



supporting those born unable to swallow

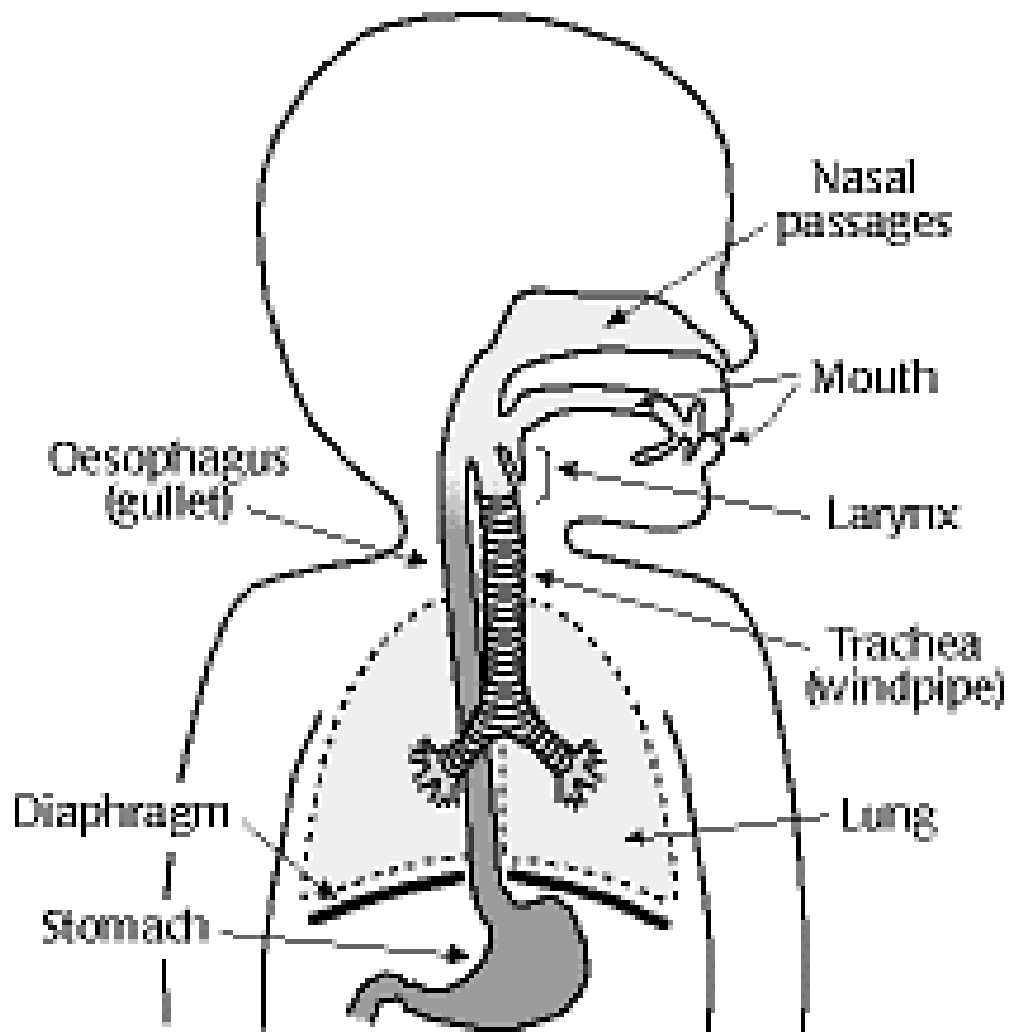
# **About the TOF / OA condition**

**John Pearce, Treasurer & Trustee**

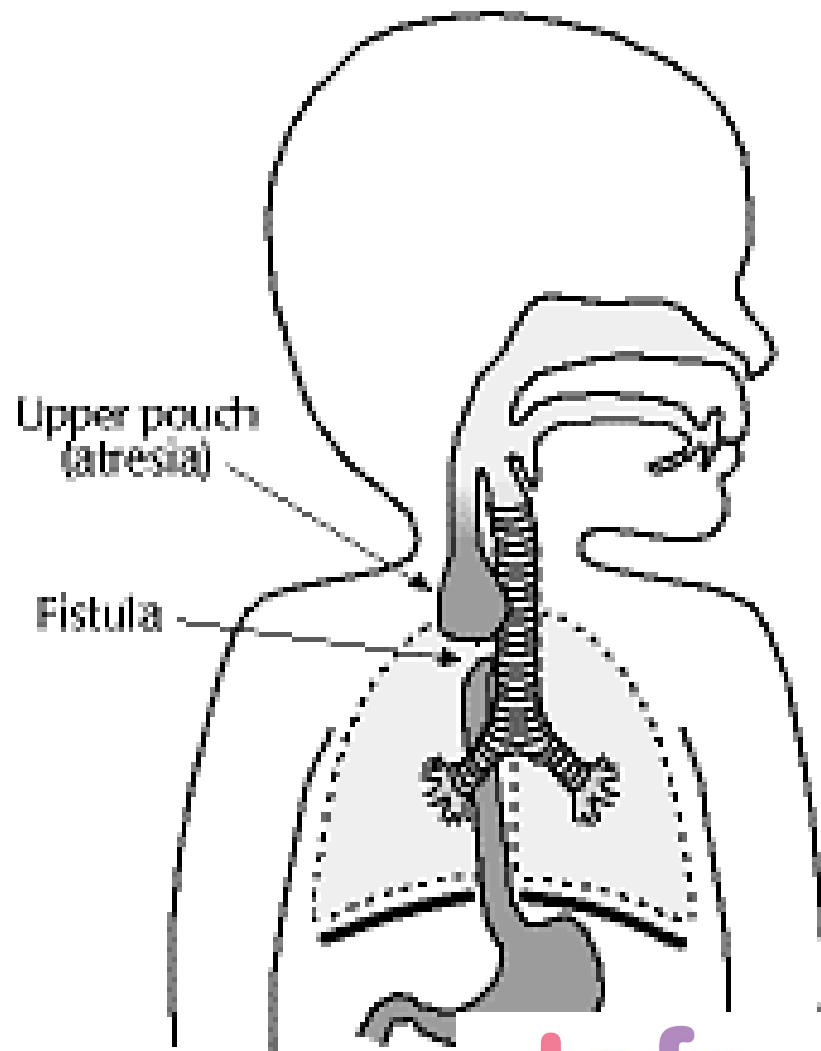
# TOF / OA: A rare birth defect ... (1 in 3000)

- Tracheo-Oesophageal Fistula and/or  
Oesophageal Atresia
- Imperfect formation of gullet and windpipe
- At 4-8 weeks gestation.....
  
- About 180 born p.a. in UK
  - UK population of c 3000 children,
  - c. 6000 adults

# Child without TOF/OA



# Child with TOF/OA



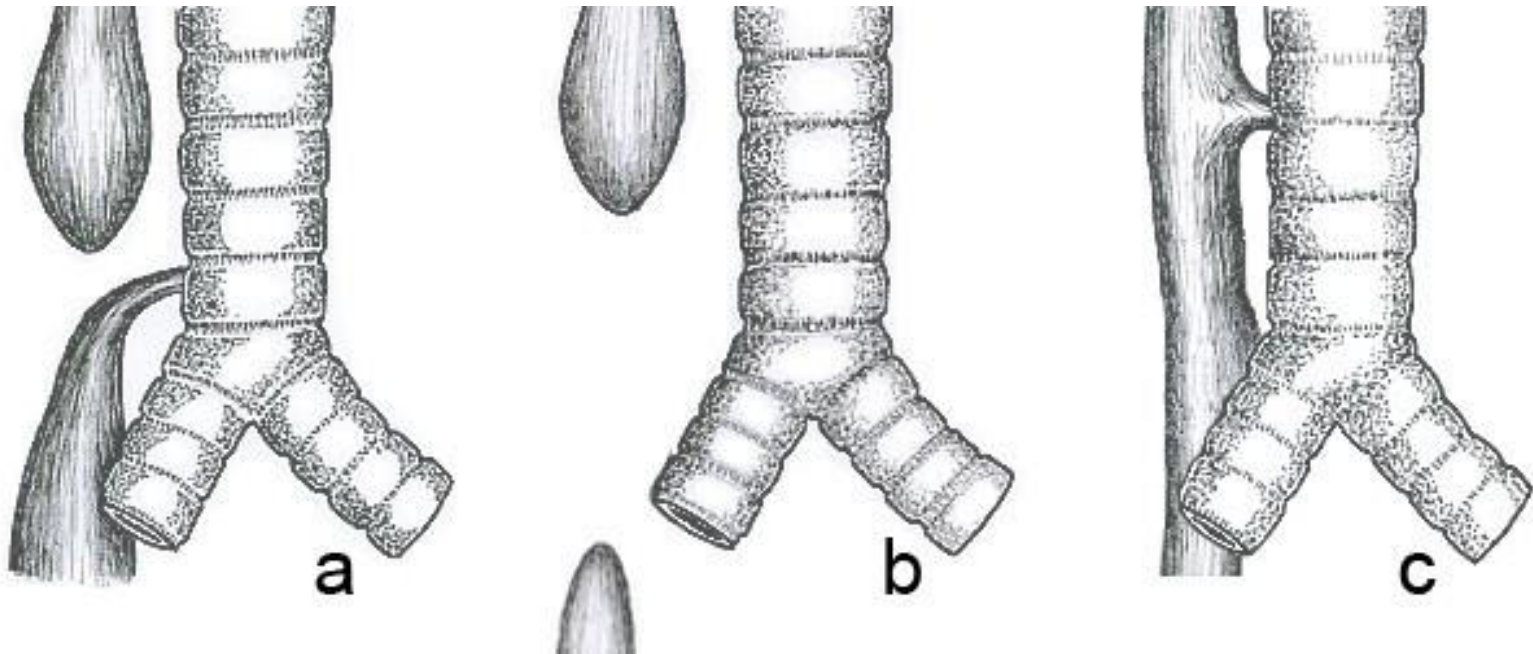
# TOF / OA: No single clear “cause”

- Environmental factors suggested
  - Alcohol, some drugs, diabetic mother
- 5-10% of TOFs have a trisomy ... Downs
- c.5% of TOFs have other genetic causes
  - Many rare syndromes include TOF/OA
  
- TOF incidence higher for twins
- About half also some other defect



# TOF / OA: Several broad “types”

- 86% are OA & lower pouch TOF (A)



- 7% OA but no fistula (B)
- 4% fistula but no OA (C)

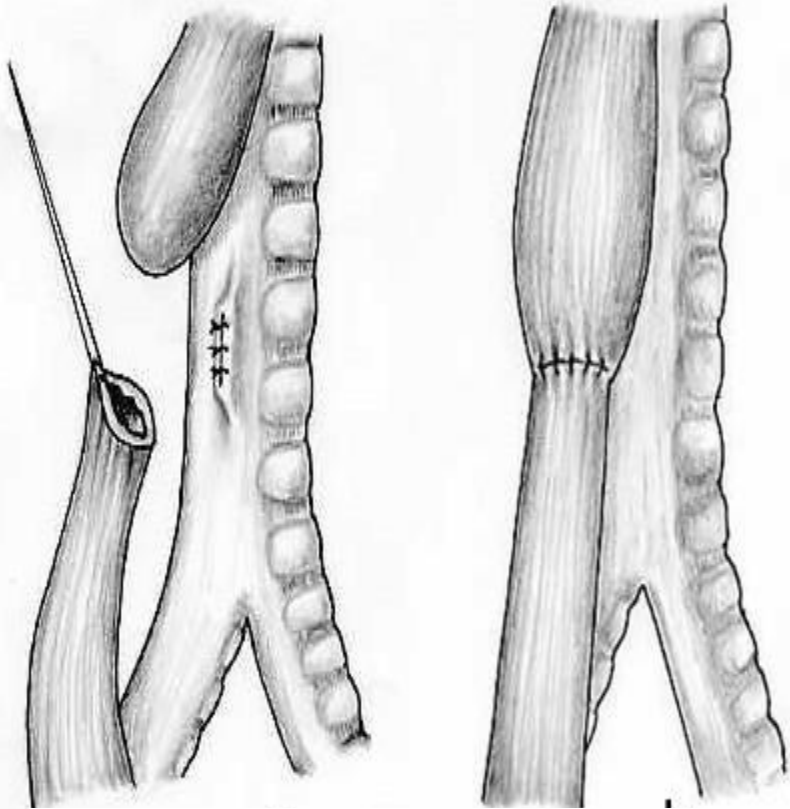
# TOF / OA: surgery within days

- Terminal prior to 1941
- First success in Europe in 1947
- Requires immediate surgery ...c.30 UK centres
- Most cases:
  - straightforward repair surgery
  - discharged in days / weeks
  - 50% only ever have one procedure

# Much progress in 70 + years...

- Survival rates now 95% +
  - straightforward TOF / OA >>95%
  - lower for renal, cardiac problems
- But surgery not a “cure”.
  - difficulty swallowing
  - problems with reflux
  - recurrent chest infections
  - TOF cough
- Life-long issues remain for some

# Swallowing...obvious oesophageal problems

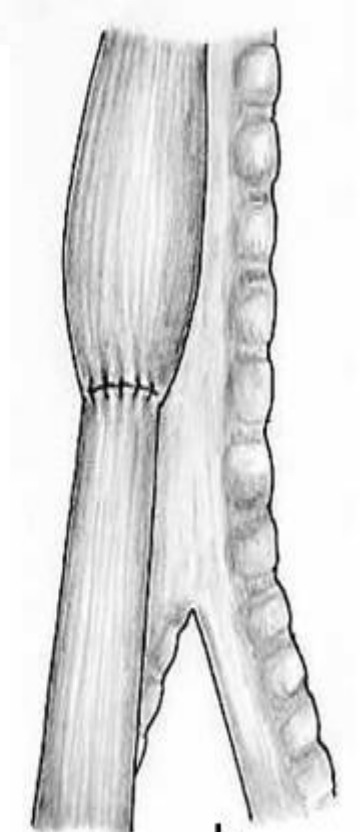


- Partial blockage  
–at site of join
- Stricture at join line
- lower oesophagus  
narrower than upper



# Swallowing...more oesophageal problems

- During gestation:
  - independent development of U & L oesophagus
  - foetuses normally suck thumbs, swallow, etc
  - peristalsis not successfully learned
- Once born
  - weak, absent or contrary motility
  - larger problem than strictures

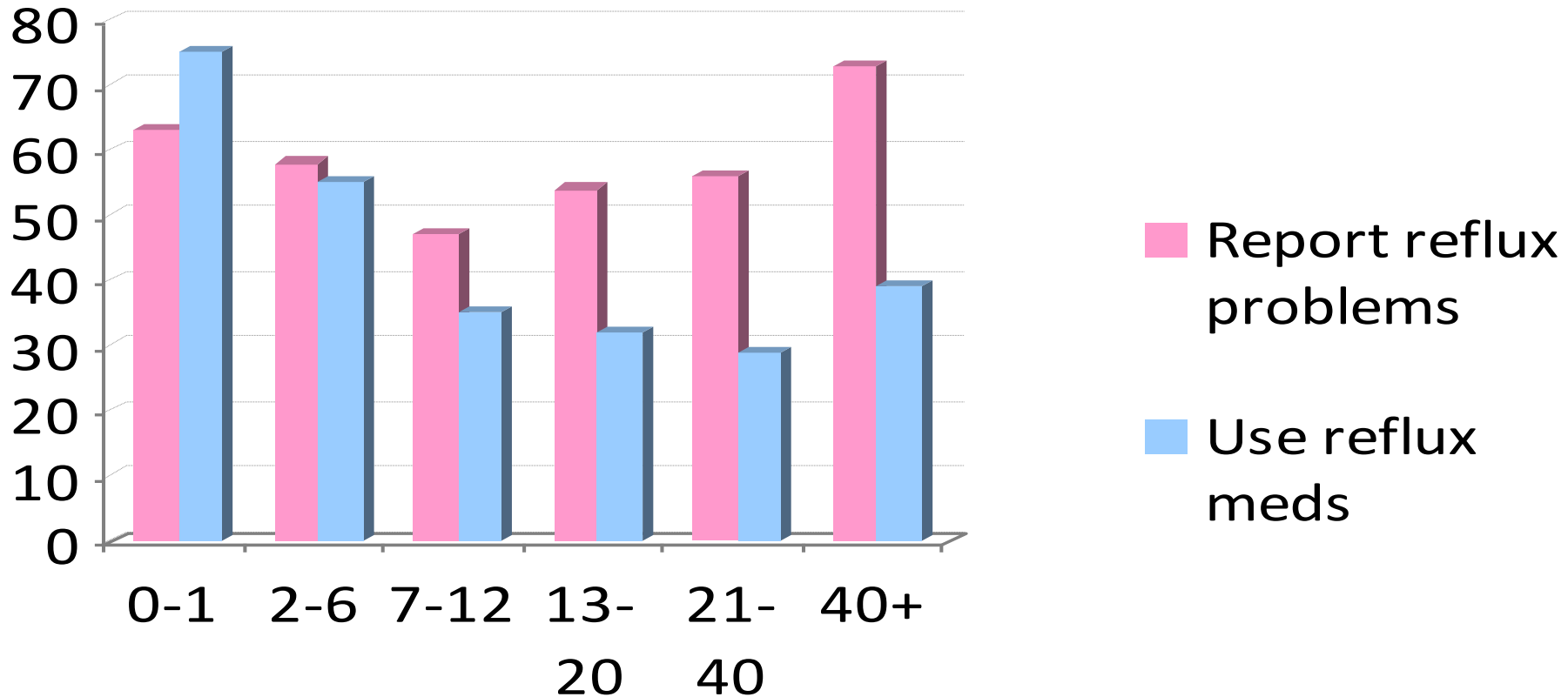


# Solid food

- Weaning often delayed in TOF / OA babies
- Swallowing challenge increases with solids
- Occasional “stickies” common
  - 0-1 yr olds: 42% report “often” or “sometimes”
  - strictures at the join
  - lack of / contrary peristalsis

# Reflux: **very** common

- Stomach acid reflux common for TOFs
  - sphincter “valve” issues?



# Reflux

- Irritation to lower oesophagus
- Very serious long term
  - can lead to real damage,
  - Barrett's oesophagus, cancer
- High rates of Barrett's amongst TOF adults
- 30 yrs ago, 1/3 had anti-reflux surgery
  - often between ages of 2 and 6
  - Nissen's fundoplication
- PPI drugs widely used now
  - don't actually stop reflux, though



# Respiratory

- TOFs usually have slightly abnormal trachea
  - fistula join site – some cartilage missing?
  - usually strengthens in time
- Tracheomalacia – collapse upon expiration
  - TOF Cough .... Barking sound
- TM can need surgery – aortopexy

# Respiratory

- Frequent chest infections are the norm
  - aspiration
  - reflux
- Improve as child grows
- Lung damage not uncommon
  - Non-CF Bronchiectasis

VACTERL association.... (1 in 6000)

**About 1 in 3 TOFs are VACTERL**  
**3 or more of:**

- Vertebral abnormalities
- Anorectal atresia
- Cardiac abnormalities – VSD most common
- TOF with / without OA (TEF...EA)
- Renal anomalies
- Limb abnormalities  
– generally forearm



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# Everyday life - being Practical

- Avoiding some foods / packed lunch to school
- Cutting food up well and chewing thoroughly
- Drinking lots (water!) at mealtimes
- Eating little and often – small meals
  
- Sloping bed
  - tackles reflux,
  - helps with  
bronchiectasis





# TOF / OA: mostly a childhood issue

- TOF troubles ease during childhood eg 0-5
  - They get stronger, more able to cope
  - age 7-12 ...respiratory infections < 1p.a. ...50%
    - ...stickies seldom / never ... 55%
    - ...can eat everything ...76%
- TOF teens mostly OK
  - age 13-20 ... respiratory infections < 1p.a. 66%
  - high levels of tiredness?
  - teenage stresses heightened by health fears

# TOF / OA: adult outlook

- Most TOF Adults are relatively OK
- Most have normal work and lives
  - >50% suffer reflux
  - about 1/3 say “somewhat” or “significant” impact
  - around 10% have seriously impacted lives
- Raised levels of illness, tiredness
- But greatly raised risk of Oesophageal disease, inc cancer
- Life expectancy not yet established

# TOF / OA: to end

- Major impact on neonates' swallowing
- Ongoing upper GI issues – reflux, dilatations
- Tracheal & respiratory issues too
- They DON'T all “grow out of it”
- Adult outlook: mostly OK

# Acknowledgements

- Images from:
  - TOFS Website
  - Spitz: Oesophageal Atresia. Orphanet Journal of Rare Diseases, 2007. Creative Commons Licence 2.0
  - Hospital for Sick Children, GtOrmond Street
  - TOFS Chew photographs
- Key sources:
  - Spitz (above)
  - Oesophageal atresia, tracheo-oesophageal fistula, and the VACTERL association: review of genetics and epidemiology. C Shaw-Smith. Journal of Medical Genetics 2006
  - Recent EAT survey of EA patients (Jul-Aug 2014)
  - A national observational study to assess antenatal diagnosis, associated anomalies and infant outcomes prospective survey of Current practice with oesophageal atresia in the UK: Shah, Burge, et al BAPS-CASS 2012
  - Long term oesophageal problems for adult TOFS                      TOFS *Chew* Spring 2012