E10/S/c

**NHS STANDARD CONTRACT
FOR COMPLEX GYNAECOLOGY: CONGENITAL GYNAECOLOGICAL ANOMALIES**

PARTICULARS, SCHEDULE 2 – THE SERVICES, A - SERVICE SPECIFICATIONS

Service Specification No.	E10/S/c
Service	Complex Gynaecology : Congenital Gynaecological Anomalies
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

- **Population Needs**

  - **National/local context and evidence base**

- Complex Congenital Gynaecological Anomalies is the gynaecological management of rare congenital anomalies of the genital tract. In some cases, these rare conditions may also present in adult life. Some that present in adolescence will need long term adult gynaecological management.

- Multi-professional and multi-disciplinary input is required either because the patient has co morbidities which render the care of her gynaecological disorder especially complex e.g. endocrine problems such as Congenital Adrenal Hyperplasia or because she may require complex treatment including reconstructive surgery to optimise future gynaecological, psychological and reproductive outcomes. This is best provided by a specialist centre co-located with other specialised services, such as paediatric gynaecology, urology, radiology and genetics.

- Units that provide this service will also need defined links to other definition sets and should be co-located to the relevant services. The Provider will deliver high quality services within an agreed network of Providers whose population may also use this service.

- Accurate incidence figures are not available but it is estimated that these

conditions overall affect 1 in 2-3000 females.

- **Scope**

- **Aims and objectives of service**

- The aim of the services is to provide patient centred specialist care for children, adolescents and adult women with a congenital abnormality of their genital tract and promote family centred specialist care and to optimise future reproductive function and quality of life.

- The primary aims are:
- To provide a safe and effective care pathway for women with congenital anomalies of the genital tract.
- To provide social, economic and psychological benefits for children, adolescents and their family requiring the service
- To provide continuity of care through the whole care pathway encompassing other specialised services included within the pathway including paediatric and adult services

- **Service description/care pathway**

- Services will provide the defined activities outlined below as part of a multidisciplinary team approach associated with interdependent services. Patients will follow a care pathway as described below and in the appendix.

- The patient journey will include:
- Referral from GP or secondary care
- Investigations and diagnosis
- In patient or outpatient treatment
- Outpatient follow up
- Discharge where appropriate

- The multi-disciplinary team wil include:
- Co-located:
- Gynaecologist
- Psychologist
- Specialist nurse
- Interdependent:
- endocrinologist, urologist, biochemist, geneticist, plastic surgery, radiologist as required

- **Management of congenital anomalies of the genital tract**
- **This includes 2 main diagnostic groups:**

- Disorders of Sex Development (DSD). Older terminology now abandoned byclinicians (but still present in ICD classification) includes intersex, indeterminant sex and hermaphrodite/pseudohermaphrodite.
- Complex Mullerian Anomalies. This includes Rokitansky syndrome (mullerian agenesis) and mullerian obstruction/duplication. Some complex mullerian anomalies are associated with complex urological anomalies such as bladder extrophy and cloacal anomaly.

- **Outpatient Appointment**

- All patients will require assessment by a multidisciplinary team. The composition of the team may vary slightly. Patients with complex mullerian anomalies will need to see a gynaecologist, clinical nurse specialist and psychologist. Gynaecology input should include access to advanced minimal access surgery. Those with a DSD will also need specialist endocrinology and urology input. The service must be supported by specialised imaging, biochemisty and genetics. Family members may need genetic referral.

- At least two appointments will be required. The second appointment will include review of results and disclosure of information including atypical karyotype. Further psychology input is usually required for this in the majority of cases.

- **Investigations**

- As listed. Complex imaging should include magnetic resonance imaging **(**MRI). Genetics will include karyotype but also specialised genetic testing in the case of some Disorders of Sex Development (e.g. androgen insensitivity syndrome).

- **Treatment Strategy**

- Treatment is diagnosis dependent and may be non-surgical (dilation or hormone replacement) or surgical.

- Non-surgical dilation for neovaginal creation is the first line treatment of choice for vagina agenesis and is successful in 85% of cases. It is a nurse led treatment but requires two weekly contact with the patient (face-to-face or telephone) every 2-3 weeks for approximately 3-6 months. Women who fail dilation should be offered the laparosopic Vecchietti vaginoplasty.

- Surgical treatment will vary due to the diagnosis and previous surgical procedures. Procedures such as gonadectomy, excision of obstructed uterine horn, Davidov procedure, uterovaginal anastomosis and excision of some high transverse septums should be performed laparoscopically. Patients will previous vaginal or abdominal reconstructive surgery may require vaginoplasty with skin flaps or intestinal vaginal replacement. These procedures should be performed jointly with specialised urology

input. Plastic surgery input may be required.

- Induction of puberty will require regular follow-up initially frequently and then on an annual basis. Regular bone density monitoring will be required over the age of 18.

- **Follow-up**

- Follow-up for non-surgical dilation is described above.

- In addition some women will be required to perform vaginal dilation after surgical vaginoplasty. This will require follow-up visits for supervision with the nurse specialist.

- Other surgical procedures such as excision of an obstructed uterine horn will require a single post-operative visit only.

- Patients with a DSD will usually require long term surveillance for chronic disease. This will mean life-long specialist follow-up. Depending on the disease either an endocrinologist or gynaecologist will be the lead clinician.

- **Further investigations/treatment**

- As in pathway.

- **Second treatment episode**

- If dilation fails, a laparoscopic Vecchietti procedure is required.

- Vaginal stenosis is a recognised medium and long term risk of vaginoplasty and may require further treatment with dilation or surgery.

- **Pregnancy**

- Pregnant women with pre-existing conditions as discussed in this specification require assessment and/or management from highly specialist tertiary maternity care delivered within a dedicated multidisciplinary service staffed by a maternal medicine specialist, a physician, and supporting multidisciplinary team with extensive experience of managing the condition in pregnancy.

- In view of this, nationally commissioned condition specific services must have outreach arrangements with highly specialised tertiary maternity units with access to appropriate tertiary medical, surgical, fetal medicine, clinical genetics and level 3 Neonatal Intensive Care services. These specialised maternity services must have a critical mass of activity to maintain expertise, ensure best practice, training opportunities and for the organisational infrastructure, staffing, facilities and equipment to be

clinically and economically efficient. They should have robust risk management and performance monitoring processes.

- All such women must receive personalised pre-pregnancy and maternity care planning from specialised tertiary maternity services to allow optimal disease management in the context of the pregnancy. This will reduce avoidable morbidity, mortality and unnecessary intervention for mother and baby.

- Women with conditions discussed in this specification must be referred immediately once they are pregnant to plan their care. This must include access to termination of pregnancy and specialist advice re contraception. The individualised care plan must cover the ante natal, intrapartum and postnatal periods. It must include clear instructions for shared care with secondary services, when appropriate including escalation and transfer protocols and clear guidelines for planned and emergency delivery.

  - **Population covered**

- The service outlined in this specification is for patients ordinarily resident in England*; or otherwise the commissioning responsibility of the NHS in England (as defined in Who Pays?: Establishing the responsible commissioner and other Department of Health guidance relating to patients entitled to NHS care or exempt from charges). * - Note: for the purposes of commissioning health services, this EXCLUDES patients who, whilst resident in England, are registered with a GP Practice in Wales, but INCLUDES patients resident in Wales who are registered with a GP Practice in England.

- Specifically this service is for children, adolescents and adult women with congenital anomalies of the genital tract requiring specialised intervention and management as outlined within this specification.

- Currently 7 units in England offer this multidisciplinary service. Accurate incidence figures are not available but it is estimated this affects 1 in 2-3000 women.

  - **Any acceptance and exclusion criteria**

- **Referral Pathway Type Source**

- The service will accept referrals from primary or secondary care clinicians in adult or paediatric services. Patients will congenital anomalies will normally be under the care of a gynaecologist with appropriate specialist qualifications.

- The service will accept referrals from other providers particularly where the referring service is not accredited to undertake the clinical role the patient requires.

- There are two types of presentation:
- Presentation at birth or in childhood with a DSD (commonest presentation is with ambiguous genitalia) or complex urological anomaly. These patients will require transition from paediatrics for specialised adolescent and adult services.
- New in adolescence or early adult life e.g. primary amenorrhoea with or without pubertal development, virilisation at puberty, obstructed menstruation. These patients will present to their GP who will usually refer them to local gynaecologyservices.

- The Provider will be expected to use evidence based approaches and to demonstrate efficiencies whenever possible.

- Eligible patients will be referred using a defined referral system that can be audited for waiting times

- A discharge plan will be prepared offering support and facilities required providing care at home.

- Appropriate referrals to specialist colleagues will be documented and GP informed of any transfer of care.

- All paediatric, adolescent and adult patients with congenital anomalies of the genital tract will be referred to a specialist centre.

    - **Interdependencies with other services**

- The paediatric, adolescent and adult congenital anomaly service will be part of a multidisciplinary team working together, networking and linking with other healthcare services across both community and hospital settings. These services include, urology and colorectal services, genetics, imaging, psychology, plastic surgery and community.

- **Applicable Service Standards**

    - **Applicable national standards e.g. NICE, Royal College**

- **Care Excellence (NICE), Royal College STANDARDS OF CARE**

- All patients should be under the care of an experienced multidisciplinary team

- Expert psychological input targeting emotional and social adjustment is crucial.
- Patients are likely to require interventions to cope with an atypical karyotype, the emotional challenges of infertility, complications related to sexual function, orientation and identity, communication barriers within the family and partnership and problematic transitions.

- A supervised course of vaginal dilation should be the first line treatment for vaginal agenesis.

- Surgery should be performed by gynaecologists experienced in these procedures and where appropriate with input from other specialist surgical services such as urology, plastics and colorectal.

- Specific procedures should only be performed laparoscopically. These include gonadectomy, excision of uterine/mullerian remnant, uterovaginal anastomosis, Vecchietti procedure, Davidov procedure and some high transverse vaginal septums.

- Units should have close links with paediatric services and a specific focus on transition for adolescents.

- Specialised diagnostic support including genetics, biochemistry and imaging

- Fertility input may be required in some conditions for example in vitro fertilisation (IVF) and ovum donation in Premature Ovarian Failure (POF), Turner's and
- Swyer syndromes as well as surrogacy in mullerian agenesis.

- Centres managing patient with complex congenital anomies have a duty to:
- Audit surgical and non-surgical outcomes
- Build links with patient peer support groups
- Participate in clinical networks mapped by British Society for Paediatric and Adolescent Gynaecology
- Contribute to the body of knowledge on The Service by publishing original research as well as commentaries and reviews
- Contribute to the education of Specialist Registrars and other health professionals

- **References:**

- Hughes et al 2006 'Consensus statement on management of intersex disorders'
- *Arch.Dis.Child* ;**91**(7) 554-63.

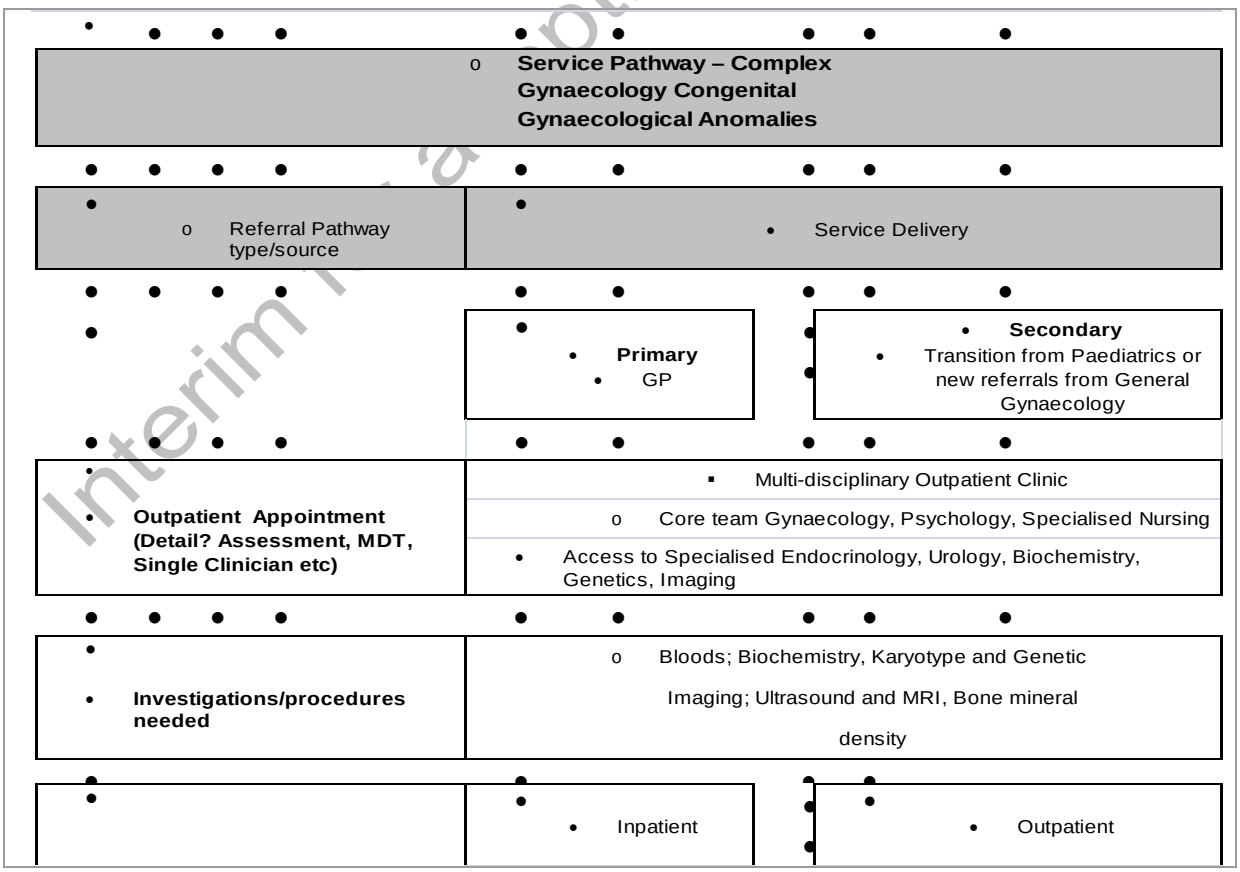
- Clinical Standards for Service Planning in Paediatric and Adolescent

Gynaecology. British Society for Paediatric and Adolescent Gynaecology. January 2011. Available from: www.britspag.org. Accessed November 2012.

- **Key Service Outcomes**

- **Planning**

- The Service will provide a tertiary service to support women requiring specialist support within a network of care and pathways.
- There will be an agreed planned and mapped pathway of care for women with a complex gynaecological problem
- The Service will be part of a multidisciplinary team working together, networking and linking with other healthcare services across both community and hospital settings.
- The Service will be expected to use evidence based approaches and todemonstrate efficiencies whenever possible.
- Eligible women will be referred using a defined referral system that can be audited for waiting times
- A discharge plan will be prepared offering support and facilities required providing care at home.
- Appropriate referrals to specialist colleagues will be documented and GP informed of any transfer of care
- It is the responsibility of the Provider to recruit/provide suitable and appropriately competent and qualified personnel in the provision of this service.
- Expert psychological input targeting emotional and social adjustment is crucial.
- Patients are likely to require interventions to cope with an atypical karyotype, the emotional challenges of infertility, complications related to sexual function, orientation and identity, communication barriers within the family and partnership and problematic transitions.
- Surgery should be performed by gynaecologists experienced in these procedures and where appropriate with input from other specialist surgical services such as urology, plastics and colorectal.
- Units should have close links with paediatric services and a specific focus on transition for adolescents.



Treatment Strategy (please provide detail)	Procedures include; Gonadectomy, Vaginoplasty (all types), Clitoral reduction, Excision of septum, Excision of obstructed o Psychology Vaginal Dilation Therapy Induction of puberty
Follow up (detail)	Post-surgical (dilation) 1, 6 and 12 weeks follow-up. Post-surgical (no dilation) 6 weeks follow up Dilation progamme (no surgery). 2 weekly nurse led follow up for up to 3 months. Psychology and psychosexual input as required. Post-gonadectomy - oestrogen replacement, 6 weeks
Further investigations/treatments required (detail)	o Post-gonadectomy - oestrogen replacement annual ▪ Psychology and pyschosexual. Fertility referral Chronic Disease Surveillance
Second treatment episode if needed (detail)	Complications such as vaginal stenosis require dilation+/- repeat vaginoplasty Failed primary vaginal dilation treatment (15%) process to lap Vecchietti ▪ Psychology and psychosexual

End.