Clinical Standards for Management of an Infant or Adolescent presenting with suspected differences of sex development (DSD)

These Clinical Standards for management of an infant or adolescent presenting with a suspected DSD have been produced by the Clinical Committee of the BSPED. They are evidence based where possible or developed by consensus with input from clinical experts and stakeholders. The standards are reviewed regularly to include any new evidence.

Overall Aim:

- Improve patient experience
- Improve clinical outcomes
- Improve equity of access to specialist services
- Identify optimal service requirements

Working Group: BSPED DSD Special Interest Group

Introduction

Differences of sex development (DSD) encompass a wide range of conditions usually presenting in the new-born period or adolescence. Their clinical features and underlying physiology/pathophysiology are very variable but usually involve atypical genitalia in infancy, or atypical pubertal development in adolescence. These pose extremely challenging and sensitive clinical presentations to families and professionals, requiring consideration of the appropriate sex of rearing and potential for future adult sexual functioning and fertility.
This requires the input of a specialist multi-disciplinary team with adequate knowledge and experience of the range of conditions that present as DSD, their investigation and management and experience in the complex, sensitive discussions required. It is essential that, whilst addressing the inevitable pressure for urgent assessment and investigation at initial presentation, a medium and long-term plan is also developed and this requires a long-term relationship between the child/adolescent, family and the specialist MDT.

Many of the conditions are rare and data on long-term outcomes are very limited. With increasing knowledge about DSD there has also been a major shift in societal views towards DSD. Aspects of previously standard clinical management are being challenged by patient groups and professionals in view of concern about long-term outcomes. It is therefore essential that strategies are developed to ensure the best care is available to patients and their families through access to specialist MDTs, that appropriate standards of care are set and reviewed and that experience and expertise is shared and outcomes recorded. This will ensure the evidence base develops to inform the best care in the future. Crucially the views of the patient and their families must be incorporated into developing services and clinical pathways.

A. Clinical Standards for Management of an Infant or Adolescent presenting with suspected differences of sex development (DSD)

No	Standard	Adapted from Source
Specialist/Network centre accreditation		
1	I. All centres managing children/young people with DSD should have a specialist multi-disciplinary DSD team that can be accessed by its regional network. II. The specialist DSD MDT must include: Endocrinology (Paediatric & Adult) Urology/Surgery Clinical Psychology/Psychiatry Clinical Genetics Radiology Gynaecology	1 -4
2	All children/young people presenting with a suspected differences of sex development (DSD) should be recorded in an approved DSD registry	2, 13
3	There should be provision to share, after 'opt in' agreement by parents, data recorded in the DSD Registry locally, nationally, and internationally as agreed by the family and the specialist DSD network.	2, 13
4.	All regions should have an agreed regional network and clinical pathway for the referral of children/young people presenting with a suspected difference of sex development (DSD) to the specialist DSD MDT	1,2,4, 14
5.	Each specialist multi-disciplinary DSD team can provide evidence of participation in audits/quality improvement projects related to their clinical activity recorded in 2 and principals of management outlined below.	13, 14

6.	Members of specialist multi-disciplinary DSD team should participate in national and international DSD related CPD	14

B.

B. Principals of Management of Infants or Adolescents presenting with suspected differences of sex development (DSD) agreed by Delphi Consensus Process organised through the BSPED Clinical Committee and DSD SIG April-August 2017

No	Principal of Management	Adapted from Source
Equity of service		

1	I. Evaluation, investigations and discussions of a child/adolescent with a suspected DSD diagnosis should only be carried out by a specialist DSD MDT along a definitive pathway agreed by the network.	Delphi Consensus 100%
	II. All neonates with a suspected DSD should be referred and discussed with a member of the specialist DSD MDT within 2 working days	Delphi Consensus 97%
	III. All children/adolescents with a suspected DSD diagnosis should be discussed within a week with a member of the specialist DSD MDT, to make a plan for referral, appropriate investigations and further management.	Delphi Consensus 96%
	IV. Every family with a child/young person with a suspected DSD diagnosis should be encouraged to engage with an experienced DSD Psychologist /Psychiatrist/Mental health professional who is an integral part of the specialist DSD team.	Delphi Consensus 100%
	V. All families with a child/adolescent having evaluation, investigations and discussions for a DSD diagnosis should have a named key worker within the specialist MDT	Delphi Consensus 90%
	VI. All families with a child/adolescent having evaluation, investigations and discussions for a DSD diagnosis should be offered access to high quality peer reviewed information.	Delphi Consensus 98.5%
Professional Expertise		

2	I. All children/young people with a suspected DSD diagnosis should have relevant endocrine investigations supervised by a Paediatric Endocrinologist who is part of a specialist DSD MDT. Testing should occur at a centre with access to a full range of hormone evaluations at accredited laboratories.	Delphi Consensus 100%
Diagnostics		
3	I. Endocrine investigations should be started within the first week for a neonate presenting with a suspected DSD. II. All children/young people with a suspected DSD diagnosis should have access to age-appropriate specialist imaging guided by a specialist DSD MDT which includes paediatric/specialist radiologists as integral DSD MDT members. III. All children/young people with a suspected DSD diagnosis should be offered investigation with a full range of specialist diagnostic genetic investigations and advice guided by a specialist DSD MDT which includes Clinical Genetics expertise as an integral part of the DSD MDT.	Delphi Consensus 98% Delphi Consensus 98.5% Delphi Consensus 98.5%
Patient Management		

4	I. All management decisions of the specialist DSD MDT and network must reference discussions with the patient (and their family for patients requiring parental/guardianship consent), and include patient/parent opinions as an integral part of the clinical decision making process. II. All families with a child/adolescent with a DSD diagnosis should be offered details of appropriate Peer Support Organisations	Delphi Consensus 100% Delphi Consensus 98.5%

C.

Patient Experience		
5	I. All specialist DSD MDTs must have a mechanism to enable patient/ family to feedback their views and opinions to the DSD MDT.	Delphi Consensus 98.5%
Transition		
6.	I. The DSD MDT must have an appropriate Transition pathway to adult care and ensure this is explored and discussed with each young person with a DSD diagnosis.	Delphi Consensus 100%

References

1. Ahmed SF, Achermann JC, Arlt W, et al. 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*. 2016;84(5):771-788.

2. Hughes I.A., Houk C., Ahmed S.F. *et al* (2006) Consensus statement on management of intersex disorders. Archives of Disease in Childhood, 91, 554–563.

3. SSNDS Definition No.23 Specialised Services for Children (3rd Edition) 2009/10, Specialised Paediatric Endocrinology and Diabetes Services pg 40. National Specialised Commissioning Group http://www.ncg.nhs.uk/index.php/key-documents/specialised-services-national-definitions-set/

4. UK Standards for Paediatric Endocrinology 2010 http://www.bsped.org.uk/clinical/docs/BSPEDPaediatricEndocrineStandardsvs130710.pdf).

5. British Society for Paediatric Endocrinology and Diabetes website: http://www.bsped.org.uk/professional/guidelines/index.htm

6. RCN Competencies: an integrated career and competency framework for paediatric endocrine nurse specialists, 2008, Royal College of Nursing

7. Medicines Standard: National Service Framework for Children, Young People & Maternity Services 2004 www.dh.gov.uk

8. General Assembly of the United Nations (1989). The Convention on the Rights of the Child. Adopted by the General Assembly of the United Nations on 20th November 1989

9. Medicines adherence Involving patients in decisions about prescribed medicines and supporting adherence, 2009 NICE clinical guideline 76

10. Reference guide to consent for examination or treatment, second edition 2009 www.dh.gov.uk

11. Working Together to Safeguard Children. A guide to inter-agency working to safeguard and promote the welfare of children, March 2010, Department of Children, Schools and Families, HM Government http://publications.dcsf.gov.uk

12. Transition: getting it right for young people. Improving the transition of young people with long term conditions from children's to adult health services 2006 www.dh.gov.uk/childrens nsf.

13. Commission expert group on rare diseases - European Commission.

http://ec.europa.eu/health/rare_diseases/publications accessed 05 Aug 2016

14. Kyriakou et al. Orphanet Journal of Rare Diseases (2016) 11:155

DOI 10.1186/s13023-016-0534-8