SCHEDULE 2 – THE SERVICES

A. Service Specifications

Service Specification No:	E10-S-c (merged with former E10-s (HSS)-b)
Service	Complex Gynaecology: Congenital Gynaecological Anomalies (Children of 13 years and above and Adults)
Commissioner Lead	*For local completion*
Provider Lead	*For local completion*

1. Scope

1.1 **Prescribed Specialised Service**

This service specification covers the provision of specialised, multi-disciplinary team (MDT) gynaecological management, including surgery in some cases, for girls from 13 years of age and women with rare, congenital anomalies of the female genital tract. In a small number of individual cases, girls may be referred at a younger age depending on their clinical diagnosis and treatment requirements.

1.2 **Description**

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

Complex Congenital Gynaecological Anomalies include the following two diagnostic groups:

- Disorders of Sex Development (DSD). Sometimes referred to as intersex conditions. Old terminology now abandoned but still present in ICD classification includes Indeterminate sex and pseudohermaphroditism. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2082839/

- Complex Mullerian Anomalies. This includes Mayer-Rokitansky--Küster-Hauser (MRKH) syndrome (mullerian agenesis, MRKH) and mullerian obstruction/duplication. Some complex mullerian anomalies are associated with complex urological anomalies such as bladder exstrophy and cloacal anomaly.

1.3 **How the Service is Differentiated from Services Falling within the Responsibilities of Other Commissioners**

The service is accessible to all eligible patients with a congenital gynaecological anomaly. All commissioned centres must accept referrals for eligible patients meeting the acceptance and clinical criteria.

2. Care Pathway and Clinical Dependencies

2.1 **Care Pathway**

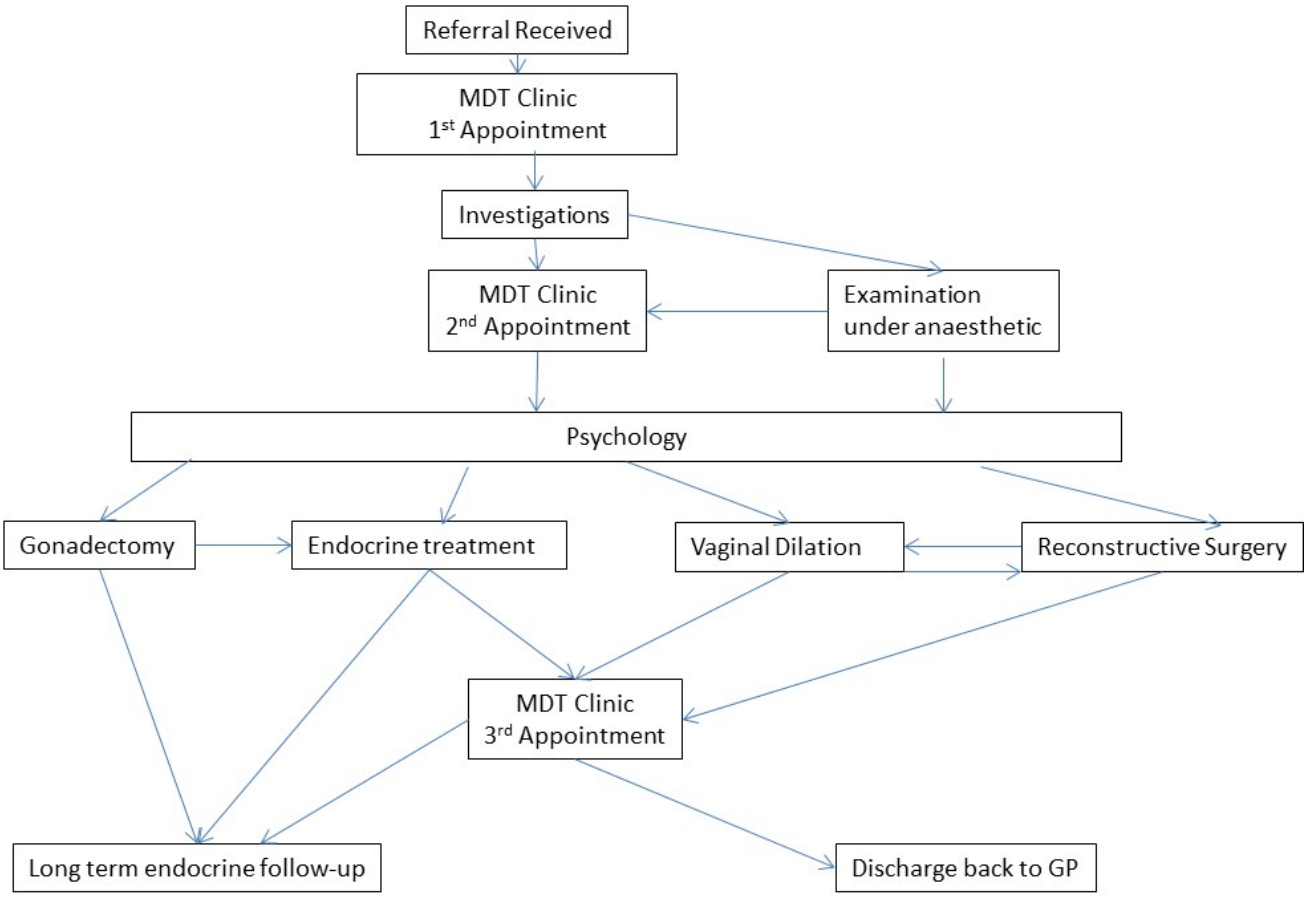
The service will ensure the provision of specialised gynaecological management including surgery in some cases, for girls from 13 years of age and women with rare, congenital anomalies of the genital tract. Some patients will need long-term specialised gynaecological management into adulthood. In a small number of individual cases, girls may be referred at a younger age depending on their clinical diagnosis and treatment requirements. The specialist gynaecologists working within the service will manage both adolescent and adult

patients, thereby ensuring a smooth transition to adult services. The service will refer to self-care guidelines for patients including engaging with peer support and with local or national support organisations should they wish to take this up.

The service will be provided by experienced, multi-disciplinary teams[1,2,] who will manage the needs of patients with complex conditions or associated co-morbidities such as congenital adrenal hyperplasia or who require treatment including reconstructive surgery where indicated to optimise future gynaecological, psychological and reproductive outcomes. Specialist centres will have defined links to other services as outlined in section 2.0; deliver high quality services within an agreed network of providers whose population may also use this service, and establish close links with other services providing care for children with these complex conditions.

Following referral, the patient journey will include: initial outpatient assessment; investigations and diagnosis; day case, in-patient or outpatient treatment, including surgery where appropriate; outpatient follow-up; discharge where appropriate; long-term management where necessary. Adolescent patients will be seen in a dedicated paediatric and adolescent gynaecology clinic located within paediatric services and in age-appropriate surroundings. If in-patient admission for surgery is required, this will be to a paediatric ward. All clinicians involved in the care of children and adolescents should have training in safeguarding to level 3. The multi-disciplinary team will be co-located and include the following staff: a gynaecologist trained in paediatric and adolescent gynaecology (PAG); a second gynaecologist with PAG experience and skills in advanced minimal access surgery; psychologist; specialist nurse; endocrinologist; urologist; radiologist. Paediatric services should usually be co-located but in some areas may be in close proximity depending on hospital configuration.

Patient pathway



As a result of interaction with the service, patients will have a clear understanding of the proposed difference that treatment will have on their condition, including urinary or sexual function as appropriate and have an increased quality of life as a result of improved urinary or sexual function.

Interdependent services: colorectal; biochemistry; genetics including karyotype and specialised genetic testing; plastic surgery; paediatric endocrinology; paediatric gynaecology, paediatric urology/surgery; paediatric psychology; specialised imaging including magnetic resonance imaging (MRI); other: family liaison, social workers; Child and Adolescent Mental Health Services (CAMHS), mental health services.
To Note: access to treatment will be guided by any applicable NHS England national clinical commissioning policies.

Outpatient Appointments: All patients will be assessed by a multidisciplinary team. The composition of the team may vary slightly depending on need, for example, 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, whereas those patients with a DSD will also need specialist endocrinology and urology input. Family members may need a referral for genetic diagnosis and if so, the service will arrange for this. The service will provide appropriate literature on the patient's condition, provide information on support groups and where indicated, request an onward referral for psycho-social support via the patient's GP. Following referral, at least two appointments will take place to determine the diagnosis. The second appointment will include: the review of results and disclosure of information including atypical karyotype; further psychology input; if surgery is appropriate, it will be planned at this visit. Age appropriate informed consent will be provided for all patients and psychological support will be made available to the patient and family to facilitate this. Psychological support and counselling will be provided to the patient prior to and following genetic testing. Genetic counselling for family members will require referral to genetic services.

Investigations: The service will include access to on-site specialised imaging including magnetic resonance imaging (MRI). Access to biochemistry and genetic testing will also be required including karyotype and specialised genetic testing, although these may be located off-site.

Treatment Strategy: Treatment is diagnosis-dependent and may be non-surgical (dilation or hormone replacement) or surgical. Psychological support will be available for all patients regardless of the mode of treatment. Non-surgical dilation for neovaginal creation is the first line treatment of choice for vaginal agenesis and is successful in over 80% of cases. The service must be flexible and offer patient choice as to the timing of dilation. Timing of treatment is an individual choice and adolescents may choose to commence treatment immediately or defer until adulthood. It will include an initial outpatient based nurse-led treatment session, with the patient then practicing the technique at home, with discussion with the patient (face-to-face or telephone) every two to three weeks for approximately three to six months. Psychological support is valuable both in preparation for dilation and support for those struggling with compliance. In-patient admission to hospital for dilation has not been demonstrated to give better results and most units will offer outpatient treatment only. Patients who fail dilation or are unsuitable for dilation due to scarring or genital configuration, will require surgical vaginal reconstruction. Options including advantages and disadvantages of each will be fully discussed so that the patient can make an informed decision and give consent. In conjunction with the patient, the type of operation chosen will depend on the patient's past surgical history. In the absence of previous genital surgery, the laparoscopic Vecchietti procedure has been demonstrated to have a high success rate and low morbidity. Units where minimal access surgery is not available will refer the patient to a specialist unit that can offer this.

In cases where the patient has genital scarring or unsuitable anatomy, a variety of other surgical reconstructive procedures are used. These include vaginal reconstruction with skin flaps, skin grafts and intestine as well as the laparoscopic Davidov procedure. These procedures are usually performed as joint reconstructive cases with input from other surgical specialties such as urology, colorectal or plastics. The rarest forms of surgery such as intestinal vaginoplasty, which involves colo-rectal surgery and some of the very rare mullerian anomalies (e.g. cervical agenesis) which require advanced techniques of laparoscopic surgery will only be provided by one or two centres in the service - the locations for which need to be agreed with the commissioners. Other units will refer into these expert centres.

Surgical treatment for other complex anomalies will vary due to the diagnosis and the previous surgical procedures that a patient has had. Individual needs and preferences will be taken into account when planning surgery. Minimal access surgery carries significant advantages in this young group of patients in terms of physical appearance and the speed of recovery, meaning less time missed form school/college. Providers of minimal access surgery will have up to date laparoscopic equipment. Procedures such as gonadectomy, excision of obstructed uterine horn, uterovaginal anastomosis and excision of some high transverse septums should be performed laparoscopically. Patients with 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 or colorectal input. Plastic surgery input may be required. Clitoromegaly is a

feature of some DSD diagnoses and can cause distress regarding appearance, identity and in sexual relationships. Initial assessment and management is psychological, including provision of psychological support to reduce distress. If surgery is required, it should be performed by surgeons proficient in this procedure. Informed consent should include discussion of potential detrimental impact on sexual sensitivity and sexual function. The service will provide psychological support both before and after surgery to maximise the psychological impact of the surgical outcome.

Some adolescents will require induction of puberty and this should be led by an endocrinologist or an appropriately trained paediatric gynaecologist. This entails regular follow-up initially frequently and then on an annual basis. Regular bone density monitoring will be required over the age of 18 years.
Investigation of delayed puberty should be done jointly with an endocrinologist. Management of patients with complex endocrinology conditions such as Congenital Adrenal Hyperplasia should be led by an endocrinologist within the setting of the multidisciplinary team. Fertility advice and onward referral to the patient's own General Practitioner may be required in some conditions, for example where the patient wishes to receive In-Vitro Fertilisation and this is indicated. Antenatal diagnosis and advice should be provided and appropriate care planning undertaken. Where endocrine input is required, the transition years (from 13 to 25 years) will involve both a paediatric and adult endocrinologist. The service will develop/make available suitable educational materials on treatment options and promote compliance.

Follow-up: Follow-up for non-surgical dilation is described above. In addition, some patients will be required to perform vaginal dilation after surgical vaginoplasty to prevent restenosis. 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 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.

Second treatment episode: If dilation fails, surgical vaginal reconstruction will be required. There are no comparative studies of the many available operative procedures. The laparoscopic Vecchietti procedure is offered in several UK units. It is widely used in Europe and research has confirmed low morbidity and high success rates. Other operations include vaginoplasty with skin graft (McIndoe-Reed procedure), rotational skin flaps and intestinal replacement. These procedures have a higher associated morbidity but are used when the Vecchietti procedure is contraindicated. Only clinicians who are trained in the specialist procedures will provide minimally invasive surgical care, whilst those who are not able to undertake these procedures will refer to appropriate specialists in designated centres. Vaginal stenosis is a recognised medium and long-term risk of vaginoplasty and may require further treatment with dilation or surgery.

Pregnancy: Some pregnant women with a pre-existing condition, such as Congenital Adrenal Hyperplasia or complex urological anomalies, will require assessment and/or management from specialised maternity team within a dedicated multidisciplinary service which is staffed by a maternal medicine specialist, a physician and supporting multidisciplinary team with experience of managing the condition in pregnancy. In view of this, the service will have outreach arrangements in place with specialised tertiary maternity units* to include access to appropriate tertiary medical, surgical, fetal medicine, clinical genetics and level 3 neonatal intensive care services.

* The specialised maternity services will have a critical mass of activity to maintain expertise, ensure best practice, offer training opportunities and have the organisational infrastructure, staffing, facilities and equipment to be clinically and economically efficient. They should also have robust risk management and performance monitoring processes.

All such patients must receive personalised pre-pregnancy and maternity planning from specialised maternity services to allow optimal disease management in the context of pregnancy. Women with conditions discussed in this specification must be referred immediately once they are pregnant to enable their care to be planned and an antenatal diagnosis made. This must include access to termination of pregnancy and specialist advice regarding contraception. The individualised care plan must cover the antenatal care, diagnosis and planning as well as the intra-partum and post-natal periods. The care plan must include clear instructions for shared care with secondary services and, when appropriate include escalation and transfer protocols and clear guidelines for planned and emergency delivery.

Information will be given to the patient and to their primary care clinicians about the implications of their condition for contraception, pregnancy (including miscarriage and termination) fertility, sexual health/function and menopause.

Service model

The majority of cases present in one of two ways:

I) either at birth, in which case patients will be managed by their local specialist paediatric team until adolescence when they will then be referred into the service. The timing of the transfer will depend on the patient's maturity and medical requirements and the configuration of local services.

ii) a new diagnosis in adolescence 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 gynaecology services who will then refer directly to an adolescent gynaecologist in the service, without the need to see a paediatrician first.

The service will include a number of specialist centres which provide tertiary specialist service to support patients within a network of care which spans the pathway of care. Where a referral is for an adolescent who is transferring to the service from local specialist paediatric teams, the service will ensure the smooth transition of care by having established close links with those paediatric services which provide care for children with complex gynaecological disorders. Ideally the service will be located in centres which also provide specialist paediatric gynaecology services. Older adolescents who are patients of the service may prefer to be on an adult ward and should be offered the choice. There will be an agreed planned and mapped pathway of care for patients with a complex gynaecological condition. Gynaecologists will manage both adolescent and adult women with these conditions. As the surgical procedures are identical regardless of age, this will ensure adequate surgical activity to maintain skills. Patients are likely to require interventions to cope with an atypical karyotype and the potential emotional challenges of infertility, complications related to sexual function, orientation and identity, communication barriers within the family, friends, partner and problematic transitions. Surgery will be performed by specialist gynaecologists experienced in these procedures and where appropriate, with input from other specialist surgical services such as urology, plastics and colorectal. Depending upon the range of surgical procedures available within the centre, onward referral may need to be made to another specialist centre which has additional surgical expertise.

Referral processes and sources: The service will accept referrals from primary or secondary care clinicians in adult or paediatric services and specialist paediatric services. Patients with congenital anomalies will 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.

Transition: Specific attention will be paid to the needs of adolescent patients during their transition to adult services. Once discharged from the service, patients with ongoing psychological or psychosexual concerns will require GP referral for local mental health services.

Discharge criteria and planning: A discharge plan will be prepared with the patient offering support and facilities required for providing care at home. Post-surgical vaginal dilation supervision will be offered if required.

2.2 **Interdependence with other Services**

i) Co-located services: The multi-disciplinary team will include: paediatric gynaecology; gynaecologist (including advanced minimal access surgery); psychologist; specialist nurse; endocrinologist; urologist; radiologist. Paediatric services will usually be co-located but in some cases may be in close proximity depending on the hospital configuration.

ii) Interdependent services: Colorectal; biochemist; genetics including karyotype and specialised genetic testing; plastic surgery; paediatric endocrinology; paediatric urology/surgery; paediatric psychology; imaging including magnetic resonance imaging (MRI); other: family liaison, social workers; Child and Adolescent Mental Health Services(CAMHS), mental health services.

Related Services: primary care; local Mental Health Services (Adolescent and Adult).

Close links will be established with the active patient peer support groups which operate in this service area. Literature from support groups will be available in clinic and offered to all patients. Support groups

will also be engaged in service planning and will be invited to provide feedback on services. The major support groups include; Androgen Insensitivity Syndrome Support Group (www.aissg.org), Congenital Adrenal Hyperplasia Support Group (www.livingwithcah.com), dsdFamilies (www.dsdfamilies.org), Turner Syndrome Support Group (www.tss.org.uk). Other smaller groups exist and have an important contribution to make.

3. Population Covered and Population Needs

3.1 Population Covered By This Specification

This specification covers for girls from 13 years of age and women with rare, congenital anomalies of the female genital tract. This service specification covers the population defined as the commissioning responsibility of NHS England. Commissioning arrangements for the devolved nations in relation to this service are as set out in "UK-wide Commissioning Arrangements of Highly Specialised Services" [web link is https://www.england.nhs.uk/publication/nhs-providers-of-highly-specialised-services/].

3.2 Population Needs

Accurate incidence figures are not available but these conditions are estimated to affect 1 in 2-3000 females. A review of the incidence of the most commonly seen conditions gives an indication of the expected numbers This is in the order of 25,000 cases in the current population of UK women. See Appendix 1 for references.

3.3 Expected Significant Future Demographic Changes

No growth expected in total numbers of patients apart from that due to improved coding and identification of patients.

3.4 Evidence Base

The specification is based on the following clinical evidence:

1. Consensus statement on management of intersex disorders, Hughes et al Arch.Dis.Child July 2006 91(7) 554-63.
2. Clinical Standards for Service Planning in Paediatric and Adolescent Gynaecology. British Society for Paediatric and Adolescent Gynaecology. January 2011 www.britspag.org
3. Congenital Adrenal Hyperplasia in the Adult. British Society for Endocrinology Guidelines 2013.
4. American College of Obstetricians and Gynaecologists. Mullerian agenesis: diagnosis, management and treatment. Committee opinion Number 562 May 2013.
5. British Society for Paediatric and Adolescent Gynaecology National Safety Standards for Invasive Procedures (NatSSIPs) NHS England, September 2015 https://www.england.nhs.uk/wp-content/uploads/2015/09/natssips-safety-standards.pdf
6. National Safety Standards for Invasive Procedures (NatSSIPs) September 2015 www.england.nhs.yk/wp-content/uploads/2015/09/natssips-safety-standards.pdf

4. Outcomes and Applicable Quality Standards

4.1 Quality Statement – Aim of Service

The aims of the service are to provide patient centred specialist assessment and management, including surgery where appropriate, for adolescent and adult women with a congenital abnormality of their genital tract; to promote family centred specialist care and to optimise future reproductive and sexual function, psychological health and quality of life. As a result, patients will feel comfortable interacting with the service and empowered to contribute to and make decisions about, their own healthcare.

NHS Outcomes Framework Domains

Domain 1	**Preventing people from dying prematurely**	
Domain 2	**Enhancing quality of life for people with long-term conditions**	√
Domain 3	**Helping people to recover from episodes of ill-health or following injury**	√
Domain 4	**Ensuring people have a positive experience of care**	√
Domain 5	**Treating and caring for people in safe environment and protecting them from avoidable harm**	√

4.2 Indicators: Please note that the interpretation of these indicators will be subject to the specific treatments received by patients depending on their individual circumstances

Number	Indicator	Data Source	Outcome Framework Domain	CQC Key question
Clinical Outcomes				
101	number of new referrals	SSQD	2,3	effective
102	number of follow up appointments	SSQD	2,3	effective
103	number of patients referred for vaginal dilatation	SSQD	2,3	effective
104	number of open surgical procedures	SSQD	2,3	effective
105	number of laparoscopic procedures	SSQD	2,3	effective
106	% patients receiving gonadectomy	SSQD	2,3	effective
107	% XY patients presenting with gonadal cancer	SSQD	2,3	effective
108	% patients stabilised on HRT	SSQD	2,3	effective
109	% patients sexually active following vaginal dilation	SSQD	2,3	effective
110	% patients sexually active following vaginal reconstruction	SSQD	2,3	effective
111	% patients seen by psychologist	SSQD	2,3,4	effective
112	% patients with vaginal agenesis who declined vaginal dilatation	SSQD	2,3	effective

113	% patients with vaginal agenesis treated with vaginal dilatation that was unsuccessful	SSQD	2,3	effective
114	% patients treated laparoscopically	SSQD	2,3,4	effective
115	number of patients receiving surgery outside the centre prior to referral	SSQD	2,3,5	effective, safe
116	Length of stay for patients receiving laparoscopic surgery	SSQD	2,3,	effective
117	Length of stay for patients receiving open surgery	SSQD	2,3,	effective
118	% patient with post-operative complications following laparoscopic surgery	SSQD	2,3,5	effective, safe
119	% patient with post-operative complications following open surgery	SSQD	2,3,5	effective, safe
Patient Experience				
201	Patient information	Self declaration	4	Responsive caring
202	Patients have a named clinical nurse specialist	Self declaration	4	responsive, caring
203	Adolescent patients have a named transition facilitator	Self declaration	4	responsive, caring
204	Patient feedback	Self declaration	4	responsive, caring
Structure and Process				
301	There is a specialist team	self declaration	2,3,5	well led, safe ,
302	Patients are discussed at an MDT meeting	Self declaration	235	effective
303	There is a multidisciplinary clinic for the assessment of patients	Self declaration	2,4	effective, responsive &caring
304	There is a dedicated paediatric and adolescent gynaecology clinic	Self declaration	2,3,4	safe, effective
305	There is a nurse led clinic for vaginal dilatation	Self declaration	2,3,4	safe, effective
306	There is easy access to paediatric facilities	Self declaration	3,4,5	responsive, caring
307	There are agreed patient pathways	Self declaration	3,4,5	effective
308	There are agreed pathways for transition	Self declaration	3,4	effective
309	There are agreed clinical protocols	Self declaration	2,3,5	safe, effective
310	There are annual audit meetings	Self declaration	2,3,5	effectiveness responsive

Detailed definitions of indicators, setting out how they will be measured, is included in the NHS England contract documentation, Schedule 6.

4.3 Commissioned providers are required to participate in annual quality assurance and collect and submit data to support the assessment of compliance with the service specification as set out in Schedule 4A-C

There is a requirement to hold national audit meetings involving all specialist centres meeting together on an annual basis. Audit meetings should address: clinical performance and outcomes including surgical and non-surgical outcomes; process-related indicators, such as efficiency of the assessment process, prescribing policy, bed provision and occupancy, outpatient follow-up; stakeholder satisfaction, including feedback.

4.4 Applicable CQUIN goals are set out in Schedule 4D

5. Applicable Service Standards

5.1 Applicable Obligatory National Standards

Providers must comply with the following:

- Management of women with premature ovarian insufficiency - Guideline of the European Society of Human Reproduction and Embryology, December 2015.
- Society for Endocrinology UK Guidance on the initial evaluation of an infant or an adolescent with a suspected Disorder of Sex Development (revised 2015) Clinical Endocrinology (Oxf) 2015 Aug 13. doi: 10.1111/cen.12857.
- BritSPAG Clinical Standards for Services Users in PAG 2011 (some parts currently being updated).

5.2 Other Applicable National Standards to be met by Commissioned Providers

Providers must comply with the standards of the British Society for Paediatric and Adolescent Gynaecologists which is a specialist group under the umbrella of the Royal College of Obstetricians and Gynaecologists. Providers should also: collaborate in establishing a national DSD clinical network for centres managing DSD. Service providers will be mindful of cultural sensitivities around potential fertility and other impacts of the condition and take into account each patient's cultural and religious backgrounds and ethnicity.

5.3 Other Applicable Local Standards

The service's specialist centres must ensure they are fully integrated into their Trust's corporate and clinical governance arrangements and comply fully with clinical negligence scheme for trusts (CNST) and Care Quality Commission (CQC) requirements in terms of quality and governance.

6. Designated Providers (if applicable)

The specialist centres already exist and have a contract to provide this service specification. NHS England Hubs will assess compliance to the revised specification.

7. Abbreviation and Acronyms Explained

The following abbreviations and acronyms have been used in this document:

BritSPAG	British Society for Paediatric and Adolescent Gynaecology
DSD	Disorders of Sexual Development
ICD classification	International Classification of Diseases
IVF	In Vitro Fertilisation
MRKH syndrome	Mayer-Rokitansky--Küster-Hauser (MRKH) syndrome
SSQD	Specialised Services Quality Dashboard
VTE	VTE (venous thromboembolism)

Appendix 1: Incidence of conditions

The table below gives incidences of some of the conditions covered by this service specification. The current female population of England is approximately 25 Million. Just taking the above conditions gives a minimum of 0.5 to 1 per 1,000 population i.e potentially 25,000 cases.

Condition	Incidence	Reference
Mayer-Rokitansky-Kuster-Hauser Syndrome	I in 5000	Attomaki et al Fert.Steril. 2001
Congenital Adrenal Hyperplasia	1 in 30,000 female	Pang SY et al. Paediatrics 1988
Complete Androgen Insensitivity Syndrome	1 in 40,000	Boehmer AL et al. J.Endocrin.Metab 2001
Turner Syndrome	1 in 3,000	Gravholt, S. Juul, et al BMJ. 1996
Complex Mullerian Anomalies (transverse septae, obstructed hemivagina, etc.)	Approx. 1 in 5,000	No recent medical literature