SITM: Prenatal Diagnosis (PD)

SECTION 1: CAPABILITIES IN PRACTICE

PD CiP 1: The doctor can use ultrasound to recognise where fetal anatomy is not normal.	
Key Skills	**Descriptors**
Demonstrates normal structural findings in all trimesters and recognises if normality cannot be demonstrated	• Performs and records a detailed, systematic ultrasound of the fetus as per FASP guidance. • Understands the strengths and limitations of ultrasound for each system within each trimester. • Explains normal anatomical views to the woman. • Documents and records normal anatomical views. • Recognises when image quality is technically poor. • Is able to explain next steps if normal views cannot be obtained.

Evidence to inform decision	
• Reflective Practice • NOTSS • TO2 • CbD • Mini-CEX • OSATs (see below)	• RCOG Learning • FASP on-line training • Local and Deanery teaching • Attendance at relevant courses and conferences • Log of cases and outcomes • Attendance at fetal medicine clinics • Attendance at MDTs • Attendance at specialist neonatal and paediatric clinics • Examples of anonymised birth plans

Knowledge criteria

- The normal appearances on ultrasound scan in all trimesters of the fetal CNS, face and neck, thorax, cardiovascular system, abdominal wall and gastrointestinal tract, urogenital system and the fetal skeleton and extremities
- Local protocols for follow up, if any, after an incomplete anatomy scan
- Normal embryology of all body systems, and how errors in these processes result in the more common fetal abnormalities targeted by FASP.
- Normal fetal behaviour and activity, and abnormalities of this
- Fetal circulation, and how it adapts at birth
- Diagnostic features of each condition targeted by FASP, their differential diagnosis and chance of structural, chromosomal and syndromic associations. These conditions are Trisomy 21, 18 and 13, anencephaly, spina bifida, congenital diaphragmatic hernia, gastroschisis, exomphalos, renal agenesis, facial cleft, hypoplastic right or left heart, lethal skeletal dysplasia

- The thresholds for diagnosing mild, moderate and severe ventriculomegaly measurements, and the potential implications of the different severities of ventriculomegaly
- The role of MRI for CNS lesions.
- The difference between Dandy Walker malformation, DW Variant and Mega cisterna magna, the implications of each and the pitfalls in prenatal diagnosis
- The common fetal tachy- and brady – arrhythmias and the role of the paediatric cardiologist in their management
- The different types of VSD and their association with cardiac, extracardiac and chromosomal anomalies. Understand the role of the paediatric cardiologist in their management
- The ultrasound features of transposition of the great arteries, atresia of either outflow tract, stenosis of either outflow tract, double outlet right ventricle or a common outflow tract (truncus arteriosus)
- The association of these conditions with further cardiac, extracardiac and chromosomal anomalies
- The role of the paediatric cardiologist in the management of fetal cardiac problems
- The ultrasound features of GI atresia, associations and surgical options following birth
- The spectrum of ultrasound findings of echogenic bowel and its association with chromosomal anomalies, cystic fibrosis, growth restriction and viral infections
- Urinary tract obstruction and MCDK: aetiology, spectrum of severity postnatal investigation and the likely short- and long-term impact of these conditions
- The local pathway for postnatal referral for talipes and the Ponsetti approach to treatment
- Limb reduction defects: associations and aetiology
- Findings suggestive of lethal skeletal dysplasia and the features of the more common non–lethal dysplasias, particularly certain types of osteogenesis imperfecta and achondroplasia
- A differential diagnosis for non-immune hydrops, the need for tertiary referral and the range of investigations likely to be offered

PD CiP 2: The doctor can assess and investigate a pregnancy where there are concerns regarding the fetus	
Key Skills	**Descriptors**
Is able to provide genetic counselling in common prenatal situations	• Takes an appropriate history and constructs, where appropriate, a family tree in women with, or chance of, genetic conditions. • Explains common modes of Mendelian and multifactorial inheritance, and recurrence risks. • Counsels for previous trisomy and monosomy X. • Counsels for previous neural tube defect.
Provides initial counselling with regard to common fetal structural abnormalities and ongoing management in	• Experienced in the ultrasound diagnosis and management of pregnancies complicated by fetal abnormalities covered by the Fetal Anomaly Screening Programme. • Discusses other potential prenatal tests appropriately.

conjunction with tertiary FM services	• Recognises when to refer to tertiary centre and how best to share care and monitoring. • Liaises appropriately with the tertiary centre and the multidisciplinary team. • In collaboration with subspecialists, formulates, implements and where appropriate modifies management plan. • Counsels individuals and their partners regarding the fetal risks, implications for the pregnancy and the long-term outcome. • Signposts to external sources of information and support. • Constructs a follow-up plan for the pregnancy. • Plans birth and appropriate neonatal support in collaboration with fetal medicine specialist.
Counsels and manages pregnancies at risk of fetal infection	• Investigates appropriately for common fetal infections. • Is able to interpret laboratory results for each infection in liaison with virology. • Explains the potential fetal, newborn and long-term effects of fetal infections. • Recognises when to refer and how best to share care and monitoring. • Liaises appropriately with the tertiary centre and the multidisciplinary team.
Counsels and manages severe early fetal growth restriction	• Is able to produce a differential diagnosis. • Knows when and which further investigations should be offered. • Liaises with the fetal medicine tertiary referral centre regarding diagnosis and ongoing management.
Counsels regarding prenatal investigations	• Understands both the non-invasive and invasive options and is able to discuss the risks and benefits, facilitating choice. • Understands the importance of the different levels of resolution of genetic testing and is able to communicate the importance of this to the parents. • Explains the risks and benefits of each procedure and any alternatives. • Communicates the scope and the limitations of these tests. • Describes how prenatal samples are processed and when, and how, the results are given. • Offers genetic counselling where appropriate.
Evidence to inform decision	
• Reflective Practice	• RCOG Learning

• NOTSS • TO2 • CbD • Mini-CEX	• Local and Deanery teaching • Attendance at relevant courses and conferences • Attendance at Clinical Genetics clinics Log of cases and outcomes • Attendance at fetal medicine clinics • Attendance at MDTs • Attendance at specialist neonatal and paediatric clinics • Examples of anonymised birth plans

Knowledge criteria

- The genetic basis for trisomy 21, 18 and 13 and the ultrasound features associated with them
- The range of tests available for screening and testing for the common trisomies and the organisation and quality control of the screening service
- Other aneuploidies: the implications of Turner syndrome (45XO), Kleinfelter syndrome (47 XXY) and 47 XXX and appreciate the approach to managing pregnancies complicated by much rarer/unique chromosomal abnormalities
- The underlying genetic inheritance patterns and prenatal testing for cystic fibrosis, muscular dystrophy and fragile X, and the need for liaison with clinical genetics
- When it is appropriate to offer invasive testing, and when not to
- The role of non-invasive testing
- The implications for the current pregnancy and the long-term prognosis for each condition, and recurrence risks for future pregnancies
- The limitations of ultrasound in detecting and diagnosing congenital abnormalities (e.g. cleft palate) or predicting prognosis (e.g. diaphragmatic hernia)
- Triggers and diagnoses necessitating tertiary referral
- Diagnostic features of each condition, their differential diagnosis and the chance of associated structural, chromosomal and syndromic associations
- The role of DNA analysis from maternal plasma

PD CiP 3: The doctor demonstrates the skills and attributes required to provide ongoing support and care to parents for whom a problem with their pregnancy has been identified	
Key Skills	**Descriptors**
Counsels on and organises or refers onwards for termination of pregnancy for fetal anomaly	• Raises the option of termination of pregnancy for fetal anomaly appropriately. • Counsels regarding the different methods of termination, when termination is offered and when fetocide is legally mandated. • Organises termination of pregnancy for fetal anomaly (or refers appropriately where there is conscientious objection or the need for tertiary involvement). • Supports the parents' journey from diagnosis to follow up with planning for future pregnancies.

	• Adjusts care around termination of pregnancy in high risk situations. • Manages complications of termination of pregnancy. • Is aware of and can signpost to appropriate organisations providing support.
Supports the woman who wishes to continue with the pregnancy where the baby will not survive to birth and the pregnancy where the baby is expected to die in the neonatal period	• Supports and empowers the parents in their decision. • Plans for delivery with the family and paediatric team to give the parents the best experience possible in the circumstances, with clarity on intervention/non-intervention in labour. • Plans an appropriate end of life pathway with the family and paediatric team.
Provides follow up and counselling after a pregnancy complicated by fetal anomaly	• Explains the role of the post-mortem and any other relevant postbirth tests (eg genetic testing, post-mortem MRI). • Explains the findings and implications of any additional post-birth investigations. • Refers, where appropriate, to the wider multi-disciplinary team, including clinical genetics. • Counsels regarding chance of recurrence across the range of conditions targeted by FASP, and arranges genetic counselling where appropriate. • Proposes a plan for future pregnancy management. • Recognises when tertiary service involvement is appropriate for more complex cases.

Evidence to inform decision	
• Reflective Practice • NOTSS • TO2 • CbD • Mini-CEX	• RCOG Learning • Local and Deanery teaching • Attendance at relevant courses and conferences • Attendance at neonatal unit ward rounds • Log of cases and outcomes • Attendance at fetal medicine clinics • Attendance at MDTs • Attendance at specialist neonatal and paediatric clinics • Examples of anonymised birth plans

Knowledge criteria

- The antenatal management, intrapartum care and immediate postnatal management of each condition
- The impact of the diagnosis and individual circumstances on the timing, location and mode of birth
- The local prenatal, birth and post-birth pathways for care of the fetus and newborn with these conditions

- The legal framework under which termination of pregnancy by feticide may be offered
- Recognise which conditions are amenable to prenatal treatment (e.g. diaphragmatic hernia, spina bifida)
- The recurrence risk and management plan for future pregnancies for each condition

SECTION 2: PROCEDURES

The trainee will provide evidence through OSATs of their competency to perform fetal anomaly scans i.e. they may choose to have an OSAT demonstrating their assessment of a single system, but should be able to demonstrate all the fetal systems to the standard of FASP. Procedures marked with * require three summative competent OSATS

Procedures	*Level by end of training*	*CIP 1*	*CIP 2*	*CIP 3*
Fetal anomaly scan*	4	X		
Fetal echo*	4	X		
Amniocentesis	1		X	
CVS	1		X	
Fetocide	1			X

SECTION 3: GMC GENERIC PROFESSIONAL CAPABILITIES

Mapping to GPCs
Domain 1: Professional values and behaviours Domain 2: Professional skills • Practical skills • Communication and interpersonal skills • Dealing with complexity and uncertainty • Clinical skills *(history taking, diagnosis and management, consent; humane interventions; prescribing medicines safely; using medical devices safely; infection control and communicable diseases)* Domain 3: Professional knowledge • Professional requirements • National legislative requirements • The health service and healthcare systems in the four countries Domain 4: Capabilities in health promotion and illness prevention Domain 5: Capabilities in leadership and teamworking Domain 6: Capabilities in patient safety and quality improvement • Patient safety • Quality improvement Domain 7: Capabilities in safeguarding vulnerable groups

SECTION 4: MAPPING OF ASSESSMENTS TO PD CiPs

PD CIP	OSATS	Mini-CEX	CbD	NOTSS	TO1/ TO2	Reflective practice
1: The doctor can use ultrasound to recognise where fetal anatomy is not normal	X	X	X	X	X	X
2: The doctor can use ultrasound to recognise where fetal anatomy is not normal		X	X	X	X	X
3: The doctor demonstrates the skills and attributes required to provide ongoing support and care to parents for whom a problem with their pregnancy has been identified		X	X	X	X	X