
Disclosing Disorders of Sex Development and Opening the Doors

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Key Words

DSD · Full disclosure · Informed consent

Abstract

This paper focuses on the importance of full disclosure in disorders of sex development (DSD), as a universal human right and closely related to informed consent. Full disclosure is not only a way of communicating a diagnosis, it is a methodological constant that permeates all the clinical moments expressed by a multidisciplinary team. As stated by The Chicago consensus, DSD should be referred to specialized centers of excellence. In these centers provided with the necessary multidisciplinary team that is able to: provide knowledge, skills and experience; deliver quality and care, and cope with the emotional barriers that often hinder the practice of full disclosure. Full disclosure is important when a person is informed about something, can participate in making a decision or is advised about something that needs to be done before he/she will be able to make a choice. However, if a person is informed about something that was done unnecessarily and could have been postponed until he/she could have been involved in making the decision, full disclosure can be seen as deception colored by rage, sorrow and regret.

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Introduction

Knowledge about one's medical condition is a basic human right. Although it has not always been the case, it is now generally accepted that full disclosure of medical information is integral to care provision for persons with disorders of sex development (DSD) [Slijper et al., 2000; Wisniewski et al., 2000; Creighton and Minto, 2001; Creighton et al., 2001; Carmichael and Ransley, 2002; Brinkmann et al., 2007; Liao et al., 2010]. Accurate information, communicated sensitively and empathically to the patient, is fundamental in the process of sense-making for the patient. With support, it can initiate the process of self-acceptance, the development of a coherent and integrated self-identity.

However, optimal communication about DSD is easier said than done; it demands 'advanced communication skills' from the clinician [Liao et al., 2010]. Even then, our restrictive, binary language of sex and gender may render the task too emotionally challenging for some clinicians and parents. In this paper, I argue for a relationship between the level of maturity and integration of the DSD team and its members' capacity for positive interaction with the patient. Care givers' feelings of safety and integration can positively influence care recipients' sense of

safety and capacity for integration. Most of all, I argue that in a mature team, we as clinicians can challenge ourselves to think outside the box, and that in doing so, we may be more able to assist service users to do likewise.

Concealment

In the not too distant past, the belief that concealment was necessary to preserve psychological well-being was widespread. DSD clinicians withheld information from patients on the premise that the reality of DSD was impossible to live with and that complete medical knowledge would lead to unbearable suffering. Surgical sexing of the body in early life followed by unambiguous sex of rearing was considered pivotal; it was a corollary of the policy of concealment [Money et al., 1955].

With the changing ethos in clinical practice, many people with DSD have stated the opposite. Some say that far from being a risk to their psychic equilibrium, the moment when they were clearly told about their diagnosis was the moment when some of their experiences began to make sense. (All the clinical quotations in this paper come from my personal practice, from direct interviews with patients or collected in AIS support group meetings of AISIA [Italian AIS Association], www.sindromedimorris.org.)

Suzy knew about her condition when she was 16 years old and presented at a gynecological clinic with primary amenorrhea. She, along with her parents, was given full information about her diagnosis and what that might imply. She said, 'I felt greatly distressed, and sometimes I feel the same now, but like all painful experiences it has helped to strengthen my character. It has matured me quite a lot and made me understand what makes life worth living. Sometimes I feel a little sad, but my basic character hasn't changed.'

Like Suzy, the reactions of the following 2 people who described their moment of disclosure were characterized by a sense of relief. If relief was accompanied by a sense of sadness, it was not experienced as unmanageable.

'After I found out the truth, I had a feeling of peace because I found an answer to the many doubts and to the many anxieties that I had had since I was a small girl, not being able to find a reason for all those examinations, the therapy, the lack of menses, the physician's comments, and all the rest.'

'I felt a grief that left behind a veil of sorrow and of awareness. I felt as if I had lost my identity and I felt disappointed that I had not been informed before. All those

feelings were softened by being able to talk about my condition and share it with someone.'

Many have said that the secrecy had contributed to their suffering far more than the condition itself [Liao, 2003], as for Hanna.

Hanna is 26 years old and was referred to a clinic in her early adolescence due to what was later identified as a form of congenital adrenal hyperplasia (CAH). 'My parents, possibly because they did not want to make me suffer or because also they did not know about it themselves, never told me. Even now I don't know my condition well. I grew up thinking that it was something momentary, that I had to take pills for something that sooner or later would pass. Then, as the years went by, I realized that something was wrong, and as my parents didn't want to talk about it, I tried to get information by myself. I was so disappointed that I refused to know more. Now, I am starting to accept all that it implies, but it is very difficult. All this not speaking meant that I accepted the things later. I was conditioned by growing up with parents that did not speak and physicians that spoke little and in very difficult terms.'

Making Sense of Past Secrecy: A Tale of Two Women

Eleanor is a 25-year-old girl. She has complete androgen insensitivity syndrome (CAIS) and she doesn't know anything about her situation. Her parents wanted to start to tell her something, as nowadays full disclosure is proposed as the best practice. In all the previous years, they were scared to traumatize her by telling her the truth about her condition. The truth was that she had a XY karyotype. The 8-hour surgery she underwent when she was 9 years old was to remove her testis and to create a new vagina, not the ovary removal she was told she had to undergo for gynecological reason. She faced this with the help of her parents, and during the weeks of post surgery she was told she could not have children. She also faced the necessity of using dilators in her childhood and adolescence. She faced every necessary and inevitable procedure due to her medical situation. Now she has to face the disclosure of her XY chromosome and that she had testis removed not her ovary.

The importance of the disclosure is related to the fact that her parents did not want to conceal any aspect of her condition from her since they think that they can trust her capacity to take care of herself. However, what will she think when she realizes that she could have spared the burden? And what would she think about her parents af-

ter more than 15 years of medical and surgical treatment carried out in secretiveness and loneliness?

Full disclosure is most important when it involves people in decision-making, not when 'the die is cast' and a door is closed.

How would the situation have changed if her parents had been advised to wait, since in CAIS there is no necessity to remove the testis and to perform a vaginoplasty in childhood? What different meaning could there have been with the progressive disclosure of her condition in order to decide whether and when to remove the gonads and to decide to have a vagina when necessary for her sexual life, with a much less invasive operation and with a motivation to perform the necessary dilating maneuvers?

Now, full disclosure is important to explain why she had gone through all of this, to break the wall of secrecy and concealment that surrounded her, to make it up with her parents and to relieve them of the burden of secrets and lies they were forced to carry. Now the 'die was cast', and the possibility of taking care of her body and making her own decision about herself was closed.

Without full disclosure, a person receives surgical procedures performed under a false pretext. To be deceived in this way is to be pulled along the path of rage, sorrow and regret. Full disclosure is important in its own right, but it paves the way for informed decision-making in relation to investigations and interventions. Ideally, it should be accompanied by psychological support that can lead to coping and management strategies on the effects of the DSD.

Eloise

Eloise is a 22-year-old girl; she also has CAIS. She followed almost the same road. Her condition was diagnosed when she was 8 years old, she had a vaginoplasty when she was 12 and full disclosure when she was 18. The vaginoplasty left a scar in her vagina and leakage that became the reason for her continuous complaints towards her parents who she felt were responsible for her present condition. She was fully informed about her situation; she is in contact with a support group, so she is fully informed that the 9 h she spent on the operation table, the following 3 weeks of hospitalization, the continuous dilation performed by her parents that upset her childhood, the scar, and the leakage were not necessary.

'Where the hell were you when they did that to me? Were you drunk when you allowed them to do that?' are her daily complaints to her parents. The scar in her vagina and the shame for her continuous leakage prevent

her from having any contact with boys, and she doesn't feel she can use her vagina. This part of her body is a secret to be hidden away and has become the source of all her troubles.

Integration versus Fragmentation

In Kanzakis' [2008] detailed interrogation of past DSD management, there is a striking fragmentation between knowledge and action. I call one of the axes a 'horizontal' fragmentation of the medical and the psychological aspects, with the former involving medical practitioners and scientists, such as neonatologists, geneticists, pediatric endocrinologists, and pediatric surgeons, and the latter involving psychological practitioners. This rather restrictive vision of team work may give rise to a collection of disjointed parallel professional activities without convergence towards a common goal and may give a message to individuals and families that different areas of their experience may not fit together.

The split between the psychological and the medical appears to be greater in DSD than in other clinical situations. For instance, psychological sessions are frequently necessary after the medical and the surgical parts and the psychological intervention is required to help patients to cope with something still unknown. Perhaps the emotions generated by patients with DSD are so threatening that they trigger a natural recoil amongst clinicians and institutions [Sutton, 1998]. Full disclosure is a litmus paper of how active these protective mechanisms are. The daily difficulties in creating a framework supported by a fluid language that enables us to integrate the multiple perspectives of physicians, surgeons, psychologists, families, and patients show us how much work there is still to be done.

I call the second axis a 'vertical' fragmentation, the separation between pediatric and adult perspectives, between parents' of children with DSD, the children themselves, and adults with DSD. The lack of integration has resulted in a fragmented knowledge about clinical outcomes of pediatric intervention and subsequently experiences across the life span.

A New Chapter

The Chicago Consensus Statement [Hughes et al., 2006] signals a new chapter in clinical management of atypical sexed anatomies. It replaced previous terms such

as 'intersex' and 'hermaphroditism' with DSD. It also attempted to respond to some of the difficulties documented by service users and clinicians, by endorsing 5 care principles for all DSD services:

(1) gender assignment must be avoided before expert evaluation in newborns;

(2) evaluation and long-term management must be performed at a center with an experienced multidisciplinary team;

(3) all individuals should be assigned a gender;

(4) open communication with patients and families is essential, and participation in decision-making is to be encouraged;

(5) patient and family concerns should be respected and addressed in strict confidence.

These principles safeguard people's right to be informed about their condition and uphold the concept of informed consent to interventions by DSD patients or their parents in case of children. They argue for expert, multi-disciplinary care from centers of excellence with the necessary knowledge, skills and experience to deliver quality care.

Emotional Barriers

Opinions about DSD are often rooted on a concrete way of thinking that had a great influence on generations of doctors that dealt with DSD and also are deep down inside of us today in the moments of particular uncertainty.

Further studies should be focused on the specific emotional aspects linked to the encounter with DSD.

Emotional aspects linked to DSD led us to focus on the concrete reality of the body: we need help in order to have access to a more symbolic way of thinking, going beyond the binary logic of yes/no, presence/absence and male/female.

The impact with a pathology, above all severe and chronic illness, puts health care operators in contact with deep anxieties and the inner fantasies that accompany everyone from their childhood on [Menzies, 1960]. Professional choices often derive from the attempt to take care of the parts that we, as therapists, feel to be immature, ill, not valued or irreparably damaged in ourselves. We may say that the ensuing professional maturational process is a progressive coming to terms with these parts in a more realistic way, living up to fantasies of omnipotence and recognizing our limits and the limits of our therapeutic possibility.

One of the possible reasons why the practice of the past dies hard is that the encounter with DSD, except for a few specialized centers, stands as an exceptional event in the routine of most health care professionals.

The presence of DSD reactivates fantasies, fears and conflicts; it exposes operators and parents to areas of anxiety and worry that shake the foundations of their emotional balance. Our emotional balance is achieved progressively from early infancy, working through primal identification with parents, sexual and aggressive impulses, curiosity about child sexuality, bisexual fantasies, and anxiety linked to the discovery of sexual differentiation.

If we do not cope with the irrational emotional factors linked to DSD, also our ability to think may be hindered. Sutton [1998] recommended that in this field we should be able to maintain an 'ability to think properly'.

'The crux of working (in this field) is that we must simultaneously put our competencies, which have been developed at least in part from the healthy sublimation of those original infantile sexual interest, to work on issues that were originally the objects of those interests. The freedom to explore, investigate and learn will have been influenced by the extent to which the process has been one of healthy harnessing of the energy and vitality of these early impulses. The converse is that splitting and denial occurs; these impulses are encapsulated in an area of the mind where they cannot be used because a threat is contained in them which has to be controlled, but at cost to the overall functioning of the individual. Mental work has to be done to keep them in their place, and experiences that might somehow incite them, giving them more force, have to be avoided' (ibid.).

The Importance of Team Development

The story of Georgia, a newborn baby with apparently female genitals and XY chromosome referred to our clinic, gives hope. It illustrates the importance of a functioning team.

When Georgia was one month old, molecular genetic analysis confirmed 5 α -reductase deficiency. Standard practice involves early feminizing surgery and gonadectomy, followed by estrogen therapy from puberty. While the surgeons considered their technical intervention, our team of pediatric endocrinologists and psychologists recalled our previous experiences and the adult literature. Although we found ourselves oscillating between different hypotheses, we did not give up on the possibility of an

alternative procedure. It was of course important to consider how best to relieve the parents' anxieties. However, we were also able to remind ourselves that there was no evidence that such relief could be brought about by surgery. In any case, it could only be short-lived. We also invoked the question raised by DSD activists about the ethics of relieving parental anxiety via irrevocable cosmetic surgery on the child.

In a meeting with the parents, who, after the first meetings with the surgeons had begun to orient themselves towards a female assignment, we shared what we knew with them, but also our uncertainty. We knew first of all that surgery was unlikely to be one-off. We knew that some adults who had undergone standard management have expressed a great deal of pain and dissatisfaction with standard practice. We also knew that many adults continue to question the decision made on their behalf without their consent. We could not offer certainty to the family, but we could offer them support if they wished to delay surgery and await the impact of dihydrotestosterone treatment on the development of the external male genitalia. They asked for some time to consider the information. At a follow-up meeting, they spoke of an uncle who lived in their country of origin who, when considering their present level of information, has the same condition. We were told that this uncle had lived as female in infancy, but, during pubertal development, he masculinized and acquired his current male identity, even though he has severe hypospadias.

The parents decided to delay surgery and begin treatment with dihydrotestosterone. In the following 2 months, penile development ensued. The parents were pleased with their choice. Rather than feminizing surgery, the child received treatment to repair the hypospadias and turning of the labia majora to form a scrotum. The next step was to prepare a medical report to enable parents to ask the local authorities to register a male identity.

Also Mario's mother took the risk of raising her child with partial androgen insensitivity syndrome (PAIS) as a

boy. When she was asked, 'What will he feel when all the other children pee standing and he does not?', his mother replied, 'I don't know; we will sort something out!' Basically she reflected that when a child grows up surrounded by sincere and warm relationships, it is able to admit that something else within himself is missing and will not be forced to deny it during the rest of his life.

Conclusions

Full disclosure, closely intertwined with informed consent, needs to be an inclusive process, involving health professionals, parents of children with DSD or adults with DSD, from the beginning of the reality of DSD. In the ideal world, there would be a peer support forum that clients can access, because reassurance from people who have lived through the experience is something that health professionals cannot offer. Such an inclusive process can feel uneasy; the collision of scientific and emotional truths cannot be experienced as controllable and predictable. But, with a shared vision of the possibility of living well and authentically with DSD, the emerging integration of biological and psychological reality can awaken and open doors for all participants. Attempts to obliterate the reality of a body born different, without asking permission from its owner, reflects either an excess need to control life or an excess fear of uncertainty that is intrinsic to life. Successful obliteration protects us and our society from discomfort; discomfort that arises from what we have known all along, that maleness and femaleness have never been mutually exclusive. In shutting down possibilities, fear may be reduced, but also hope of thinking outside the box.

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