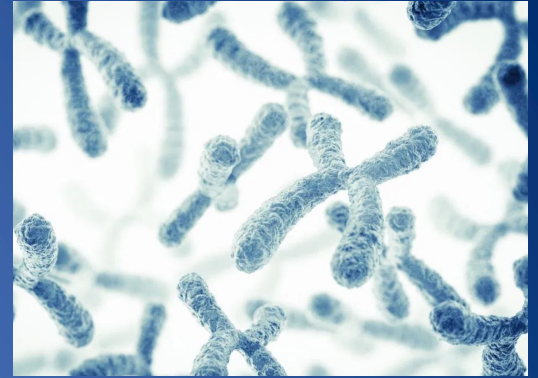


Insights into  
**MA**naging  
**G**rowth for  
**E**ndocrine  
**N**urses



# Explaining the diagnosis of Turner Syndrome to patients and families

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# Disclosures

- Honoraria
  - Springer IME
  - Novo Nordisk
  - Ipsen Ltd

# Introduction

- The different stages where a diagnosis of Turner Syndrome can be made
- The Nurse's role
  - Points to consider
- Disclosure of diagnosis to children and young people
- How to give a diagnosis to families
  - Communication
  - What to communicate
- What can Nurses do for the future

# Turner Syndrome diagnosis – prenatally

- Ultrasound abnormalities can be detected in TS, usually within the first trimester
  - Cystic hygromas
    - High risk for foetal outcome
    - Typically characteristic of Turner Syndrome
  - Foetal hydrops
  - Increased nuchal fold
  - Renal defects
  - Cardiac defects
  - Echogenic bowel



*Thayalan et al, 2018*

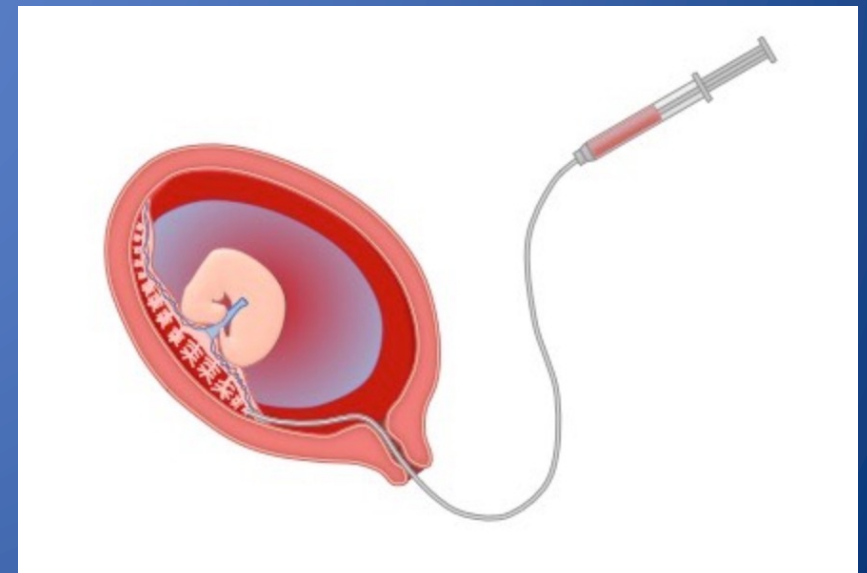
# Screening and testing

- Maternal serum testing
  - Combines nuchal translucency and serum biochemical markers
- Results from ultrasound and maternal testing will facilitate the need to determine formal karyotype

Amniocentesis



Chorionic villus sampling



# Genetic counselling

- Key factors are
  - WHO does the counselling
  - Parental perceptions
- Enhanced awareness about the improved post natal outcome should be reflected in prenatal counselling

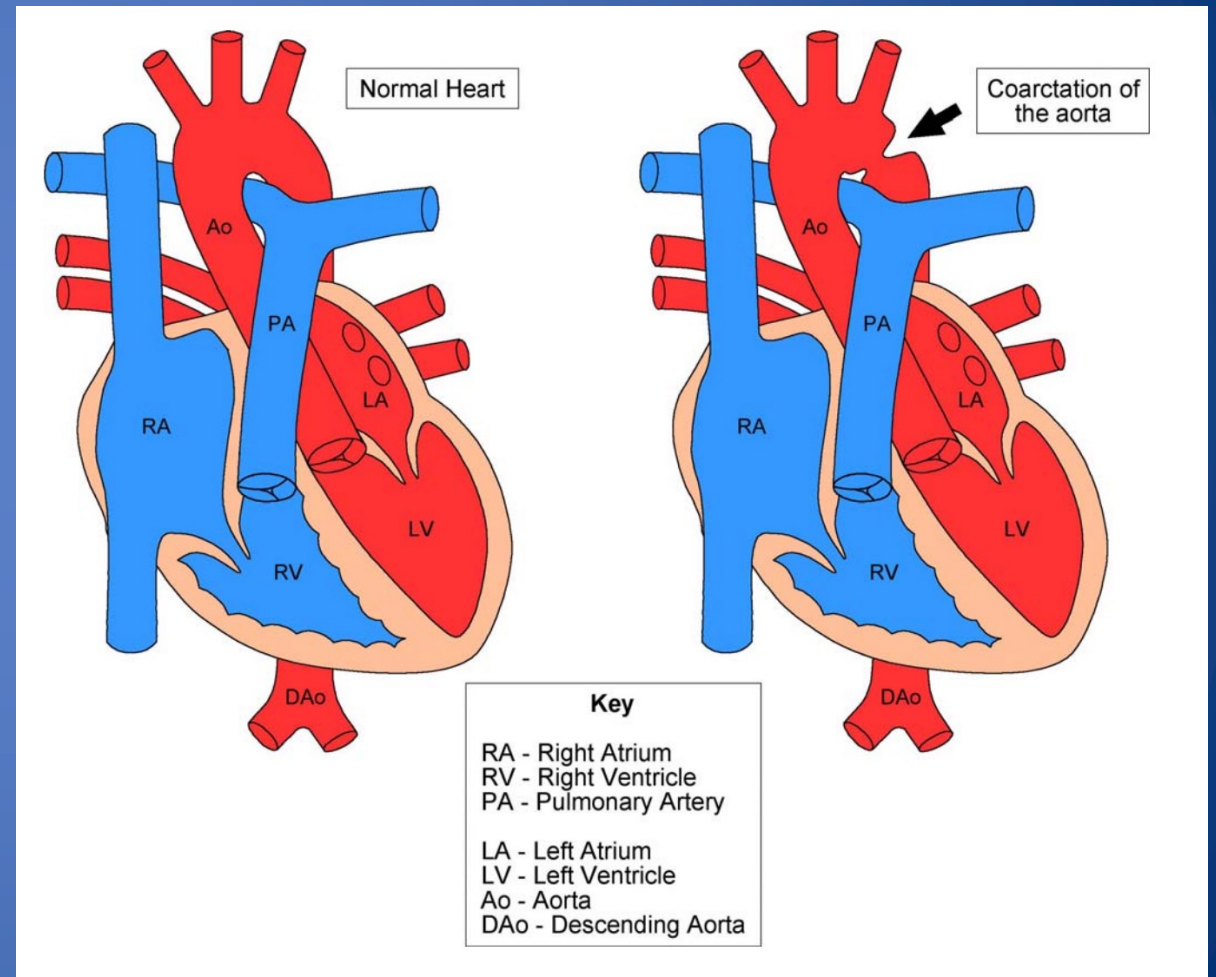
## Genetic Counselling

Being told that your baby or child has a genetic condition is extremely distressing and worrying. You will have so many questions that you want to ask about Turner syndrome. You and your family will need information, advice and support. This leaflet outlines just one source of excellent information and support - genetic counselling - and explains how you can obtain such help.

# Diagnosis in infancy

- Phenotype
  - Lymphoedema
  - Webbed neck (*Gravholt et al, 2017*)
- New genomic tests for postnatal diagnosis
  - Serum samples
  - Buccal swabs
- Cardiovascular screening
  - 5% of girls presenting with coarctation of the aorta have TS

(*Bondy, 2014*)



# Diagnosis in childhood

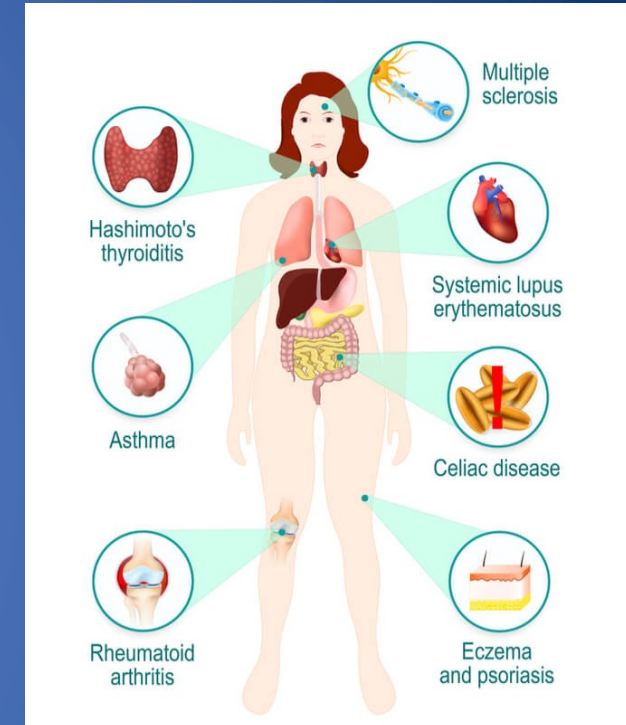
- Most girls diagnosed at this stage
- History of middle ear disease in a girl with short stature
  - ? TS diagnosis (*Massa, 2005*)
- Difference in height compared with friends becomes obvious in early childhood (*Culen et al, 2017*)
- Dependent on growth monitoring in different countries
  - Role of school nurse (*Ardary, 2007*)





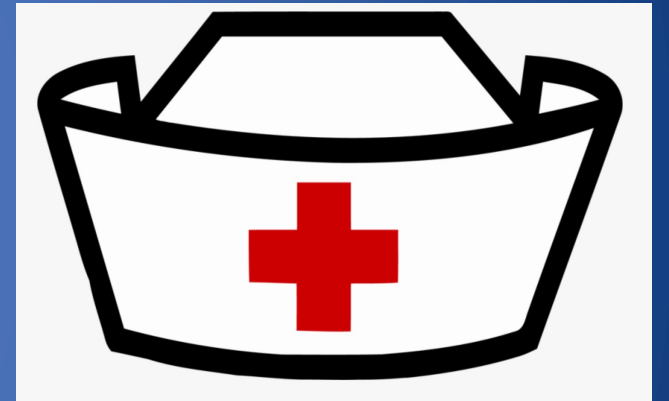
# Diagnosis in adolescence

- Short stature
- Delayed puberty
- Increased prevalence of autoimmune disease
- 20% of girls with TS are diagnosed between the ages of 12 – 18 years (*Apperley et al, 2018*)
- Psychosocial aspects
- Late diagnosis and late pubertal induction
  - Adverse outcomes
  - Behavioural traits can interfere with education (*Lee & Conway, 2014*)



# The Nurse's role

- Consider our unique role with girls and their families
  - Advanced skills
- Early intervention and support
  - Needs of child and parent
- Family / Cultural / Racial / Ethnic considerations
- Act as the patient's advocate
  - Follow the patient's directives
  - Explore their beliefs
  - Provide information on resources and treatment options



*(Duderstadt, 2014; Gerber, 2018)*

# Diagnosis to families

- Needs to be clearly communicated
- Honesty
- Provide written information
- Health care professional
  - Familiar with all aspects of TS
  - Inform about growth hormone therapy and pubertal induction
- Family counselling with multidisciplinary team
  - Guilt
  - Socio-economic class



*(Culen et al, 2017)*

# Disclosure to children



- Consider the best age to tell the child
  - Earlier knowledge gives children time to adapt gradually in steps appropriate to their age and emotional maturity
  - If girls have online access may start to research own symptoms (*Turner & Hozjan, 2019*)
- Honesty is the best policy
- Timely, caring and sensitive manner
- Waiting for the 'right time'
  - May become more difficult as time goes on
    - Signs and symptoms
    - Active treatment

# Recommendations at Infancy

- Encourage positive child / parent attachment
- Commence TS specific growth charts
  - Reduces guilt
- Become attuned to normal sleeping and feeding habits
  - Irregularities stressful (*Culen et al, 2017*)



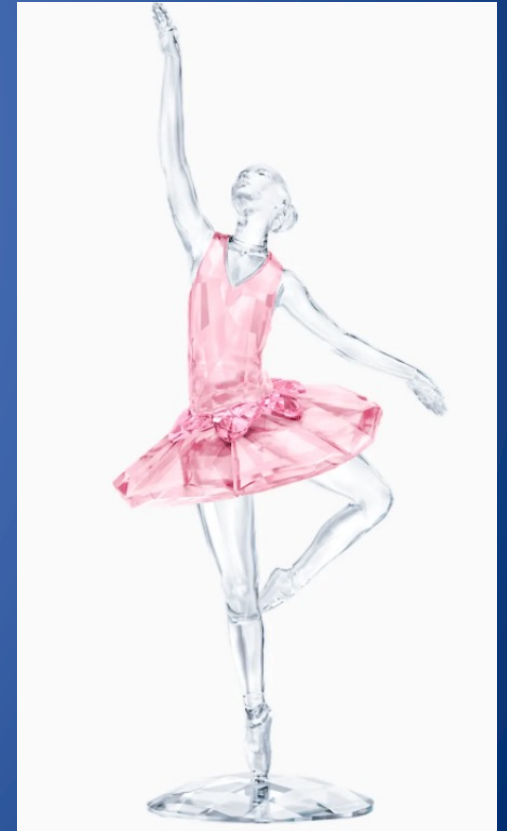
# Recommendations in early childhood 2 – 6 years

- Short stature
    - Psychosocial and Auxological factors need to be considered
    - Growth hormone therapy
  - Childhood developmental stages
  - Psychological counselling
  - Occupational therapy
    - Sensory processing
    - Functional / coping skills
- (Culen et al, 2017)*



# Recommendations in later childhood 6 – 12 years

- Reactions and comments from friends and peers regarding short stature
  - Open discussion with paediatrician, child and parents
- Consider possible motor deficits
  - Encourage activities
- Explore school matters
  - Educational psychologists / extra time
- Social behavioural interactions (*Culen et al, 2017*)



# Recommendations in adolescence

- Need for oestrogen substitution
- Ovarian insufficiency information
- Progress in educational and vocational planning
  - Cognitive and psychosocial development evaluation essential
- Discussions surrounding relationships, sexuality, fertility
  - Healthcare professionals to provide information
- Autonomy and transition
  - Gynaecology (*Culen et al, 2017*)





# Communicating the diagnosis – why and how

- Frequent but stressful task
- Patients and families want the truth
- Ethical and legal implications
- Clinical outcomes
  - Consider HOW it is discussed
    - Understanding
    - Patient satisfaction
    - Psychological adjustment
    - Medication adherence
- Follow a six step process: **SPIKES**  
*(Baile et al, 2000)*



# 1 – Setting up the consultation

- Mental preparation
  - Review the plan
  - Predict the emotional response
- Ensure privacy
- Involve significant family members
- Connect with the family
  - Eye contact
  - Establish rapport
- Manage time constraints / interruptions



## 2 – Perception of the patient / family

- Find out how the family perceives the medical situation
  - Opportunity to correct mis-information
  - Tailor the bad news
- Understand their engagement
  - See if there is any denial
- Explore health literacy



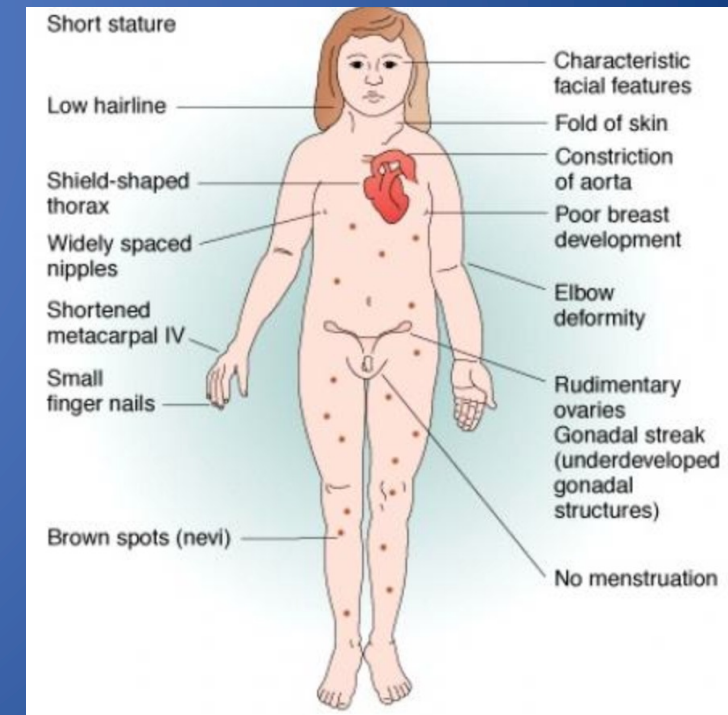
– “The motivation and ability of individuals to access, understand and use information in ways which promote and maintain good health” (*Rowlands et al, 2015*)

– Consider those at risk

- Risk for error and poorer health outcomes (*Graham & Brookey, 2008*)

# 3 – Obtaining the Patient's Invitation

- Most patients want ALL the information
  - Some do not..
- May depend on who is present in the consultation
- The amount of information given may lessen the anxiety
  - Desire for less information?
- Consider where to start



# 4 – Giving Knowledge and Information

- Pre-empt the discussion with an introduction to facilitate information processing
  - *“So we now know what the blood results are”*
- Giving medical facts could be improved
  - Start at the level of patient understanding
  - Do not use technical words
  - Avoid excessive bluntness
    - *“Your daughter is infertile”*
  - Give the information in chunks and check understanding regularly



# 5 – Addressing the Emotions with Empathy

- Reactions may vary
  - Silence
  - Disbelief
  - Crying
  - Denial
  - Anger



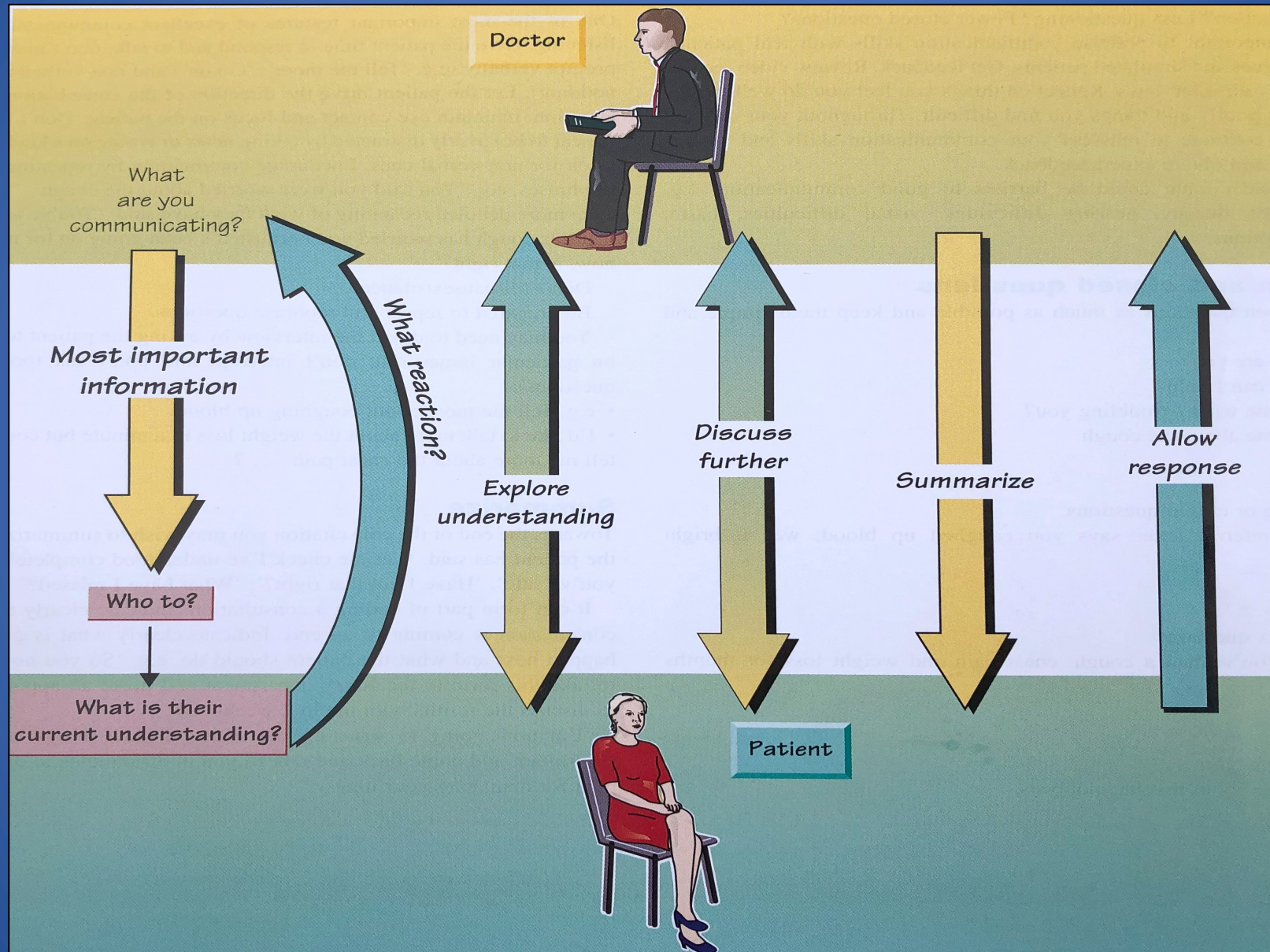
- Observe for emotion
  - Tearfulness / shock
- Identify the emotion
  - Verbalise to patient / parent
- Discuss the reason for the emotion
- Deal with the reaction
  - Wait for calm before continuing

# 6 – Strategy and Summary

- Clear plan for the future
  - Less anxious and uncertain
- Discuss treatment options
  - Share decision making – give patient autonomy and responsibility



# Communicating information





# The future role of the paediatric endocrine nurse

- Explore opportunity for advanced skills
  - Physical assessment
  - Mental health
  - Genetic counselling
  - Communication
- Patient support groups
  - Vital in providing support outside the clinic environment
  - Belonging to a peer support can have a positive impact for the girl and her family  
(Gravholt et al, 2017)

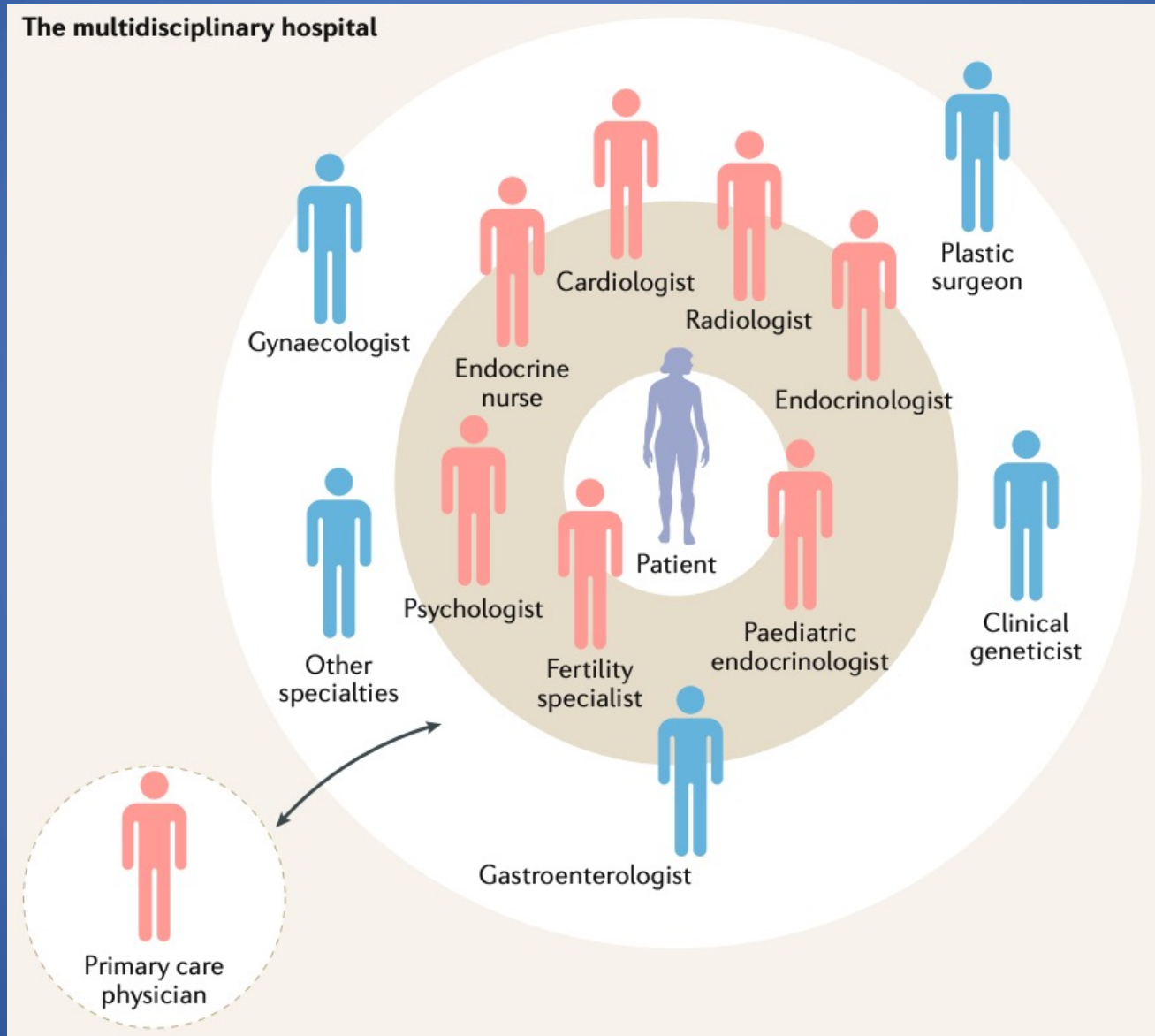


# Support groups

- UK
  - [www.tss.org.uk](http://www.tss.org.uk)
- America
  - [www.turnersyndrome.org](http://www.turnersyndrome.org)
- Canada
  - [www.turnersyndrome.ca](http://www.turnersyndrome.ca)
- France
  - [www.turneretvous.org](http://www.turneretvous.org)
- Australia
  - [www.turnersyndrome.org.au](http://www.turnersyndrome.org.au)



# The multidisciplinary team



# Conclusion

- Many factors to consider
- Thorough planning needed
- Multidisciplinary team
  - Paediatric Endocrine Nurse
- Patient support groups



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