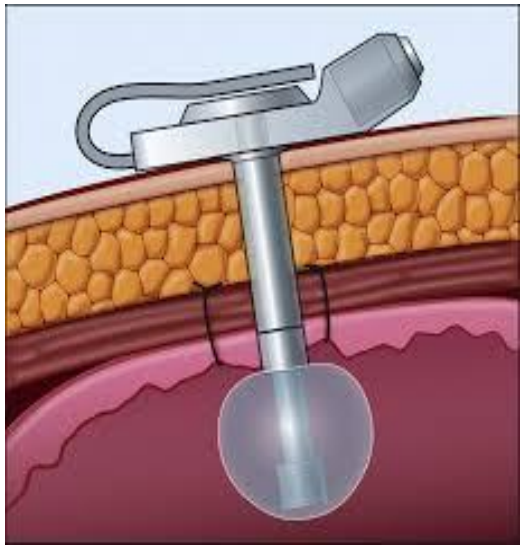


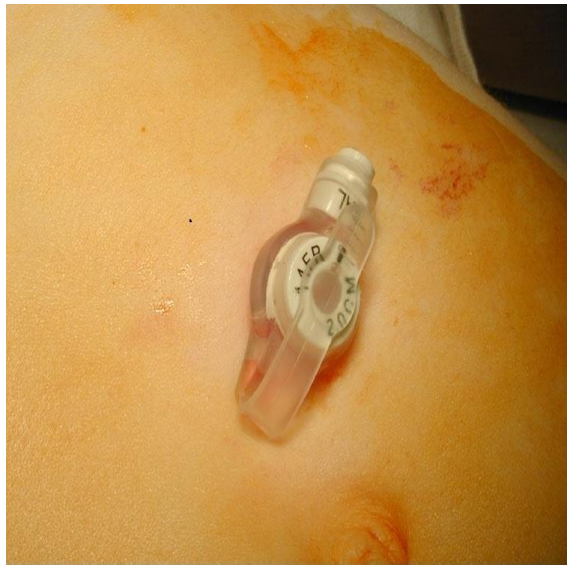


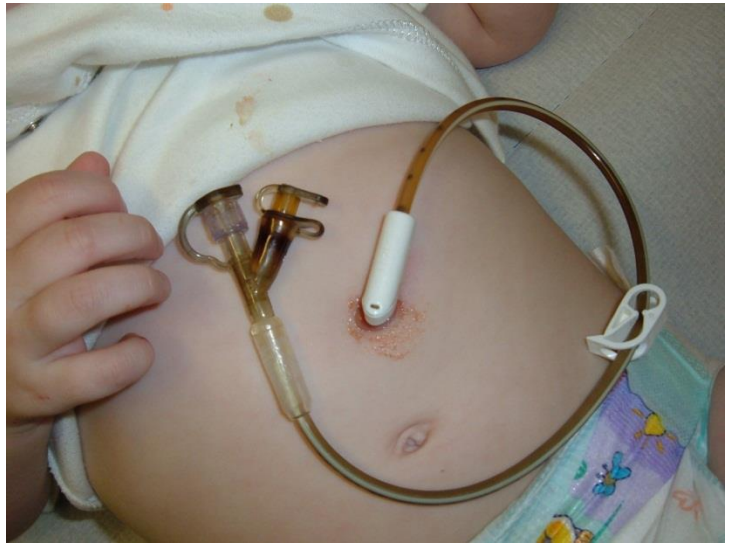


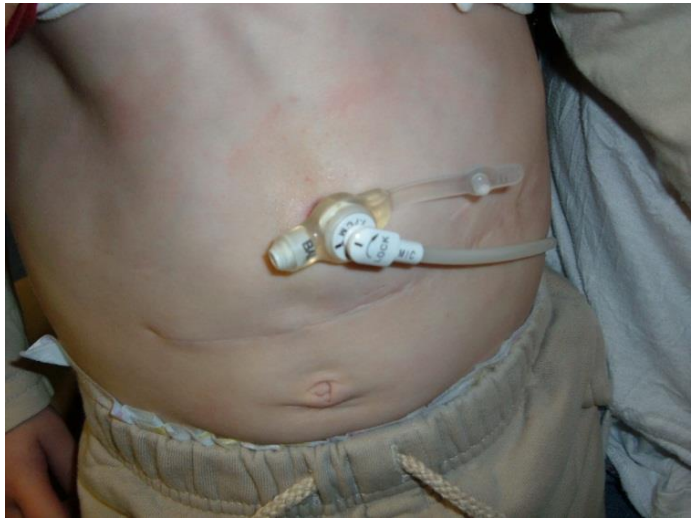
Gastrostomy tubes / buttons

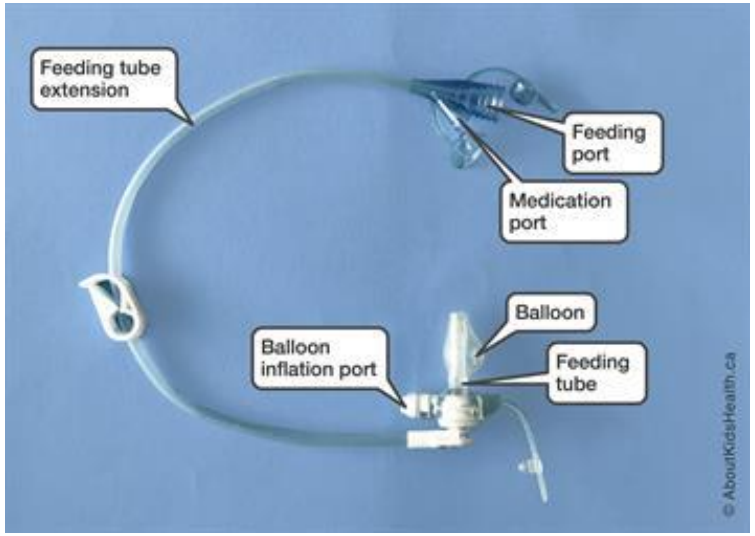
PEG

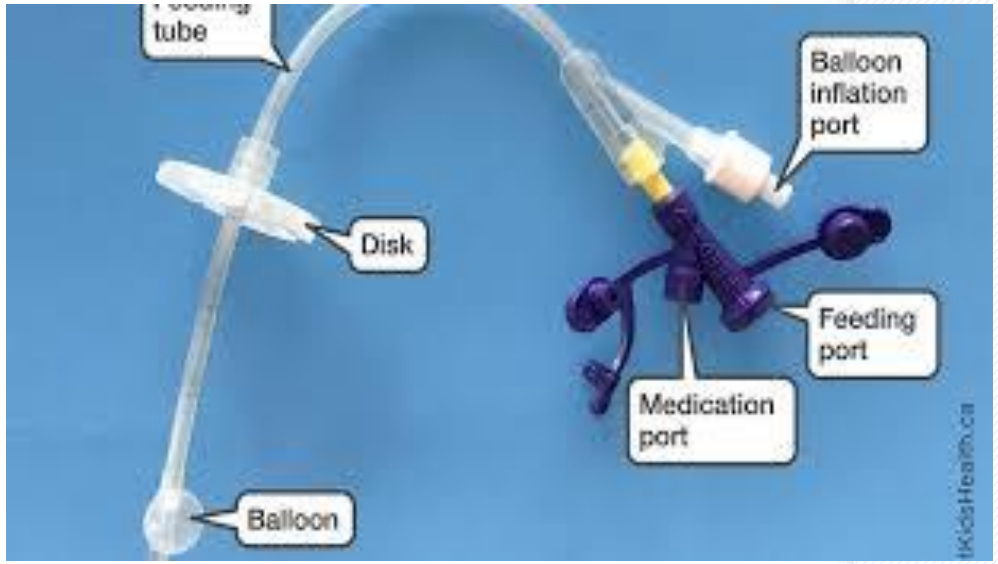


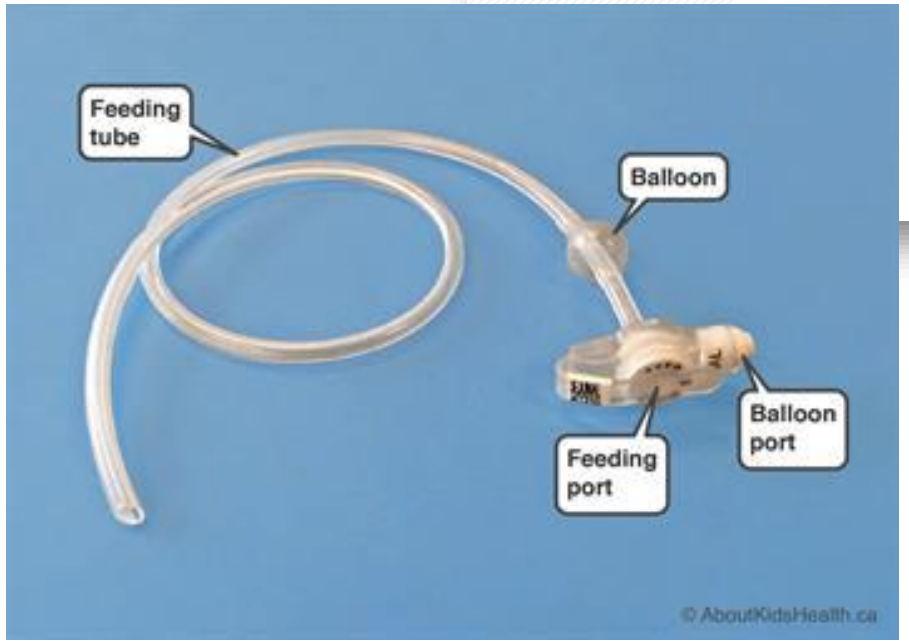












AboutKidsHealth Trusted answers from The Hospital for Sick Children

MENU

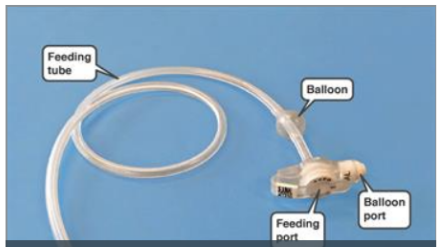
Listen

English

G/GJ tubes: Mic-Key low-profile GJ tube

By SickKids staff

A low-profile GJ tube is a type of feeding tube. Learn how to care for your child's GJ tube and how to give feeds and medication.



Click on image to drop down to the related article text

When feeding and growth are issues too….

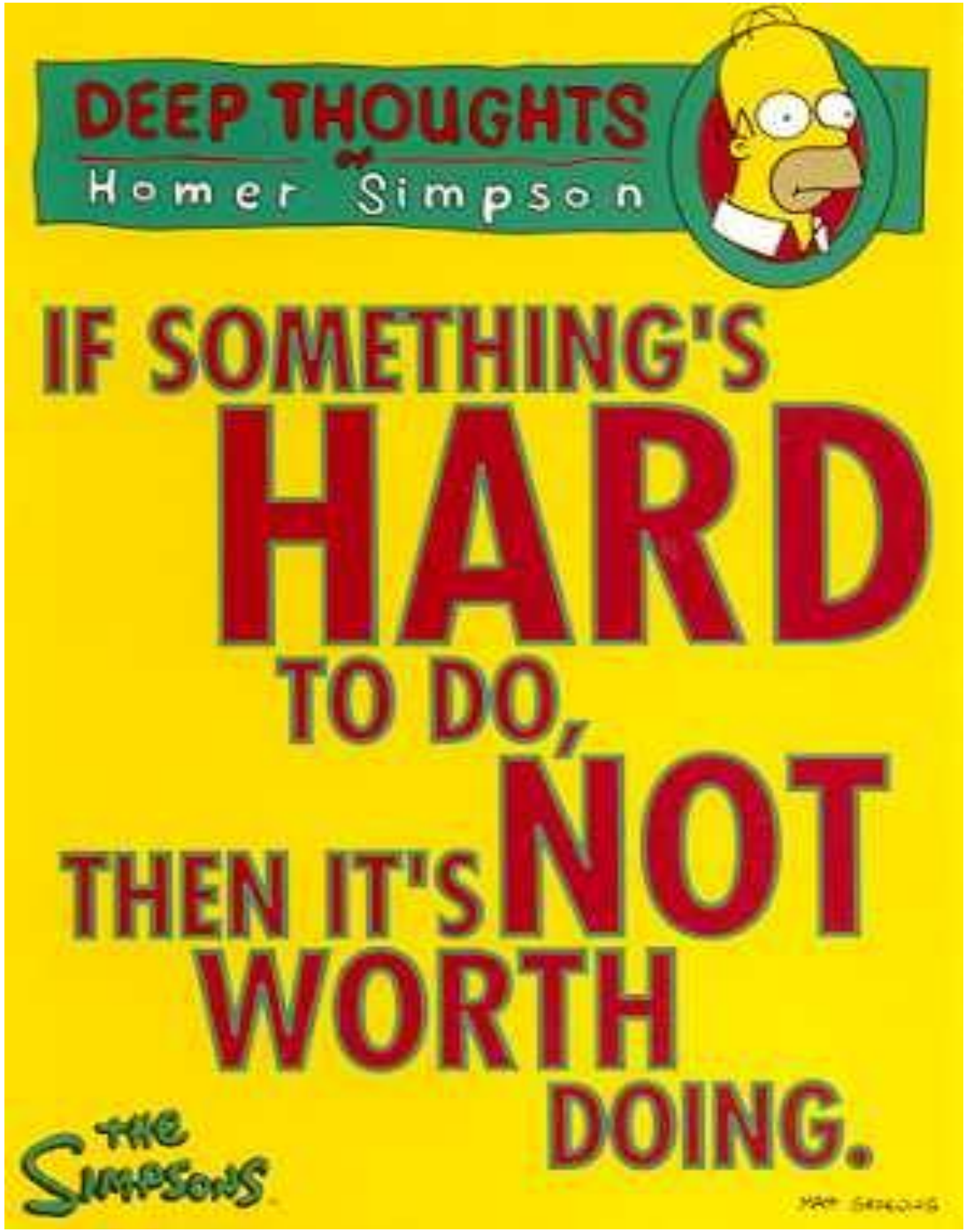
Nutrition and oral skills

- Big topic !
- Dietetic input is mandatory
  - Specific growth pattern
  - See CdLS specific growth charts*
  - Energy requirements must be met
  - Specific problems related to vomiting or poor absorption of calories
- SALT input – feeding clinics, MDT clinics

Take home messages

- Regular Infant reflux usually resolves by 1-2 years* CdLS many very different
- Symptom complex and decision making
- GERD issues:
  - Are any investigations required at all? Or do we Just treat?
  - Know limitations of diagnostic tests, use them wisely – will it change what we do- always ask !
  - Tailor management to the child/ young person/adult and what you think!
- Pain and self injury are common themes – Acid and pain are major issues, but not the only ones
- Surgical management has a big part to play – airway safety, reflux management and feeding / nutrition
  - Tailored management and MDT discussions to influence what best plan is for each person
- Importance of AHPs / therapists – SALT, OT, dietitian

Questions?


DEEP THOUGHTS
by
Homer Simpson
IF SOMETHING'S
HARD
TO DO,
THEN IT'S NOT
WORTH
DOING.
THE SIMPSONS