Brochure on dsd conditions for non-specialist clinicians

Many clinicians feel confused when a patient presents who is diagnosed with or suspected to have a Disorder / Difference or Variation of Sex Development (DSD or intersex condition).[1] Because some of these conditions are very rare, correct information is often difficult to find.

Below we have gathered information that may be of help in finding answers to some of the general questions clinicians not specialized in DSD may have about DSDs. Some websites describe typical and atypical sexual development, others describe specific DSD conditions. A number of issues that come up repeatedly in the clinical care of people with DSD are described in this brochure as well.

The information is available in six languages; English, Dutch, French German, Polish and Swedish.

What is a DSD?

Most people assume that girls have girls' genitals, female reproductive organs, female hormone levels and a female chromosome pair (XX), whereas in boys all the characteristics are typically male (testes, testosterone, XY chromosomes). Yet this is not always the case. In a number of individuals some parts of their development will have been typical for boys and other parts will have been typical for girls. These situations are more common than most imagine, and in many cases do not involve medical interventions.

DSD is an umbrella term for a variety of conditions, and within DSD they are categorized into groups with similar features. The most prevalent DSD conditions are categorized as follows:

1. Conditions with an uncommon sex chromosome configuration, such as Turner syndrome, Klinefelter syndrome, mixed gonadal dysgenesis (45,X0/46,XY), and 46,XY/46,XX.

2. 46,XY chromosome conditions, including testicular dysgenesis (underdevelopment of the testes) and disturbances of testosterone synthesis or action.

[1] There is currently a debate about the most preferred term.. In this document we will use the term DSD, because this is currently the most often used term among clinicians.

3. 46,XX chromosome conditions, including ovarian dysgenesis (underdevelopment of the ovaries), congenital adrenal hyperplasia (CAH), and non- development of the uterus or vagina.

People who have a DSD are not all alike. There is much variation between DSDs and not all that is known about a specific DSD will be applicable to every child with that type of DSD.

The following websites provide an overview of these various conditions. In this brochure existing information on the various conditions is brought together.

Websites with general information relating to typical and atypical sex development and DSD in general

English
http://www.dsdgenetics.org/
http://www.dsdgenetics.org/index.php?id=2
http://www.dsdgenetics.org/index.php?id=7
http://www.dsdfamilies.org/index.php
http://www.aboutkidshealth.ca/en/howthebodyworks/sexdevelopmentanoverview/pages/default.aspx
http://www.nhs.uk/Conditions/Disorders-Sex-Development/Pages/Introduction.aspx
http://www.yourhormones.info/

Importance of multidisciplinary teams

Depending on the time at which people learn about their own or their child`s condition, they might face a number of medical and psychological issues that they may never have heard of before. Even their family doctors will not have answers to all of their concerns.

Most people with a DSD will develop into happy and healthy individuals. They may nevertheless require support from health professionals other than medical doctors. This type of care is offered by some, but unfortunately not all, DSD teams. Holistic management in DSD implies that, besides medical treatment, a team can offer the families ample emotional and psychological support.

A multidisciplinary team (MDT) for DSD will help families to understand the diagnosis and guide them through any intervention that might be needed. Such teams should include a (paediatric) endocrinologist, a (child) psychologist, an urologist and, later in life, a gynaecologist and possibly a plastic surgeon. Other disciplines may also be involved, such as a geneticist. These experts piece together different aspects of the DSD in order to help to understand which type of intervention, if any, may be the right one for the person with DSD . This will help to make truly informed consent decisions on these interventions.

Such a specialized MDT needs to liaise with the family doctor to ensure that the information is well communicated. If no contact has been made yet with a MDT, the family doctor may help to find a centre that ensures multidisciplinary care.

For psychologists it is important to know the impact of the many challenges. Parents often struggle with questions such as “How will my child be treated in school? Who can I rely on? Will my child be able to enjoy sexuality? Will my child have a family?” A multidisciplinary specialized team should help the family to find their way through these questions. Clinicians should encourage parents and older persons to ask for help (e.g. counselling) elsewhere, if such help is not available in the hospital where the person is treated. Sometimes, due to long distances from the specialized hospital, psychological care closer to home might be preferred.

Information management

Families with a child or adult with a DSD face many questions. Some relate to the *information they are receiving* from healthcare professionals and other sources. Doctors might not always realize that parents or family members do not understand all information that is given so clinicians should be aware of this and check whether the information they provide is understood. Caring for a child with a DSD and understanding their condition is complex, and even good explanations are not always easy to comprehend.

Other important issues concern the *information on the condition a parent or clinician gives to the child*. Parents and clinicians may wonder when they should share information, what kind they should give, who should do it and what words they should use. Parents may feel insecure about such matters and may want to know where they can get guidance and support, either within the specialized team or elsewhere. Internet information is increasingly available, such as the example booklet " Amazing you" written by a mother. Available at www.dsdfamilies.org

What the child should know very much depends on his or her condition and at what age the diagnosis is made. When children are very young they may need explanations for why they visit doctors, why they need to use medication and why they might look different from other children. Somewhat later, when they approach puberty, they may need information about why they have to use hormones. With time, various aspects of their condition may be explained, including information about reduced or absent fertility. When they approach adulthood the team should have given them complete information about their DSD. As an adult, they should never unexpectedly be confronted with unknown aspects of their DSD.

Persons with DSD and their parents (when the child is young) may also wonder what others should know about the condition. However, there is not one single correct answer to this. Clinicians can help, to find a balance between openness (about certain aspects) and keeping information on the condition private.

Boys or girls?

Some girls or boys may not behave as gender stereo-typical girls or boys. For instance, a girl may like boys' toys and activities, prefer to play with boys and dislike playing with girls. This does not mean that she is unhappy about being a girl and about living like a girl.

Very few children with DSD question their sex, but some may feel confused for shorter or longer periods of time when they grow older. This may happen even if there are no visible signs of their DSD and while nobody ever questioned whether they are male or female. Such concerns, whilst understandable, are unfortunate. They may lead to both parents and (older) children worrying about something which might not necessarily need to matter to them.

Instead, what really counts is how they experience themselves, whether that be as a male, a female or differently (see the sites below). Even more important is whether others acknowledge and respect who they are. No body characteristic can override their feeling of being male or female. A girl with typically male XY chromosomes who has always experienced herself as a girl ***is*** a girl. Trying to force someone into an identity is harmful.

If DSD patients are raised as girls they usually experience themselves as girls; if they are raised as boys they usually experience themselves as boys. But a small number of children might need help in untangling their feelings about being a boy or a girl. Some of them may eventually develop an identity that is neither typically male or female. The few children who are clearly very unhappy about being a boy or a girl for long periods of time may want to talk to people who are specialized in this area. See below for sites of specialty clinics.

Sites on gender

http://binarythis.com/
http://everydayfeminism.com/2014/03/intersex-awareness/

Gender identity services

UK (specifically for children and adolescents)
http://www.tavistockandportman.nhs.uk/care-and-treatment/information-parents-and-carers/our-clinical-services/gender-identity-development

Clinical care of DSD

As soon as clinicians are confronted with a child with DSD they should refer the child to a specialized centre and inform the family about the procedures they can expect. The precise diagnosis of a child may influence decisions as to which interventions or treatments should be used, or whether they should be used at all. Each parent or guardian should be informed about timeframes for when these medical interventions might be offered or applied.

-Some patients (with CAH) may experience a life-threatening condition referred to as 'salt wasting'. Most persons with a DSD do not face a medical emergency. Yet the care for children with these relatively unknown conditions may be perplexing for clinicians .

-Sometimes external and/or internal genitalia of the individual may look different from those of other people ['Early Days' made by 'dsdfamilies' is a very clear and extensive brochure and available on the dsd LIFE website in various different languages]. At birth, these children can only after careful examination be assigned to the male or female sex.

-Removal of the testes may be necessary if hormone production needs to be stopped or if there is an increased chance for malignancy if they are not removed.

-People with DSD may need hormone therapies. In some cases it may be necessary to start medication (e.g. cortisone) at birth. In other cases, hormone therapies will start at puberty. If ovaries or testes have not developed, do not produce enough hormones or have been removed, hormone treatment is necessary to start puberty and keep the bodies of the children healthy as they grow older.

-After childhood , some individuals may want to have plastic genital surgery, whereas most will be satisfied with their bodies.

-Young adults who wish to enjoy intercourse may need treatment to make this possible. The non-surgical Do-It-Yourself method of pressure dilatation gives better results than surgery, and should be tried first (with support of gynaecologist and psychologist).

-Early monitoring of bone density may be advised since some DSDs appear to carry a high risk of osteoporosis.

DSDs often require complex decisions relating to clinical care and other important aspects of life. Families need guidance and support in making these decisions. For instance, in cases where sex assignment is not straightforward, parents may wonder what they should tell other people about their child before the sex assignment has taken place and the child does not have a first name yet. As soon as children are old enough they should be involved in making these decisions, but they might need more or different support than ~~that~~ their parents required.

Ethical issues

Surgery to the genital area, particularly in young children, should be considered very carefully as it may lead to problems such as the alteration of sensation, and the development of scar tissue which may limit the treatment options later in life. In addition, it is important that children can make their own decisions about their bodies and therefore wait until they can participate in decision making.

Irreversible medical procedures such as surgery or hormone treatment should therefore be postponed until a child is capable of giving consent to any medical intervention as they mature. Allowing children to understand and agree to their own clinical care gives them control over their lives and may better equip them to overcome challenges in their lives and develop healthy self-esteem.

Realize that …

Care providers who have no experience in the field of DSD should **no**t provide care relating to DSD if they are not in close collaboration with specialized centres.

If at all possible, it is better to wait until someone is old enough to be able to provide full consent before irreversible interventions are planned. Such interventions should not be undertaken without age appropriate explanation to the child.

When it comes to treatments, whether they be hormonal or surgical, enough time in advance should be taken to understand the interventions and plan the next steps. Only very few interventions are urgent in a young life. Parents may need the support of family doctors or other health care providers when they need to make difficult decisions when a child is too young to make these.

Clinicians should be aware that adults with DSD, parents of a child with DSD and the child with DSD might need persons outside the family who know about the diagnosis and can give support.

Websites on CAH are

http://www.aboutkidshealth.ca/En/HowTheBodyWorks/SexDevelopmentAnOverview/CongenitalAdrenalHyperplasiaCAH/Pages/default.aspx
http://www.childgrowthfoundation.org/CMS/FILES/06_CongenitalAdrenalHyperplasia.pdf
http://www.dsdgenetics.org/index.php?id=33
http://www.dsdgenetics.org/index.php?id=34

Booklets on CAH are

Congenital Adrenal Hyperplasia, a guide for parents and patients. Obtainable at 'The Growth Foundation', 2 Mayfield Avenue, Chiswick, London, W4 IPW

Websites on AIS are

http://www.aissg.org/pdfs/Broch_AISSG_UK.pdf
http://www.dsdgenetics.org/index.php?id=31
http://www.dsdgenetics.org/index.php?id=32
http://www.dsdgenetics.org/index.php?id=34

Websites on Turner syndrome are

http://www.geneticalliance.org.uk/self-management-turner-syndrome.html

Websites on Klinefelter syndrome are

http://www.scotgen.org.uk/documents/Klinefelters%20for%20professionals.pdf
http://www.dsdgenetics.org/index.php?id=32

Booklets on Klinefelter syndrome are

English
Klinefelter Syndrome, an information sheet for professionals,