



Scottish Differences of Sex Development (SDSD) Neonatal Care Pathway

Release: V1.0

Date: March 2017

Updated: March 2021

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Document control

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Title:	Scottish Differences of Sex Development (SDSD) Neonatal Journey for initial assessment of Uncertain Gender at Birth
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Owner(s):	SDSD Managed Clinical Network

Version history

Version	Date of revision	Summary of changes
0.1		First draft prepared by Katie Clayton
0.2	31 July 2016	Revisions following Steering Group
0.3	5 October 2016	
0.4	7 December 2016	Shared with SPEG / Regional Neonatal Networks and changes incorporated
V2	July 2020	
V3	March 2021	

Distribution

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SDSD Steering Group	Various

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1. Introduction

This DSD Neonatal Care Pathway is designed to ensure all babies born with a difference in sex development (DSD) in Scotland receive care that is safe, effective and centred around the needs of the baby and family.

It is specific to the first few days of life, providing clinical staff who may not have encountered DSDs before, with the information they need to ensure that babies are managed and referred appropriately and their parents are supported throughout. Each Health Board will have a detailed clinical referral pathway for management of newborns with suspected DSD which should be followed.

This Pathway and supporting guidance have been developed by SDSD for use within NHS Scotland. The Pathway can be found in Appendix 1. A detailed clinical pathway for DSD specialists can be found at (1)

The following principles underpin the Pathway:

Where DSD is suspected:

- 1.1 Care should be safe, effective and centred around the needs of the baby and family.
- 1.2 The Pathway is initiated when one or more of five referral criteria are evident (see 2.2)
- 1.3 Meeting immediate clinical needs takes priority over assigning sex to the baby. Parents will be encouraged to provide all appropriate personal care to their baby by the local neonatal team.
- 1.4 The parents of the baby will have their concerns listened to, be given reassurance, and offered psychological support, this includes signposting to relevant support materials and resources
- 1.5 Health care professionals encountering DSD for the first time will have access to key messages about DSD and use them to help reassure and support parents.
- 1.6 The local neonatal team will contact the local DSD Link Clinician according to the local neonatal guidance document for the health board. The local DSD Link Clinician will organise further tests and refer to relevant regional DSD specialist if required.
- 1.7 Local DSD Link Clinician will keep parents informed and agree a management plan with the family for ongoing care needs.

2. DSD

The term 'Differences of Sex Development' covers a wide range of conditions. Some, such as undescended testes and hypospadias are common but others are rare and will require specialist DSD input. DSDs occur at an approximate rate of 2-3 per 1000 live births.

2.1. Assignment of Sex

For the majority of children, sex is assigned at birth. However, if the child is born with atypical genitalia, sex assignment at birth may not be possible. When a baby is born with a suspected DSD, referral to specialist care and investigations to establish the sex of rearing may be required.

Concerns may be raised regarding atypical genitalia antenatally and should be discussed with fetal medicine specialists. Where possible families should meet with a Neonatologist or Paediatric Endocrinologist with DSD expertise and information leaflets should be given if appropriate.

2.2. Criteria for Referral to DSD Specialist care

Any of the following are indications for initiating the DSD Neonatal Care Pathway for referral to DSD specialist care.

1. Infant is sick and requiring specialist endocrine or surgical intervention
2. Presentation of ambiguous or atypical genitalia
3. A decision on sex of rearing cannot be reached at birth
4. Appearance is not consistent with any prenatal genetic tests
5. DSD is suspected from clinical presentation

3. Neonatal Care Pathway

The local neonatal team should initiate the pathway immediately in response to a baby born with suspected DSD as defined in 2.2, and:-

3.1. Assess and meet immediate clinical needs of baby

Some conditions that are associated with a DSD are also associated with acute medical complications that require urgent treatment, e.g. Salt losing congenital adrenal hyperplasia (CAH). When a baby is born with a suspected DSD the priority will always be to meet their immediate clinical needs.

3.2. Encourage contact between baby and parents

Contact between parents and baby should be encouraged, even where the infant is very ill, to facilitate bonding.

3.3. Provide support to parents

Support for the parents should be offered immediately. Specialist DSD training is not essential for delivery of effective, caring support for parents in the post natal period and can be provided by the Midwife or Neonatal Paediatrician.

In the first 24 – 48 Hours:-

3.4. Initiate Referral and First Tier Clinical Investigations

The Neonatal personnel will refer to their local Neonatal Guidance document and contact the on-call link clinician who will organise the first line investigations set out below. These may include;

- PCR or FISH analysis - Define sex chromosomes (available within two working days)
- Plasma Glucose
- Serum electrolytes
- Any immediate surgical referral e.g. imperforate anus/ cloaca/ bladder exstrophy

3.5. Contact local DSD Link Clinician (Appendix 2)

Once initial investigations have been initiated, the local Link DSD clinician will be contacted to co ordinate care and further investigations. It is essential that any infant with a suspected DSD is assessed by an experienced clinician with knowledge of the range of conditions associated with DSD. The list of DSD Link clinicians by health board can be found in Appendix 2. In most health board areas there will be a Neonatal Guidance Document. (Please note Appendix 2 contact details will be omitted from online version of the pathway)

4. Key Messages

Identification of a DSD can be a disconcerting time for parents. How staff support and communicate with them in these first days can have a lasting impact. SDSA recognise the need to empower local staff to provide consistent, supportive and reassuring messages to families.

4.1. What staff should do:

- Encourage parents to provide all appropriate personal care to their baby, i.e. bathing and feeding the baby.
- Keep parents informed about what is happening. Listen to and acknowledge parents' concerns and questions
- Repeat information given. This is a confusing time for parents and in most cases will be the first time they have heard of these conditions
- Explain to the parents the initial tests that will help us look after their baby, and the colleagues to whom we may refer the baby
- Make peer advice available to parents through the DSD Families website

4.2. What staff should not do:

- Do not make gender specific references, instead use gender neutral language 'baby is'
- Do not make statements to parents about sex of rearing
- Do not get pushed into offering parents an opinion. Explain that we need to gather information from the tests.
- Do not use terms Intersex or Hermaphrodite
- Do not avoid speaking to parents or isolate them, they will need support
- Do not try to manage alone, you should ask colleagues for support if you are uncertain

4.3. Supporting parents to communicate

- What can the parents tell their family?

This is a problem when we are not sure of the baby's gender in a case of DSD. We can help the parents to find a form of words that they can communicate to their family.

Clinical psychology is valuable in this situation. We appreciate this may not be immediately available, but we can help parents sensitively to provide a simple explanation for their friends and family

Example:

The doctors are running some tests on the baby as there seems to be a slight hormone problem, so we're not quite sure what to say at present except that both mum and baby are fine (or are being well looked after).

- The DSD Families website link is useful for all sorts of DSD situations, including when the baby is mistakenly identified as one gender, but is another gender.

5. Resources and support for parents

For parent of newborns with DSD

- <http://www.dsdfamilies.org/docs/brochures/DSD.pdf> 'When your baby is born with genitals that are different (early years)'
- SDSO Website <http://www.mcns.scot.nhs.uk/sdsd/>

5.1. Support Organisations

Support organisations can offer valuable support and information for families. These include:

- AIS (Androgen Insensitivity Syndrome) support group - AISSG.com
- CAH (Congenital Adrenal Hyperplasia) support group - livingwithcah.com
- DSD families - dsdfamilies.org

5.2. Resources and supporting information for Healthcare Professionals

- Julie Griffiths (2015), Dealing with DSD <https://www.rcm.org.uk/tags/midwives-magazine-summer-2015>
- Society for Endocrinology UK guidance on the initial evaluation of an infant or an adolescent with a suspected disorder of sex development <https://www.bsped.org.uk/clinical/docs/CEN.pdf>

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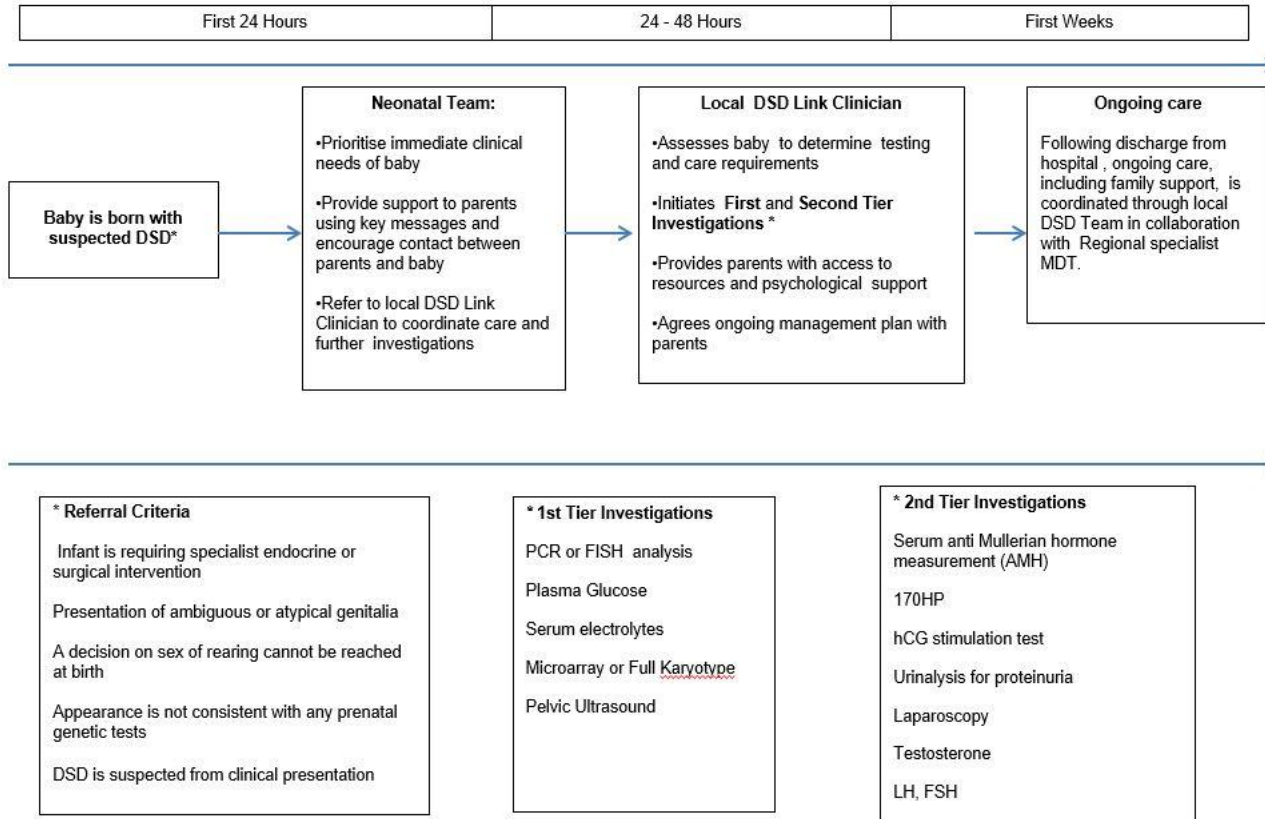
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4. Duguid et al. (2007), The psychological impact of genital anomalies on the parents of affected children, *ACTA PAEDIATRICA* 96, 348-352
5. What to do when your child is born with genitals that look different... <http://www.dsdfamilies.org/>

Disclaimer

This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined on the basis of all clinical data available for an individual case and are subject to change as scientific knowledge and technology advance and patterns of care evolve. Adherence to guideline recommendations will not ensure a successful outcome in every case, nor should they be construed as including all proper methods of care or excluding other acceptable methods of care aimed at the same results. The ultimate judgement must be made by the appropriate healthcare professional(s) responsible for clinical decisions regarding a particular clinical procedure or treatment plan. This judgement should only be arrived at following discussion of the options with the patient, covering the diagnostic and treatment choices available. It is advised, however, that significant departures from the national guideline or any local guidelines derived from it should be fully documented in the patient's case notes at the time the relevant decision is taken.

Appendix 1: SDDS Neonatal Pathway

SDDS Neonatal Care Pathway



Appendix 2: DSD Link Clinicians for NHS Board – point of contact for Referral

(Omitted from online pathway document)

Appendix 3: The clinical members of the SDSD MDT and their roles in providing care to the patient and the parents

Ahmed. F et al. (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, 17 Guidelines 18

Neonatologist or General Paediatrician

- Initial explanation
- Management of the unwell child
- Initiation of first-line investigations
- Seek advice from paediatric subspecialist (endocrine or surgical) with an interest in DSD

Paediatric Endocrinologist

- Detailed explanation over multiple visits
- Management of the unwell child
- Interpreting first-line investigations and planning second-line investigations
- Organize timely and appropriate involvement of other members of MDT
- Act as the link between the parents and MDT
- Initiate and monitor long-term medical therapy such as steroid or sex steroid therapy

Paediatric Radiologist

- Interpret and often perform ultrasound scans on the newborn
- Judge the reliability of ultrasound scans in the newborn especially when the results may influence sex assignment

Paediatric Urologist

- Assessment of external anatomy
- Explanation of the anatomy and results of imaging
- Explanation of pros and cons of reconstructive surgery
- Develop a plan for complex imaging (other than pelvic ultrasound) and further assessment of the anatomy
- Perform procedures such as laparoscopy, biopsy, reconstructive surgery and gonadectomy
- Organize timely and appropriate involvement of other members of MDT

Paediatric Specialist Nurse

- Provide general support to the patient and parents in addition to that provided by other members of the MDT
- Arrange specialist investigations
- Liaise with the rest of the DSD team, including the clinical psychologist

Clinical Psychologist	<p>Provide specialist support to parents soon after birth</p> <p>Provide support to the growing up child and the parents</p> <p>Develop an individualized plan for each family</p> <p>Guide the MDT on timing and tempo of explanation of the condition to the older child and adolescent</p>
Clinical Endocrine Biochemist	<p>Facilitate timely analysis of samples</p> <p>Provide specialist support and interpretation of results</p> <p>Guide subsequent biochemical tests</p> <p>Facilitate storage of samples for analysis at a later stage</p>
Clinical Geneticist	<p>Facilitate timely analysis of karyotype</p> <p>Closer involvement in the child with dysmorphic features</p> <p>Oversee the process of genetic analysis</p> <p>Facilitate storage of samples for analysis at a later stage</p> <p>Genetic counselling</p>
Gynaecologist	<p>Availability at an early stage to discuss future outcome and map long-term care pathway in the affected girl</p> <p>Discuss issues related to sexual function, reproductive function and surgery</p> <p>Assess the understanding and review the diagnosis</p> <p>Assess the need for psychology support in the adolescent girl</p> <p>Initiate and monitor long-term sex steroid therapy</p> <p>Perform examination, investigative and therapeutic procedures in the adolescent girl</p> <p>Oversee vaginal dilator training with specialist nurse</p>
Adult Endocrinologist	<p>Investigate and manage the adolescent presenting for the first time after the age of 16 years</p> <p>Liaise with other members of the MDT</p> <p>Act as the link between the patient and MDT</p> <p>Initiate and monitor long-term medical therapy such as steroid or sex steroid therapy</p>