

# Familial Adenomatous Polyposis

David Foreman RN BSc(Hons) PGDipMedEd FHEA

Colorectal Clinical Educator, Derby Hospitals



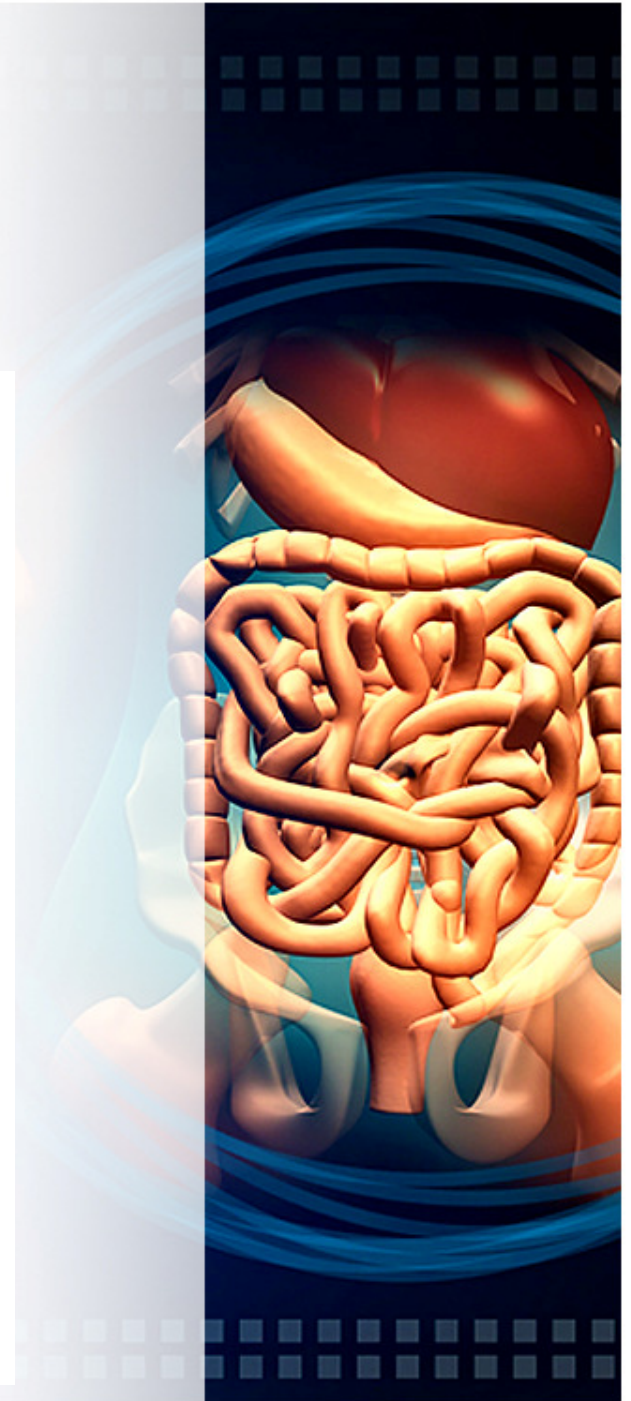
# Familial Adenomatous Polyposis

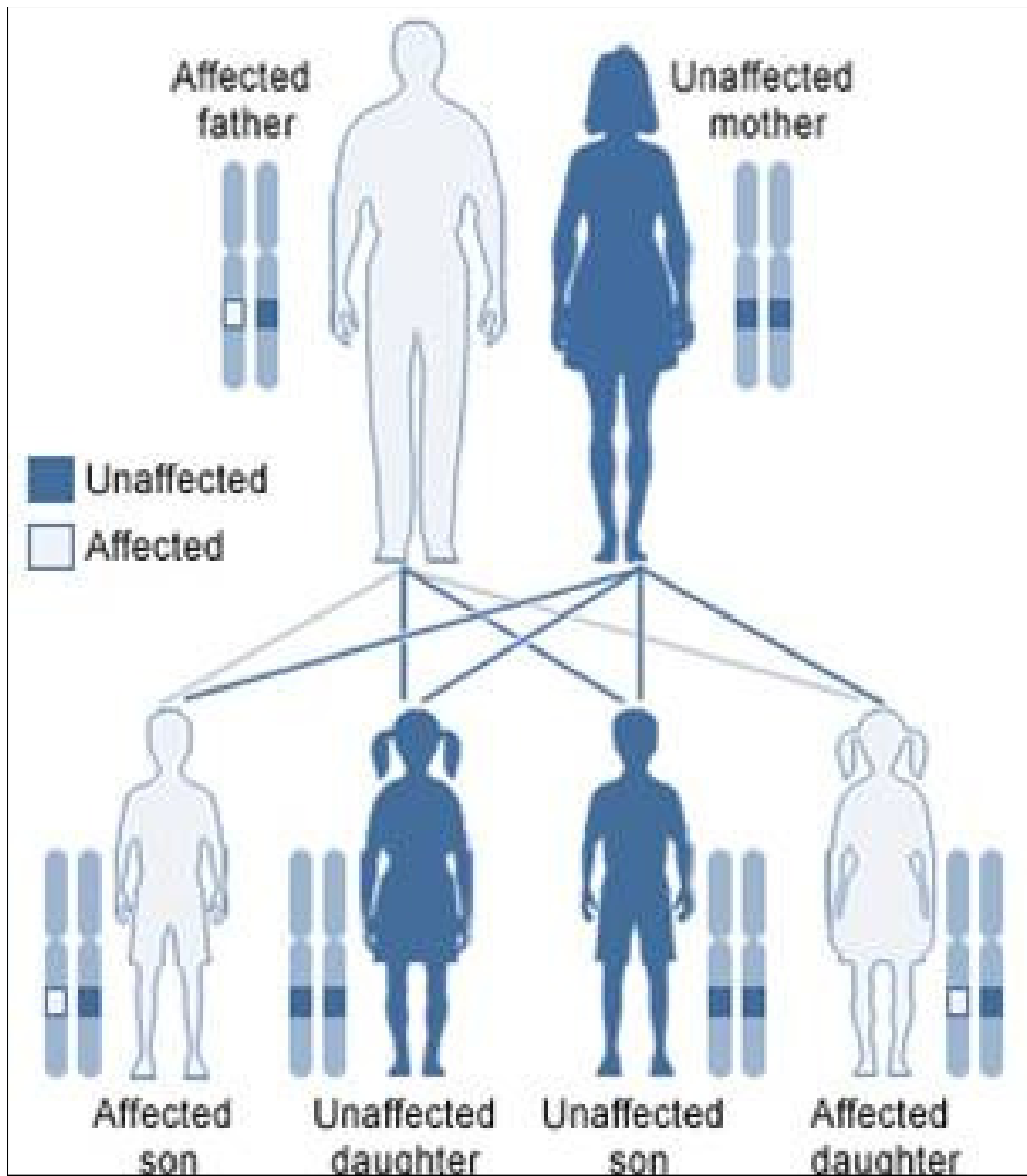
- Familial adenomatous polyposis (FAP) is an autosomal dominantly inherited syndrome characterised by the early onset of multiple colorectal adenomas, one or more of which will inevitably become malignant if not treated.
- The disease is due to a mutation of the APC (adenomatous polyposis coli) gene located on the long arm of chromosome 5.



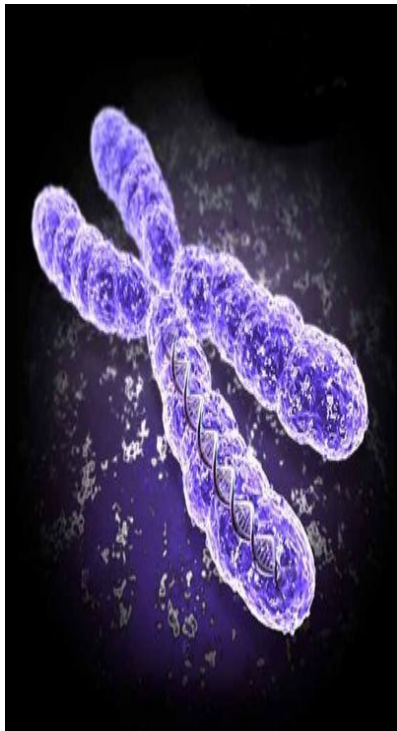
# FAP is a *Genetic* disease

- FAP can be passed from parent to child although genes can *spontaneously go wrong*
- Each child has a 50% chance of acquiring FAP



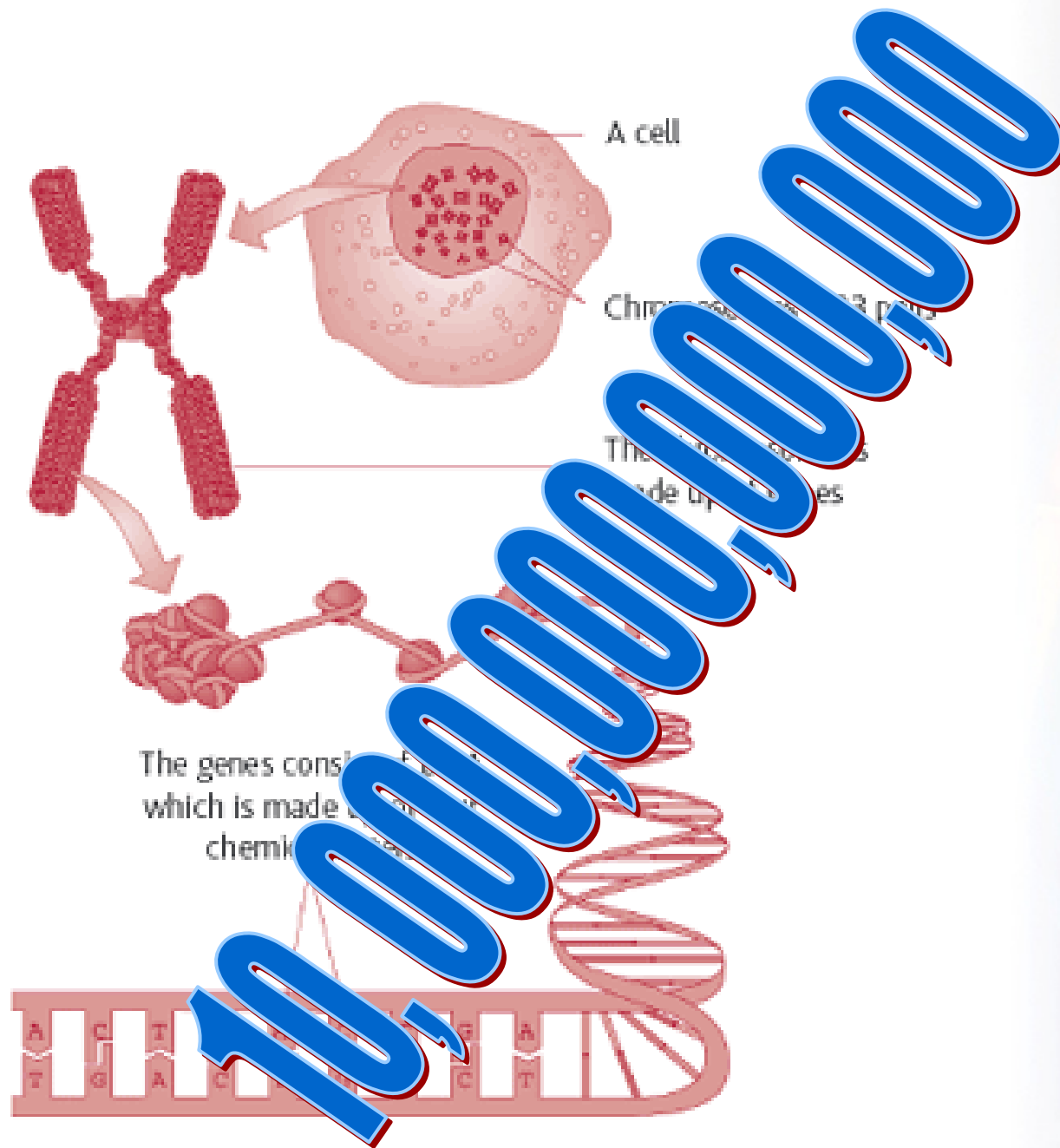


# Humans have 23 pairs of Chromosomes



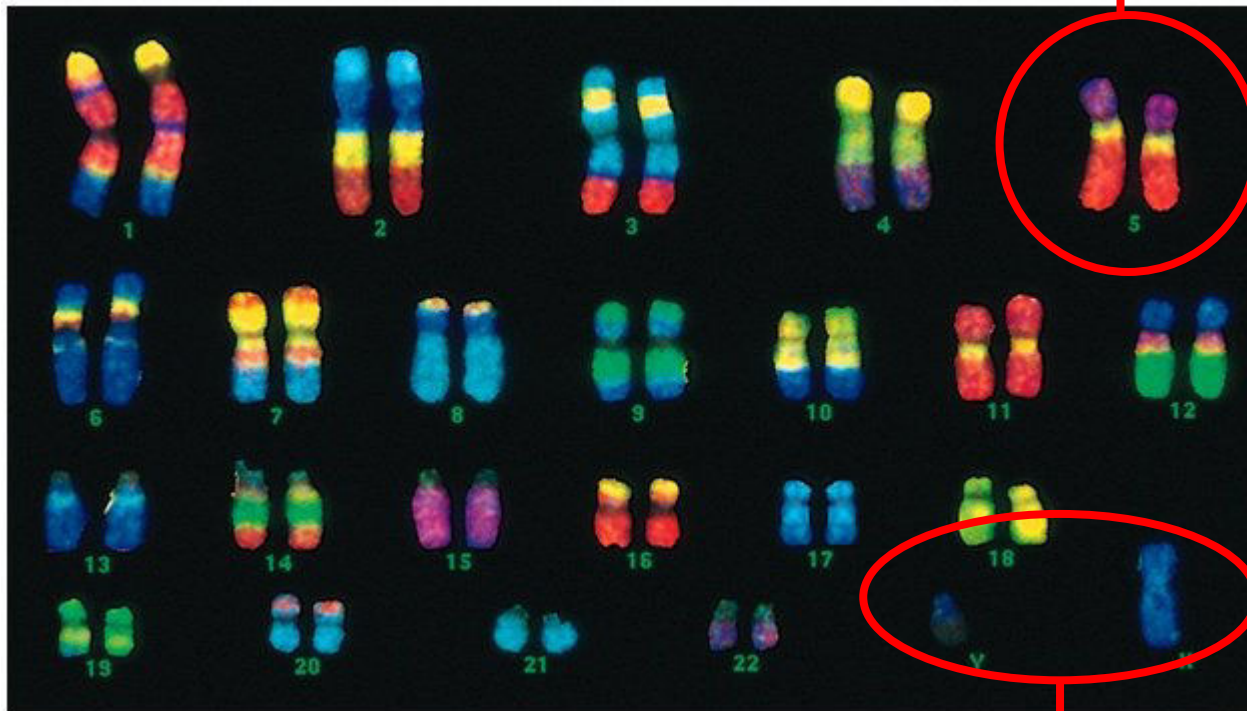
- A Chromosome is an organised series of genes
- A gene is a structure composed of DNA
- DNA contains all of the information to make you who you are.



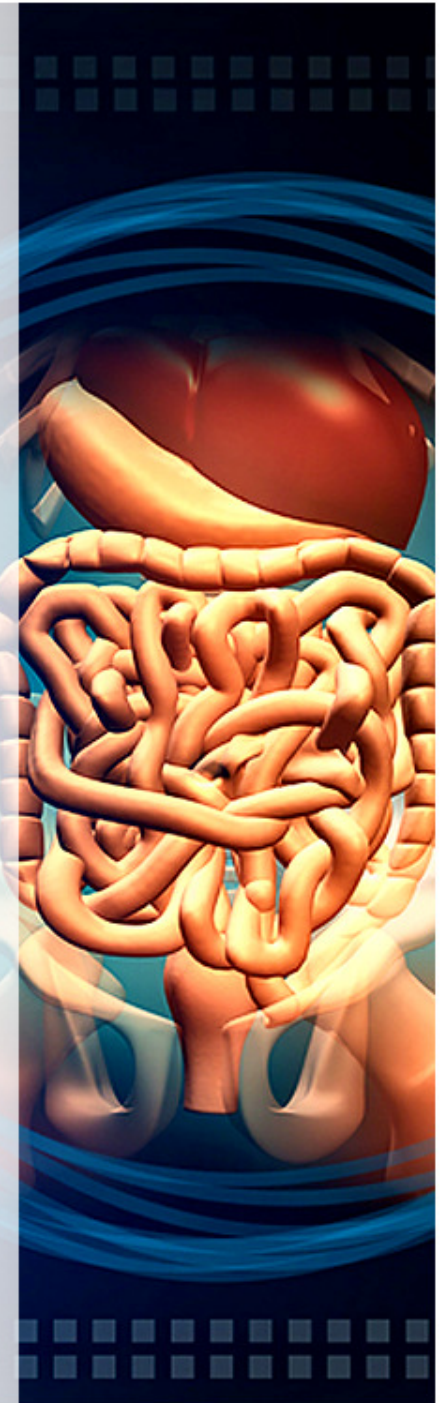


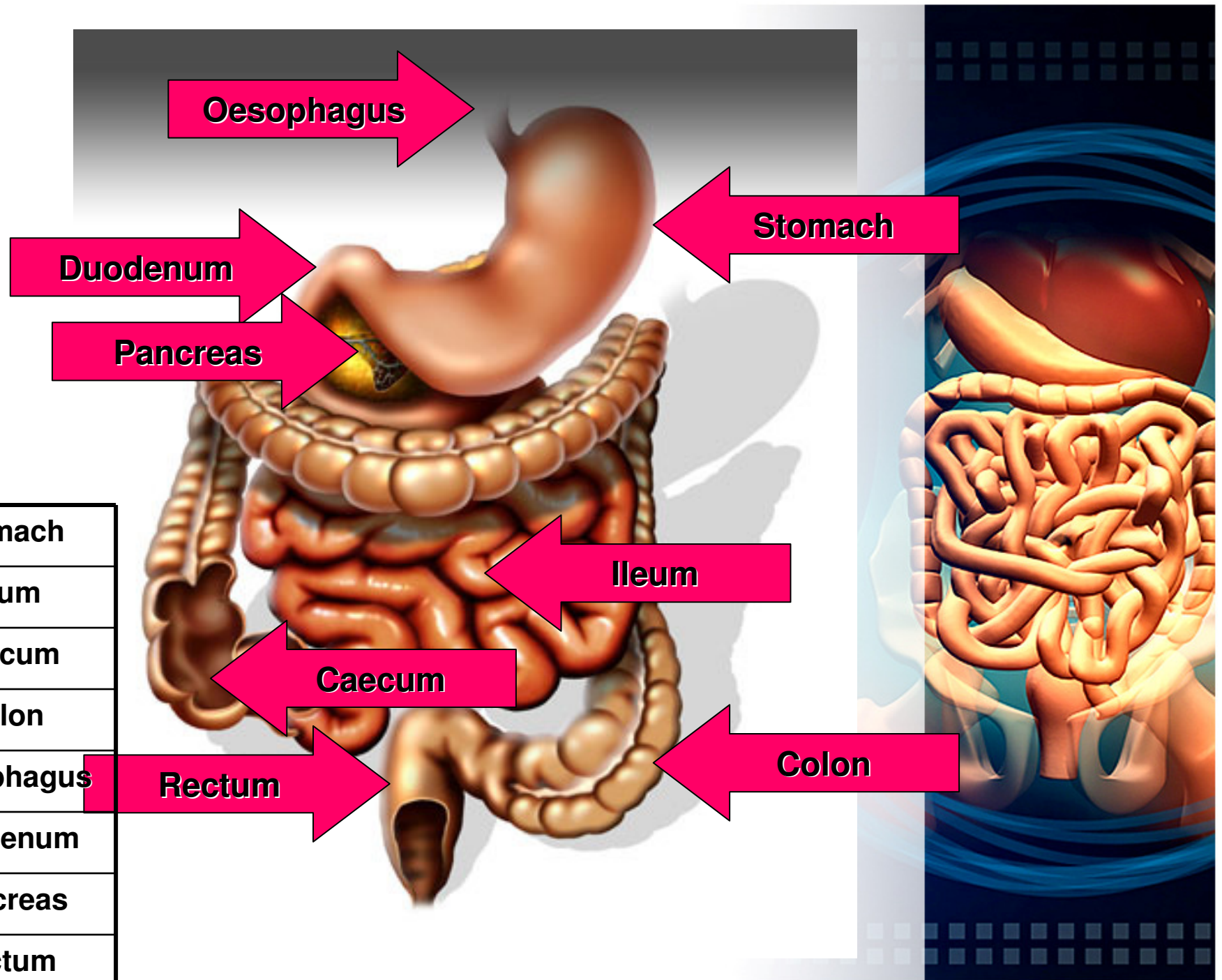
# Chromosomes

Chromosome 5

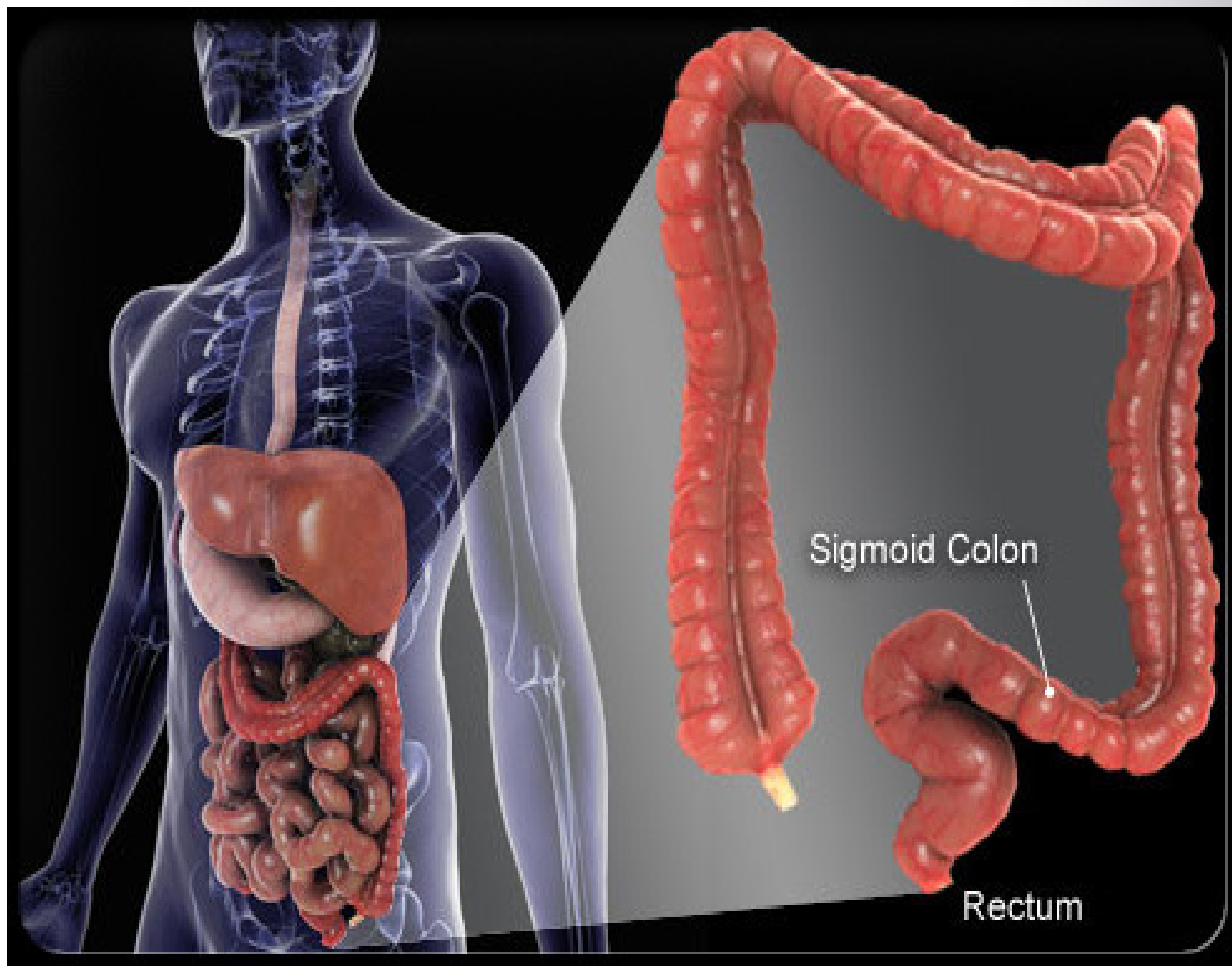


Sex Chromosomes  
XY = Male XX= Female



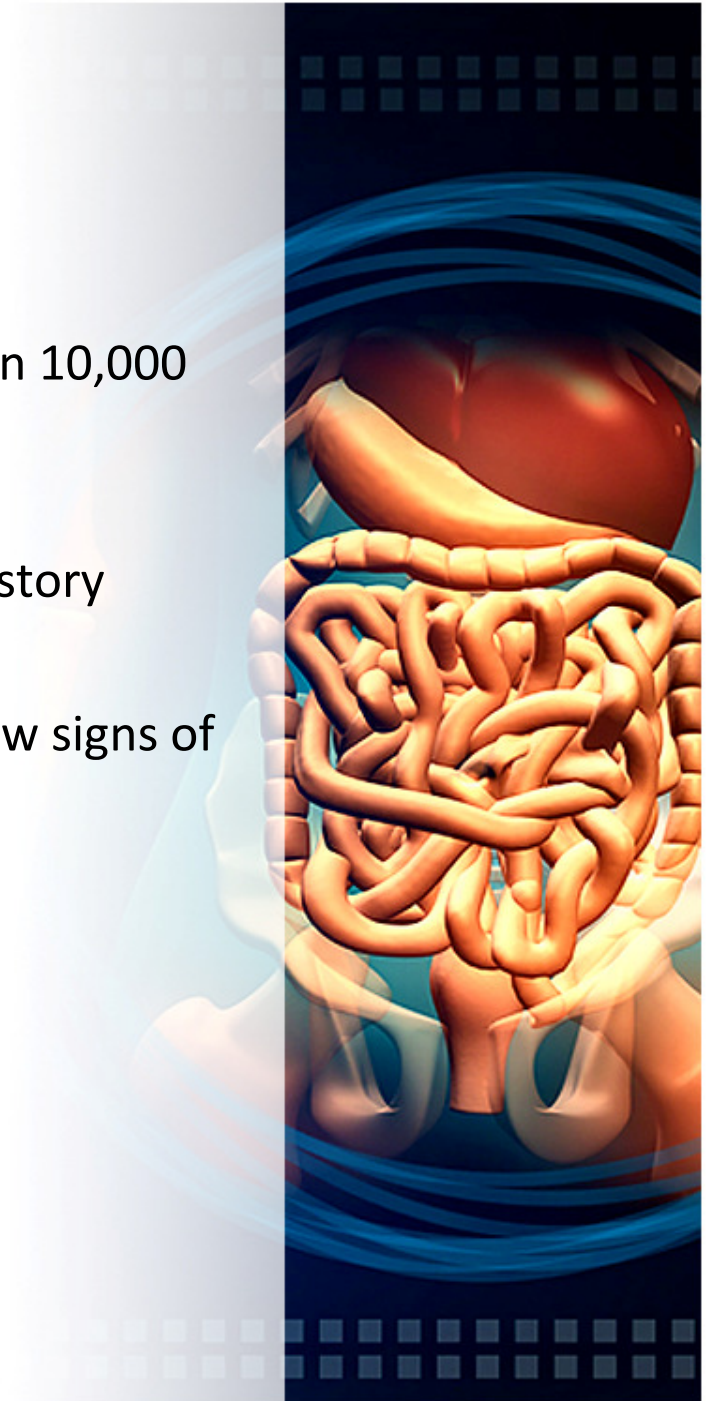


Stomach
Ileum
Caecum
Colon
Oesophagus
Duodenum
Pancreas
Rectum

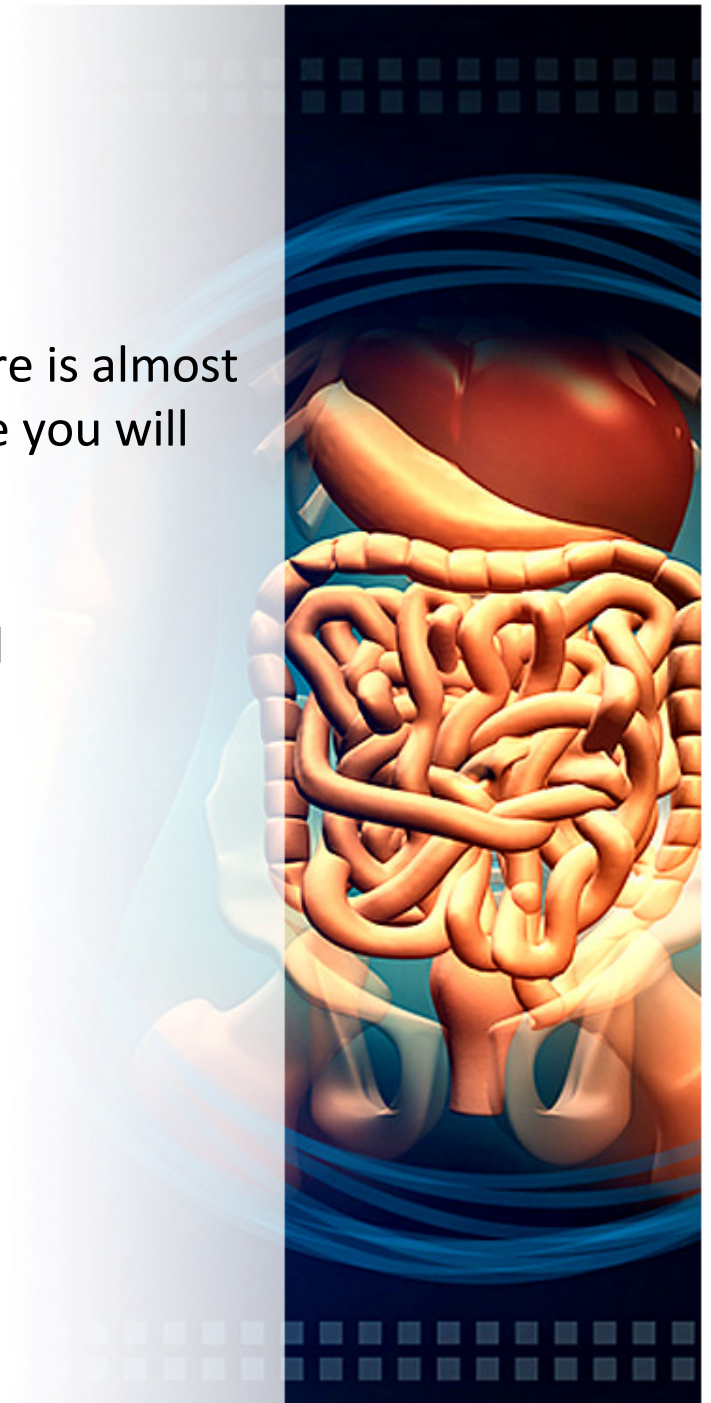


# Incidence

- It is thought that incidence of FAP is around 1 in 10,000 live births
- 10-30% of FAP type patients have NO family history
- Their genes may be the first to change and show signs of the disease.



- If you have FAP and you are left untreated there is almost a 100% chance that at some stage in the future you will get colorectal cancer!
- This can be avoided with regular screening and preventative surgery



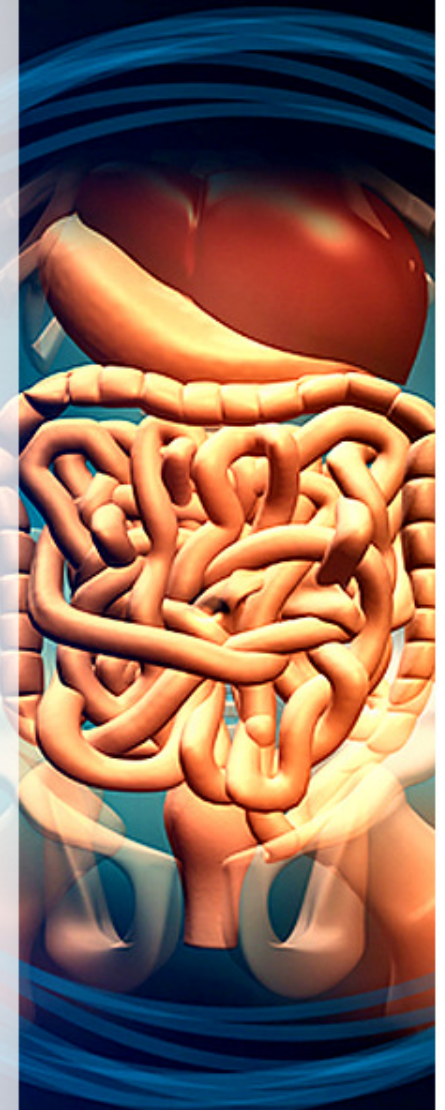
# So how can we tell if a patient has FAP?

- Firstly we listen to our patients
- 90% of the diagnosis should be made with the history

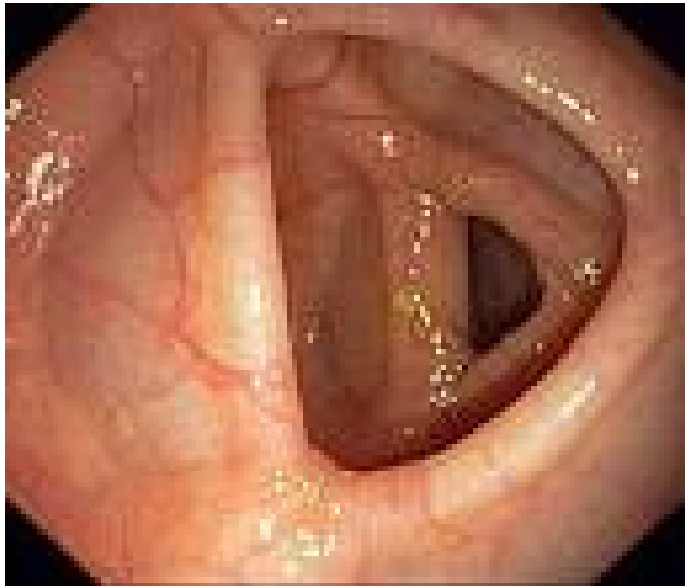
Symptoms include:

- Rectal bleeding
- Abdominal pain
- Diarrhoea
- Benign tumours (nose/face/scalp)
- Benign bone tumours (skull/lower jawbone)
- Polyps

**Family history**



# Normal endoscopy



# FAP endoscopy

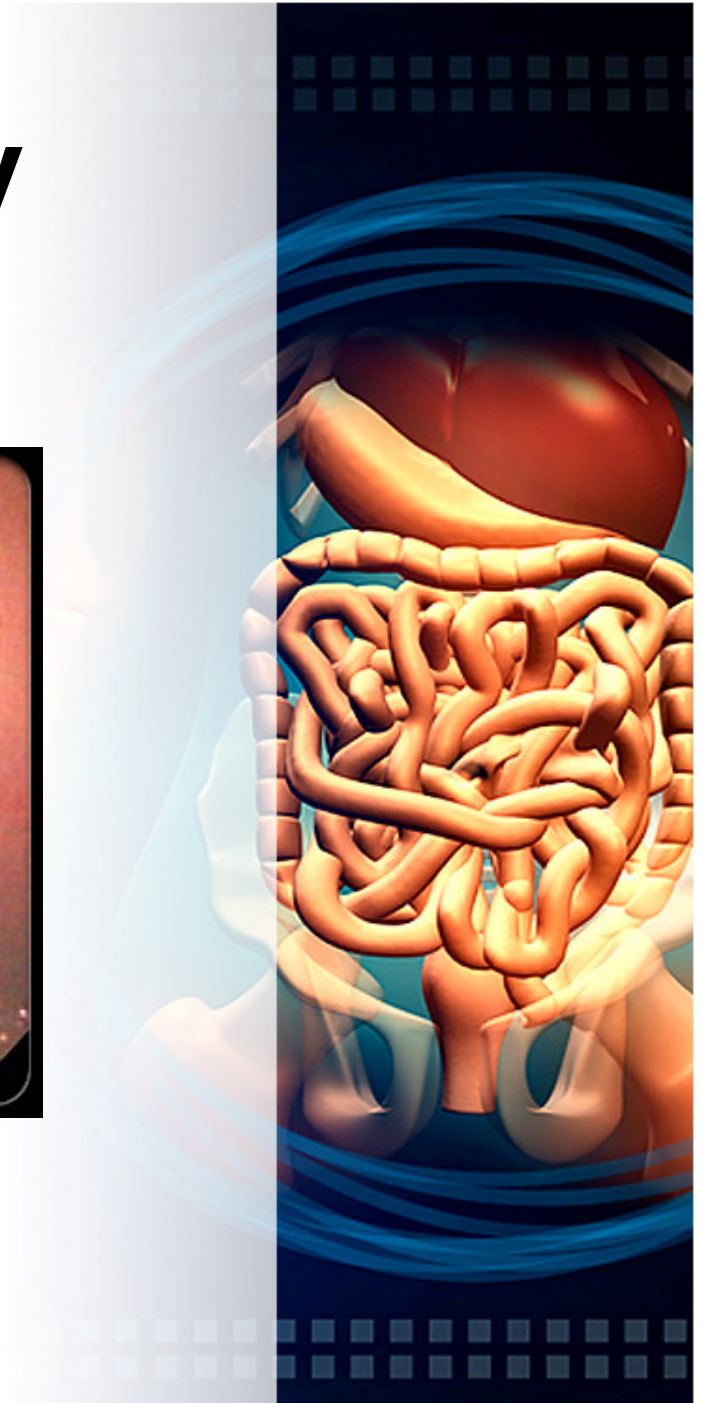
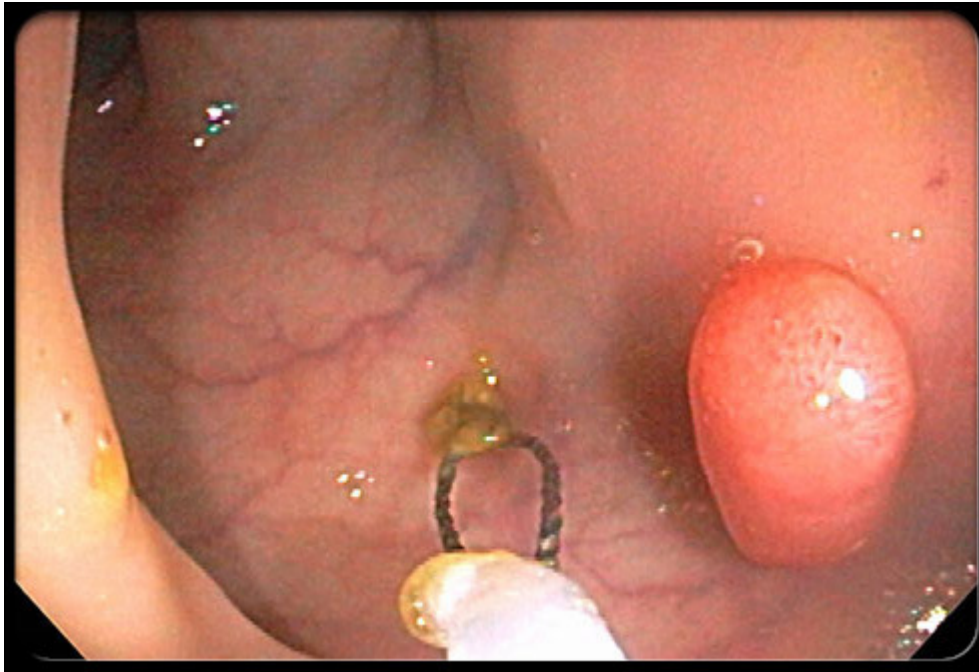


# What do we do if we find FAP?

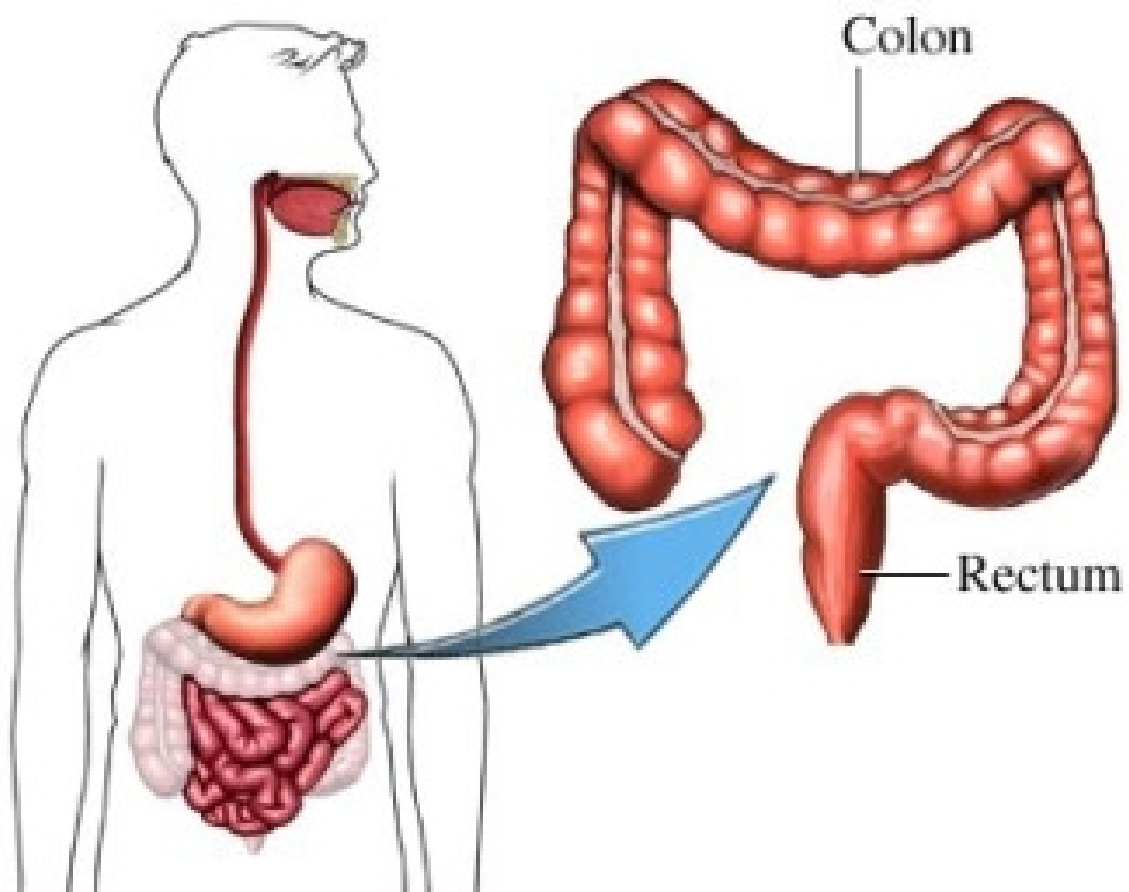
- Watch and wait with regular screening
- Therapeutic endoscopy
- Surgery



# Therapeutic endoscopy



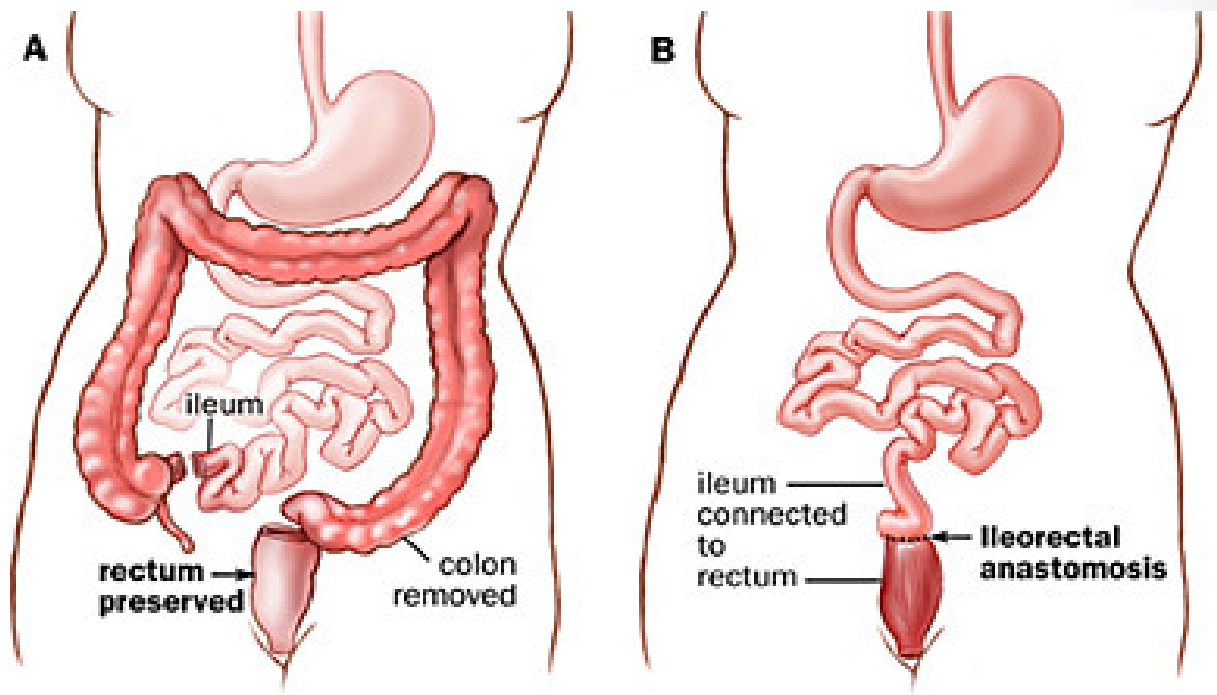
# Surgery



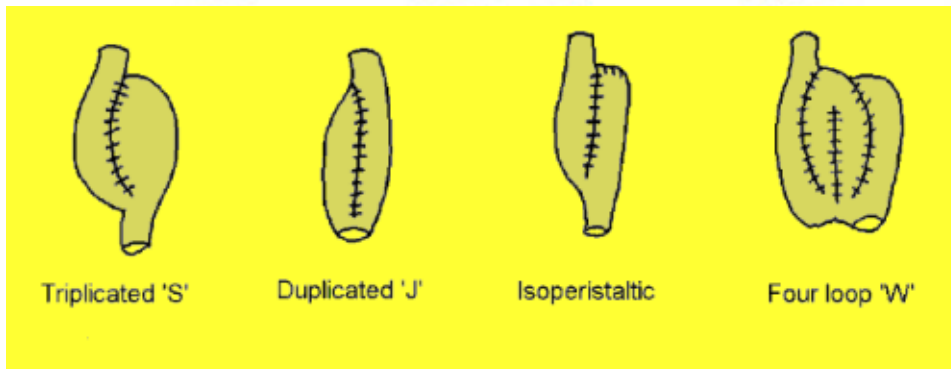
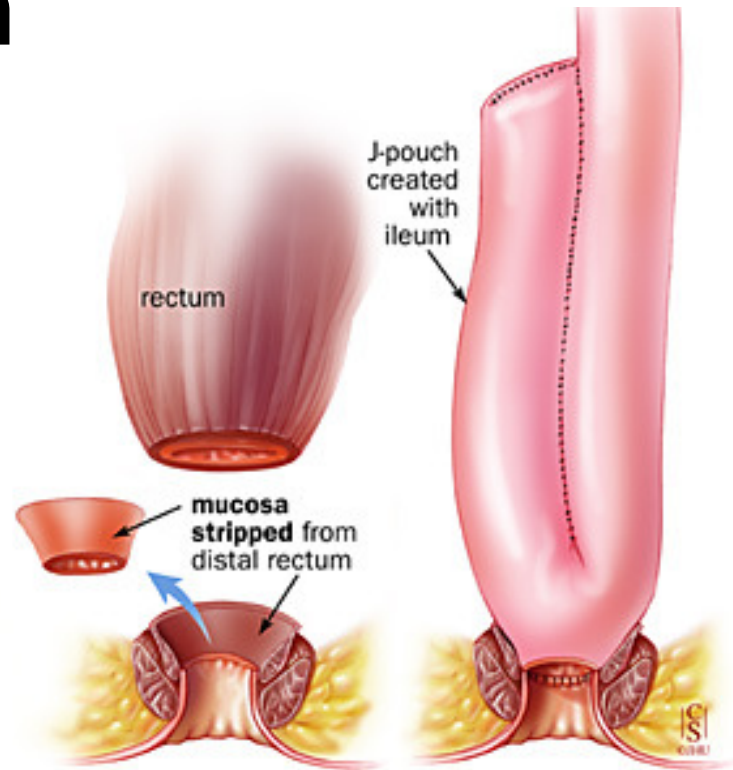
# Ileostomy



# Subtotal colectomy with ileorectal anastomosis

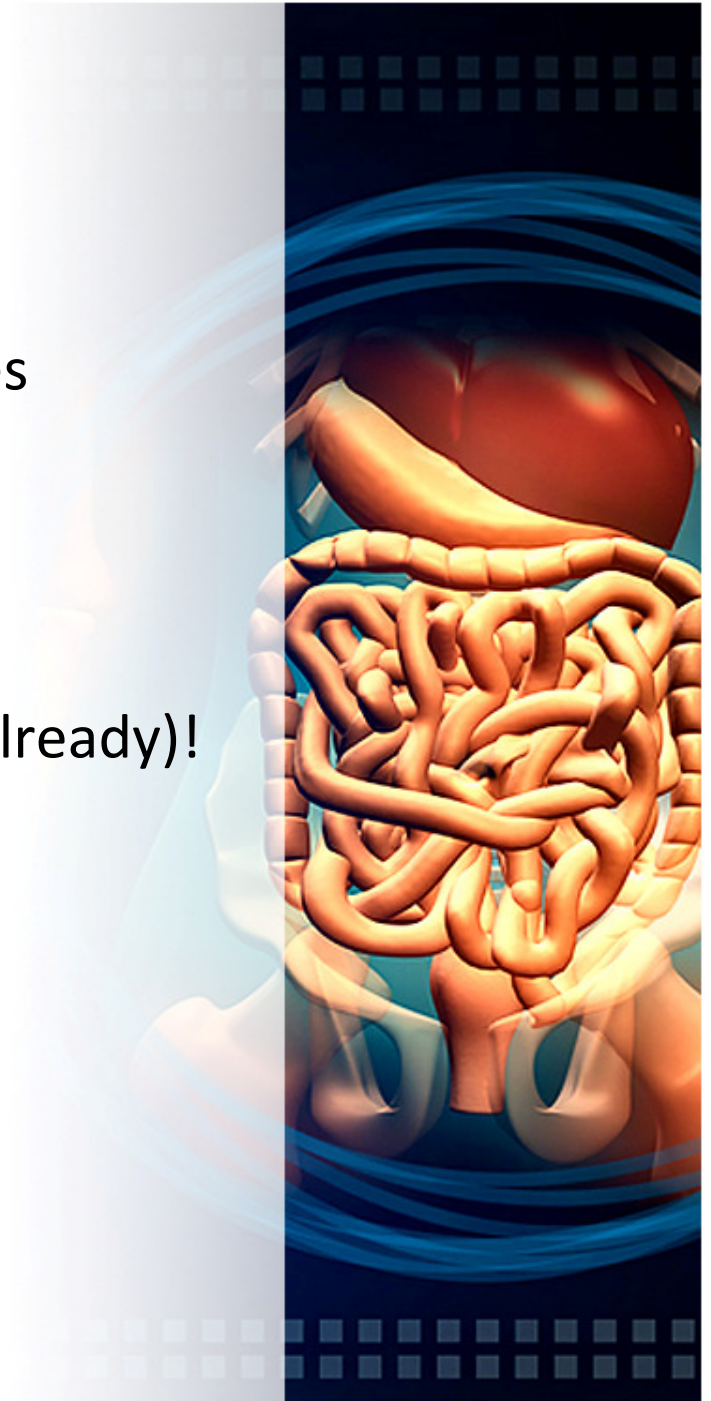


# Ileorectal anastomosis & pouch



# Future

- Regular screening upper GI endoscopies
  - Front viewing endoscope
  - Side viewing endoscope
- Contact a genetic service (if not done already)!
- Which leads me nicely onto...



**Any questions?**

