Congenital Anomalies of the Genital Tract

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Should Paediatric and Adolescent Gynaecology be Centralised?

VECKAN 2015

- Disclosure of interests:

- There is no conflict of interest to declare

What does Paediatric and Adolescent Gynaecology cover?

- Depends on your country's health system

UK: Primary care → Secondary Care → Tertiary Care

USA: Direct Access to Secondary Care → Tertiary Care

European countries: varies

Sweden: ?? Same as UK

Organization for Economic Co-operation and Development (OECD) December 2013

- Sweden is best in the world for health outcomes

- HOWEVER: “Biggest challenge is the issue of coordinating care between hospitals, primary care and local authorities

What constitutes PAG Care?

- Neonates and infants
- Children
- Adolescents
- Transition to adult services and continued care

UK System

- Neonates and infants

Paediatric endocrinology

Paediatric urology

Paediatric surgeons

Geneticists

Correction of disorders of sexual development in specialist centres.

Really no input from gynaecologists

UK system

- Children

Paediatric Endocrinology, Surgery and Urology

Disorders of growth and development inc puberty in children's specialist hospitals

UK System

- Adolescents
- Primary care
- Paediatric endocrinology
- Gynaecology – complex holistic care

Problem

- Care is now fragmented
- Multiple specialties usually in different institutions
- Consultants not specialists
- Access to specialist nurses, specialist psychology etc becomes very difficult
- Funding issues

UK System

- Transition to adult services

- Usually very poor - non-specialist doctors whether surgeons, urologists, endocrinologists or gynaecologists.
- Major lack of holistic approach to care when needed.

UK Primary Care

- Menstrual disorders
- PCOS
- Contraception
- (No cervical smears before age 25)

Only refer when unable to cope

UK Gynaecology

- Nearly all patients seen in general gynaecology out-patient clinics ie with adults. May be seen by a junior doctor.
- Menstrual disorders and Amenorrhoea
- PCOS
- Genital injuries
- Pelvic pain and endometriosis
- Congenital anomalies
- Unwanted pregnancy

UK Special Clinics

- Adolescent Gynaecology clinics
- Sexual health
- Child/Adolescent Sexual Abuse
- Female Genital Mutilation – new issue!!
- Family Planning Clinics
- Abortion Services
- Premature Menopause

USA

- Same system as UK for neonates, infants and children

- Adolescent system relies entirely on gynaecologists
- Referral rate to specialist care very low – so care very poor
- Distance is a major issue

What services should be in specialist centres?

- Specialist Gynaecologists – PAG trained
- Endocrinologist with interest in disorders of puberty, CAH, induction of puberty AND able to continue care into adulthood
- Psychologists – specially trained
- Clinical nurse specialists
- Specialist radiologists
- Geneticist if needed
- Access to reconstructive surgeons/urologists

Do all patients need to be referred?

- Absolutely not.

- Referral pathways for those conditions best cared for in the centre.
- National centre to optimize care
- Locally recognised gynaecologists to be part of a network to be involved in long term care
- National organisation to establish standards of care and monitor outcomes
- This approach delivers the best care for patients.

How does this translate in practice?

Centre for Disorders of Sexual Development and Adolescent Gynaecology

- National Centre at QCCH since 1999.
- Excludes surgical problems in children under age 12.
- Focus on holistic, multidisciplinary care.

Spectrum of problems

3 main groups

- Disorders of sexual development
  - MRKH syndrome
  - Outflow tract obstruction
  - Uterine anomalies
  - 46XY DSD
  - Turner's syndrome
  - CAH in adolescents
  - Children with vulval abnormalities

- Adolescent gynaecology
  - Disorders of puberty
  - Menstrual abnormalities
  - Complex Contraception

- Endocrine problems

  Primary amenorrhoea

  Secondary amenorrhoea

  PCO

  Hirsutism

Multidisciplinary team

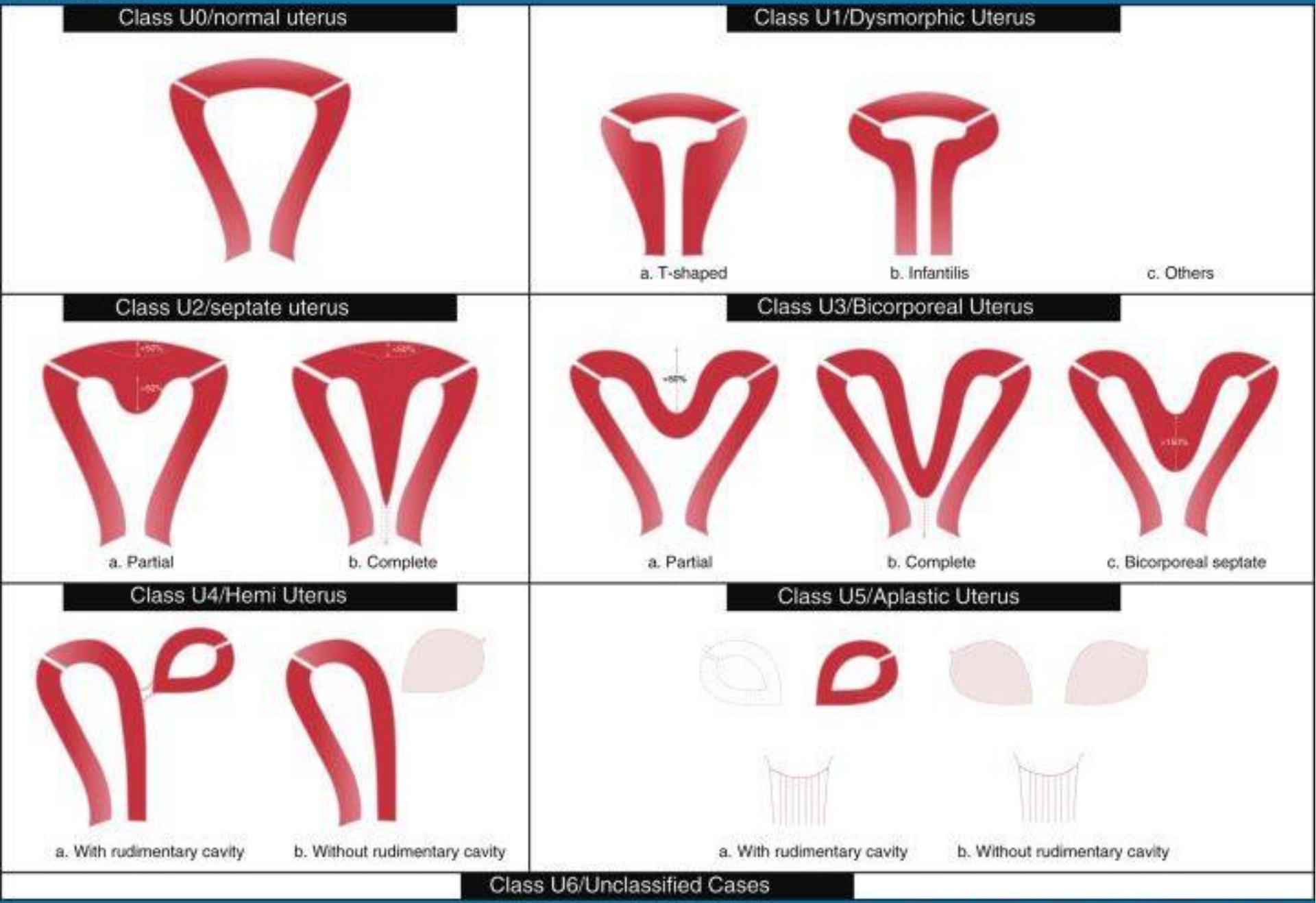
- Psychologist
- Specialist imaging expertise
- Self help group for MRKH
- Endocrinologist
- Gynaecologists

Congenital Malformations of the Genital Tract

- Classification

Numerous different attempts – all variations on the same themes

ESHRE/ESGE classification 2013



ESHRE/ESGE classification
Female genital tract anomalies

	Uterine anomaly		Cervical/vaginal anomaly
	Main class	***Sub-class***	***Co-existent class***
U0	Normal uterus		
U1	Dysmorphic uterus	**a.** T-shaped **b.** Infantilis **c.** Others	
U2	Septate uterus	**a.** Partial **b.** Complete	
U3	Bicorporeal uterus	**a.** Partial **b.** Complete **c.** Bicorporeal septate	
U4	Hemi-uterus	**a.** With rudimentary cavity (communicating or not horn) **b.** Without rudimentary cavity (horn without cavity/no horn)	
U5	Aplastic	**a.** With rudimentary cavity (bi- or unilateral horn) **b.** Without rudimentary cavity (bi- or unilateral uterine remnants/aplasia)	
U6	Unclassified malformations		
U			***C*** ***V***

C0	*Normal cervix*
C1	*Septate cervix*
C2	*Double 'normal' cervix*
C3	*Unilateral cervical aplasia*
C4	*Cervical aplasia*
V0	*Normal vagina*
V1	*Longitudinal non-obstructing vaginal septum*
V2	*Longitudinal obstructing vaginal septum*
V3	*Transverse vaginal septum and/or imperforate hymen*
V4	*Vaginal aplasia*

Associated anomalies of non-Müllerian origin:

Drawing of the anomaly

- Could be useful if everyone used it and reported all findings to a central resource within ESHRE but so far not found widespread uptake.

- Any research study would benefit as all researchers would use the same classification.

MRKH

- Second commonest cause of primary amenorrhoea after Turner’ s syndrome.

- Differential diagnosis

  XY DSD - Androgen insensitivity

  absence of pubic/axillary hair

INCIDENCE

- 1 in 5000 female births

  based on Finnish study

  (Aittomaki et al 2001)

- No reliable data for other populations

DIAGNOSIS

- Clinical
- Imaging – Ultrasound

  MRI
- Laparoscopy is un-necessary in the majority of cases

ASSOCIATED CONGENITAL ABNORMALITIES

- Renal agenesis 30%
  - Horseshoe kidney 5-10%
  - Pelvic kidney 1%
  - Duplication of ureters
- SKELETAL ANOMALIES 12%
  - Spine 60%
  - Limb
  - Rib
- URINARY TRACT ANOMALIES 40%
- HEARING IMPAIRMENT up to 10%

Classification

- Typical – sole anomaly (64%)
- Atypical – typical + renal/skeletal/hearing or other anomalies (24%)
- MURCS –Müllerian aplasia, renal aplasia and cervicothoracic somite dysplasia (12%)

Oppelt *et al* 2006

PSYCHOLOGICAL FACTORS

- SHOCK
- DEPRESSION
- DOUBTS OF GENDER
- INFERTILITY
- SEXUALITY
- WORTHLESSNESS
- CULTURAL DIFFICULTIES

PARENTAL PROBLEMS

- DEPRESSION
- FEAR
- IGNORANCE
- ACCEPTANCE OF INFERTILITY
- PATERNAL SUPPORT

IMPORTANCE OF PSYCHOLOGICAL PREPARATION

- Dealing with adolescent stress and difficulties
- Dealing with sexuality
- Dealing with support mechanisms
- Success of interventions is directly related to psychological success

Surgical Management of MRKH Syndrome

- Vulvoplasty — William's operation
- Bowel Vaginoplasty — Ileum
  Sigmoid colon
  Caecum
- Vecchetti's Operation — Laparotomy
  Laparoscopic

Success of Surgical Techniques

Vulvoplasty		95%
Vaginoplasty	Amnion	84%
	McIndoe	92%
	Davidov	88%
	Sigmoid	88%
	Vecchetti	95%
	Buccal Mucosa	90%

NON-SURGICAL MANAGEMENT

VAGINAL DILATORS

- Repeated use of graduated vaginal dilators
- Careful instruction
- 3 Times daily for 20 minutes for 2-3 months

Various Vaginal Dilators

Results of Dilator Therapy

Author	Patient Nos	Success
Rock et al (1983)	21	18 (86%)
Broadbent et al (1984)	20	19 (95%)
Roberts et al (2001)	51	46 (91%)
Gargollo et al (2009)	57	50 (88%)
Total	149	133 (89%)

Results of Dilator therapy

- Edmonds et al 2012
- Experience of **245** consecutive patients with MRKH over 12 years

  232 (95%) anatomical/functional success

- 13 did not complete therapy:

  Psychiatric or cultural issues only

- Therefore **100%** success if therapy completed.

Updated results to 2015

- Total number of patients completing dilator program **330**

- Success rate **100%**

MRKH SURROGACY

- Widely accessed
- No international data base
- Few reported series
- Success similar to IVF

- No reported female offspring with MRKH

Uterine transplantation

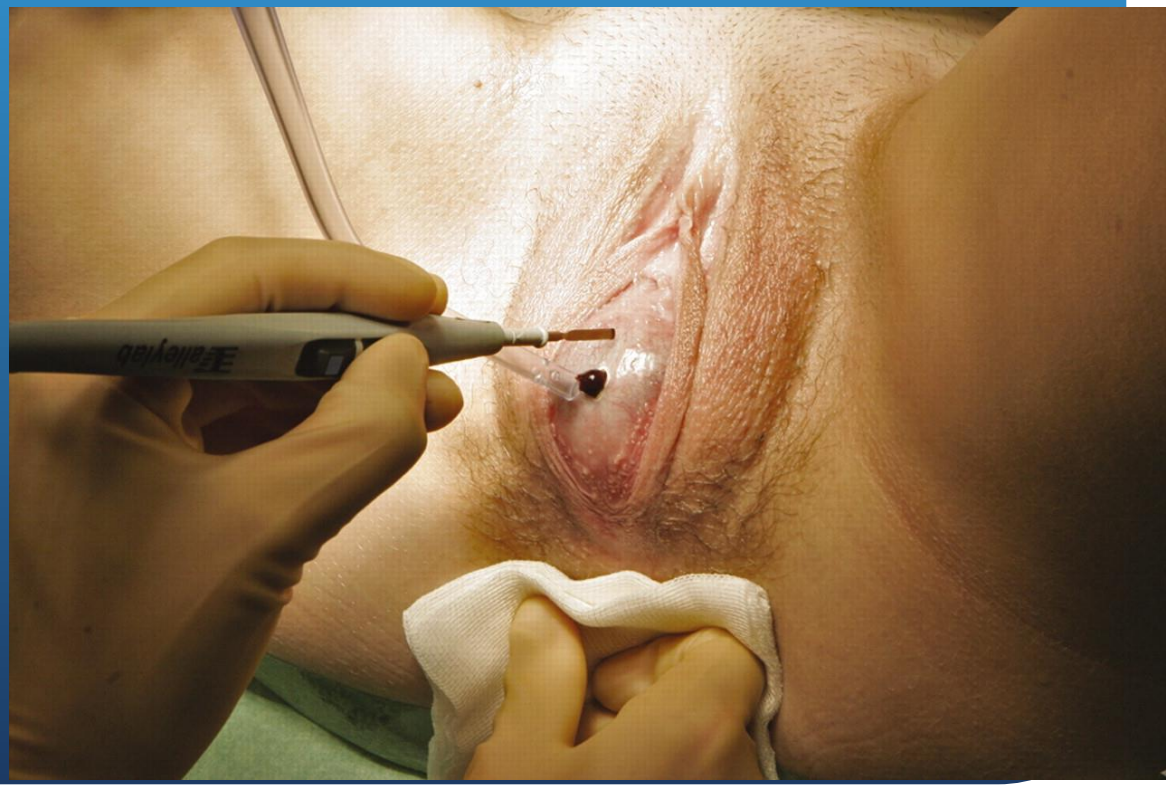
- Controversial
- Ethical issues re non-life saving transplantation
- Costs within a nationally funded health system and competing resources
- Long term health issues for recipient, donor and offspring remain unknown

IMPERFORATE HYMEN

Bulging and "blue"

Management

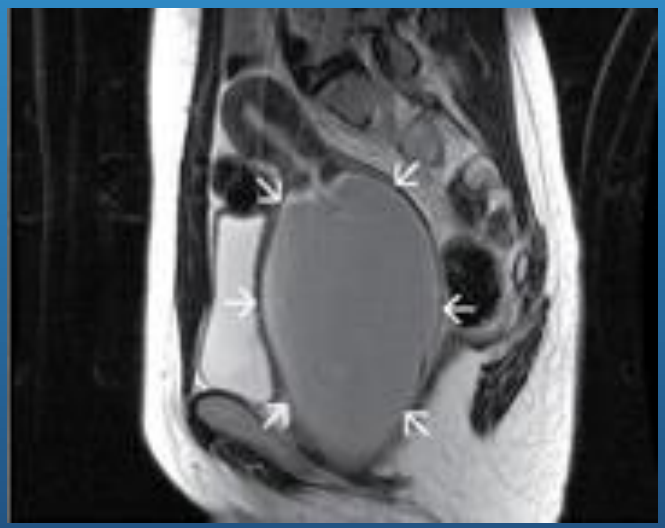
- Incision and drainage
- No sequelae



TRANSVERSE VAGINAL SEPTUM

- **CYCLICAL ABDOMINAL PAIN**
- **PRIMARY AMENORRHOEA**
- **SECONDARY SEXUAL CHARACTERISTICS PRESENT**

MRI

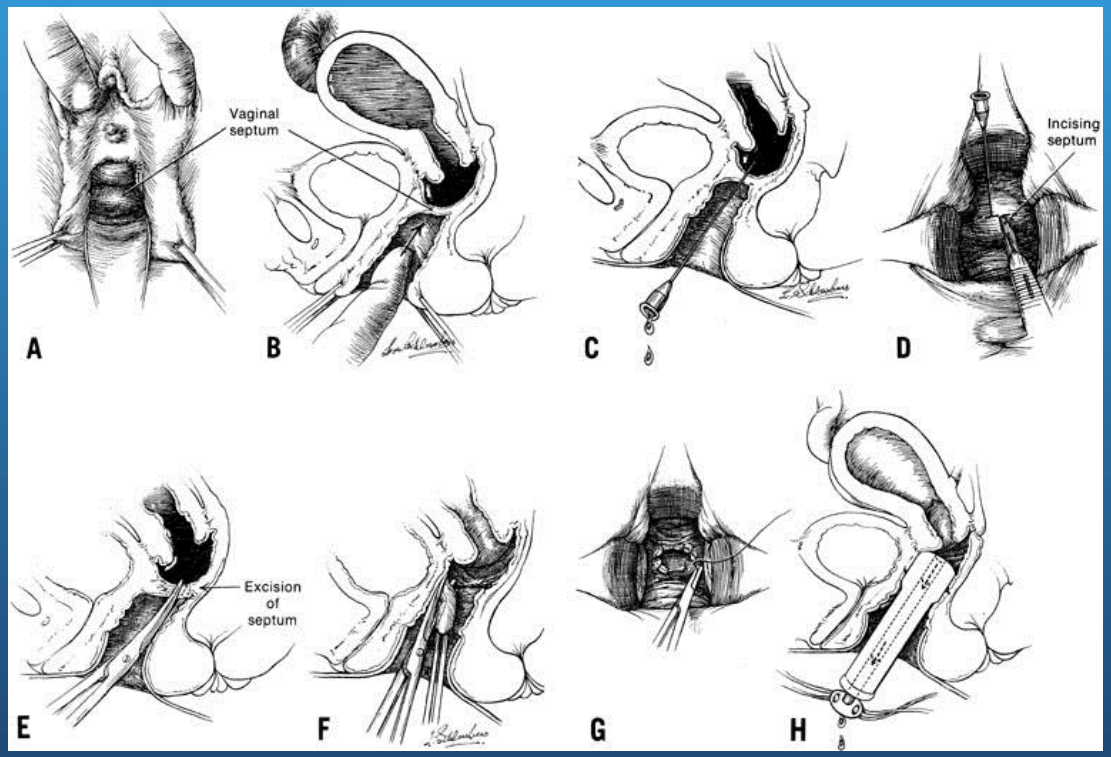


Levels of Obstruction

- a – high
- b – middle
- c - low

Surgery

- Excision of septum and vaginal advancement
- Post operative dilators/mould



OUTCOME

- Sexual function

  10% dyspareunia rate for low septae

  40% for high septae

- Endometriosis
- Reproductive performance

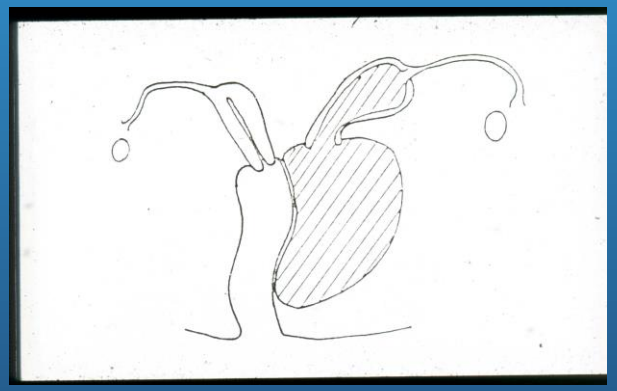
  100% for low problems

  20% for high obstructions

LONGTITUDINAL VAGINAL SEPTUM

- **DOUBLE VAGINA**
- **OBSTRUCTED**

  HEMI-VAGINA



DOUBLE VAGINA

- **DIFFICULTY USING TAMPONS**
- **DYSPAREUNIA**
- **ANTE-NATAL DIAGNOSIS**
- **INTRA-PARTUM DIAGNOSIS**

Management

- Excision surgery
  - Ligation
  - Diathermy
  - Laser
  - Harmonic scalpel
- Complications
  - Haemorrhage, infection
  - Dyspareunia

Need for a Swedish Society of Paediatric and Adolescent Gyneacology

- Education
- Training
- Research
- Communication - Professional, patients, parents
- Information
- Clinical standards
- Professional Opinion

Current Issues

- Female Genital Mutilation (FGM)
- Labial reduction
- Sexual abuse
- Childhood and adolescent obesity
- Athletic Triad
- Onco-fertility