



For immediate release

## **New UK guidance on the initial evaluation of a child with a suspected disorder of sex development**

New guidance from the Society for Endocrinology highlights the importance of a multidisciplinary team in diagnosing children with a suspected disorder of sex development (DSD). The guidance is published in the journal *Clinical Endocrinology*. This is the first time guidance on the best practice for initial evaluation and diagnosis of a DSD has been applied to a UK clinical setting.

Disorder of sex development is an umbrella term applied a wide range of conditions, present from birth, where the development of chromosomal, gonadal and/or anatomical sex differs from expected. A DSD is most often diagnosed at birth (if the appearance of the external genitalia is ambiguous) or at puberty (for example, a boy with pubertal delay, a girl who develops male characteristics or a girl who does not start menstruation). Approximately one in 300 newborns may be born with a concern about the development of the external genitalia. However, the prevalence of complex anomalies where the sex is unclear upon expert examination is more in the region of one in 5000 births.

Managing these clinical situations can be very complex, both for parents and the medical team, especially when the sex of rearing is uncertain. This guidance aims to bring together good clinical practice and standardise the UK approach to diagnosing DSDs in infants and children.

### **The guidance recommends:**

- Infants or adolescents with a suspected DSD should be treated by an experienced multidisciplinary team, accessible through a regional centre. As a minimum, this team should include specialists in endocrinology, surgery and/or urology, clinical psychology, radiology, nursing and, for infants, neonatology.
- One main contact person should be assigned to each family; in most cases, this will be the paediatric endocrinologist. The family should be told of the range of support available to them and provided with contact details for these personnel. They should also be made aware of the stepwise process used to diagnose and manage DSDs, with the ultimate goal of achieving long-term well-being.
- Access to specialist psychological support during and after the diagnostic process is essential for both the affected person and their parents. In addition, any adolescents with an existing DSD who need medical/surgical attention should be routinely offered clinical psychological support.

The pace of how information is shared should be set by the family and issues of confidentiality discussed and respected.

- The exact tests used to diagnose a DSD will vary between patients. However, as a minimum, the multidisciplinary team should take into account the patient's external appearance, internal anatomy, genetic make-up and hormone profile. The most important goals of the initial assessment period are to support the affected child and parents, assign a sex of rearing (in infants) and exclude the possibility of any early medical problems.
- Patient groups can provide important psychological support and information to affected individuals and their families. Contact details of relevant support groups should be provided as routine by healthcare professionals upon diagnosis.
- All medical personnel involved in the care of a patient with a DSD should have access to a regional DSD team. The team has a responsibility to educate other healthcare staff (including the family's primary physician) and should have a regular forum where they meet to discuss the case and review its own performance.

**Prof Faisal Ahmed, Chair of the DSD guidance taskforce, said:**

*“The aim of this guidance is to support clinical professionals in the initial evaluation and diagnosis of children with suspected disorders of sex development and to provide a framework to standardise clinical practice throughout the UK. It is of paramount importance that a child with a suspected disorder of sex development is assessed by an experienced multidisciplinary team.*

*“Having a child diagnosed with a disorder of sex development can be a very traumatic time for families. It is essential that they are offered specialist psychological support, both at initial evaluation and later on once diagnosis has been confirmed.”*

**About this guidance**

This guidance was produced by the Society for Endocrinology in association with the Androgen Insensitivity Syndrome Support Group, the Association for Clinical Biochemistry, the British Society for Paediatric Endocrinology & Diabetes, the British Society for Human Genetics, British Society of Paediatric & Adolescent Gynaecology, the British Society of Paediatric Radiology, Climb CAH Support Group, the Clinical Genetics Society and the Royal College of Nursing.

A UK DSD taskforce was convened to develop guidance on the initial evaluation and diagnosis of patients with a suspected DSD. Taskforce members took responsibility for individual sections and based their findings on observational studies and expert opinion following group discussion. Prior to publication, the guidance was subjected to open external review from the professional societies involved and their members, as well as patient group representatives.

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### Notes to editors

This guidance is published in full in *Clinical Endocrinology* (2011), **75**: 12-26. DOI: [10.1111/j.1365-2265.2011.04076.x](https://doi.org/10.1111/j.1365-2265.2011.04076.x). *Clinical Endocrinology* is the official clinical journal of the Society for Endocrinology. Visit *Clinical Endocrinology* online [here](#).

**For a copy of the full guidance, please click on the link above.**

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A patient information factsheet on this guidance is available on the SfE public information website, You & Your Hormones: [http://www.yourhormones.info/topical\\_issues/](http://www.yourhormones.info/topical_issues/)

The **Society for Endocrinology** is Britain's national organisation promoting endocrinology and hormone awareness. [www.endocrinology.org](http://www.endocrinology.org).

The **Androgen Insensitivity Syndrome Support** is a UK based charity that supports adults, families and young people affected by androgen insensitivity syndrome and related conditions. [www.aissg.org](http://www.aissg.org)

The **Association for Clinical Biochemistry** (ACB), based in the United Kingdom, is a professional body committed to ensuring that laboratories use the best possible tests when investigating a patient's illness to ensure they get the most effective treatment/advice available. [www.acb.org.uk](http://www.acb.org.uk)

The **British Society for Paediatric Endocrinology & Diabetes** (BSPED) is the UK national society for professionals who coordinate the care of children with growth, endocrine and diabetes disorders, including DSD. [www.bsped.org.uk](http://www.bsped.org.uk)

The **British Society for Human Genetics** is an independent body representing UK human genetics professionals. [www.bshg.org.uk](http://www.bshg.org.uk)

The **British Society of Paediatric & Adolescent Gynaecology** is a multidisciplinary society aimed at bringing together professionals interested in the study and practice of paediatric and adolescent gynaecology, and promoting research and education in the field. [www.britspag.org](http://www.britspag.org)

The **British Society of Paediatric Radiology** is the professional organisation that represents the interests of Radiologists involved in the field of imaging children. [www.bspr.org.uk](http://www.bspr.org.uk)

**Children Living with Inherited Metabolic Diseases** and the **Climb CAH Support Group** provide information and support to families and sufferers affected by CAH, raise awareness of these and other Inherited Metabolic Diseases and fund research. [www.climb.org.uk](http://www.climb.org.uk) and [www.livingwithcah.com](http://www.livingwithcah.com)

The **Clinical Genetics Society** (CGS) brings together doctors and other professionals involved in the care of individuals and families with genetic disorders to advance and promote the science and practice of Clinical Genetics. [www.clingensoc.org](http://www.clingensoc.org)

The **Royal College of Nursing** (RCN) is the voice of nursing across the UK and is the largest professional union of nursing staff in the world. The RCN promotes the interest of nurses and patients on a wide range of issues and helps shape healthcare policy by working closely with the UK Government and other national and international institutions, trade unions, professional bodies and voluntary organisations. [www.rcn.org.uk](http://www.rcn.org.uk)

## ABSTRACT

### UK guidance on the initial evaluation of an infant or an adolescent with a suspected disorder of sex development

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It is paramount that any child or adolescent with a suspected disorder of sex development (DSD) is assessed by an experienced clinician with adequate knowledge about the range of conditions associated with DSD. If there is any doubt, the case should be discussed with the regional team. In most cases, particularly in the case of the newborn, the paediatric endocrinologist within the regional DSD team acts as the first point of contact. The underlying pathophysiology of DSD and the strengths and weaknesses of the tests that can be performed should be discussed with the parents and affected young person and tests undertaken in a timely fashion. This clinician should be part of a multidisciplinary team experienced in management of DSD and should ensure that the affected person and parents are as fully informed as possible and have access to specialist psychological support. Finally, in the field of rare conditions, it is imperative that the clinician shares the experience with others through national and international clinical and research collaboration.