Parental Perception of Terminology of Disorders of Sex Development in Western Turkey
Running Head: Parental perception of DSD Terminology

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Abstract
Objective: “Disorders of sex development (DSD)” is a nomenclature proposed to defeat the discomfort of families and patients and has found worldwide usage. The aim of this study is addressing the perception and usage of terminology among the parents of DSD patients in a tertiary center in western Turkey.
Methods: The records of the DSD council between years 2008-2015 were reviewed retrospectively and data including details of the management process, patient characteristics and follow-up details were noted. Then inquiries reflecting parental perception about terminology were implemented.
Results: In total, 121 patients were evaluated in monthly meetings of DSD council and 79 inquiries were completed. About half of the families admitted knowing the terms; DSD, ambiguous genitalia, dubious genitals (translated from its proxy in Turkish), and intersex; however, only 2% preferred using DSD, 6% intersex, and 14% ambiguous genitalia. Fifty-two percent of the parents used a disease name in Latin (mostly hypospadias) addressing the disorder. Sixty-nine percent who were familiar with the name dubious genitals were diagnosed in the neonatal period. The department mostly involved in the management was related to the terms referring the disease.
Conclusion: Each country has its own social norms; therefore, local committees should be employed to develop proper terminology.
Keywords: disorders of sex development, intersex conditions, ambiguous genitalia, terminology

What is already known on this topic?
Few studies have been conducted to consider the perspective of families on the terminology of DSD. Despite the worldwide use among medical society, these studies failed to show acceptance among families. All of them were conducted in western countries.

What this study adds?
This is one of the largest studies questioning the parental perception of terminology about DSD. It both reflects worldwide debates and cultural problems in a Muslim country for being conducted in our country.

Introduction
Disorders of sex development (DSD) are “congenital conditions in which development of chromosomal, gonadal, or anatomic sex is atypical” (1). Assigning an appropriate name to this condition has always been controversial and the clinicians are not the only ones involved in the process. The confusing nature of the disease draws the attention of health professionals, sociologists, and activists. All these groups published plenty of papers just arguing about the nomenclature, however, few had the focus on patients’ families. Families’ perception of the disease affects their child in two ways; by affecting their decision-making and through the environment the child will grow-up in. In the era of patient-centered medicine, questioning families’ opinion is important and necessary in order to hold a responsible and an ethical management of the disease. The aim of this study was to address the perception and use of terminology among the parents of DSD patients in a tertiary center in western Turkey.
Material and Methods

Evaluation and management of DSD patients are held by a multidisciplinary team in our institution. Department mostly involved in the management changes according to the primary diagnosis of the patient (which is pediatric endocrinology in most cases) but each patient with DSD is discussed in the monthly joint meetings. Every critical decision influencing the management process is also taken in these meetings. Our core team consists of pediatric urology, pediatric endocrinology, child psychiatry, and genetics; however, adult endocrinology, psychiatry, pathology, neonatology, gynecology, and social workers are consulted when necessary.

After approval of institutional ethics committee (2016; decision number: 16-2.1/1), the records of the DSD council of our institution between years 2008-2015 were reviewed. Families who were presumably unaware of the terminology with regards to their child’s diagnosis (patients with Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome, severe hypospadias, bilateral undescended testes) were excluded from the study. Retrospective analysis of the data including details of the management process, demographics, patient history, and follow-up details was performed. Then parents were contacted to conduct inquiries focusing on the terminology the parents knew and tend to use were implemented (appendix 1) after informed consent. The questionnaire consisted of closed and short answer open questions concerning their knowledge and preference about the terms, their first contact with the disorder and details about the management. To understand their knowledge about the terms, families were first questioned about the terms they know regarding their child’s condition. The interviewer paid attention not to use any name and calling it “the disease” during the entire interview not to manipulate their answers. At the end of the interview, if they did not recall, they were told the commonly used terms in Turkey (DSD, intersex, ambiguous genitals, dubious genitals) and asked which ones they ever heard of. To evaluate which terms they were comfortable with, they were asked which names they use while talking to others such as their spouses, with their doctors, and with the relatives. They were also asked the term their doctors use and if different doctors use different names.

There were some shortcomings while translating the study to English. In Turkish, there is no term as an exact translation of intersex. Instead of it, çift cinsiyet is in use (which can be translated as double sex to English). As çift cinsiyet is used as a translation of intersex to Turkish, implementing a meaning close to a third sex, the word intersex will be used in the text for ease of reading. Another interesting term in Turkish is kuşkulu genital yapı which is a distorted translation of ambiguous genitals. It will be used with its exact translation which is dubious genitals in the text. Our perinatologists tend to use this term probably to avoid naming the exact diagnosis before consulting the DSD council. The term “Ambiguous genitals” is included in the study as a different heading because it is used in Turkey in Latin form without being translated and therefore generates a different perception. The families mostly use it as ambiguous solely without understanding the meaning. In daily Turkish language, most medical terms are used in Latin, French, or English, either in original or slightly corrupted forms. Therefore, unlike the parents in English-speaking countries, it probably appears as another disease name in Latin for them. Besides the evaluation of terminology, parents knew and tend to use, the results were analyzed to assess the effects of different parameters on the terminology families used. These parameters included primary diagnosis, age at diagnosis (as newborn or later), duration of follow-up (as less or more than 5 years), year of diagnosis (as before or after 2006- year of the Chicago consensus), appearance of external genitals, need for sex reassignment, need for name change, having a sibling with the disease, family history, history of admission to different hospitals, and the department mostly involved in the management.

To evaluate if the appearance of external genitals had an impact on families’ preference for the terminology, we divided patients into two groups as the ones that have genital atypicality and the ones that do not (2). It was defined as atypicality of genitals relative to the gender of rearing before reconstructive surgery. For patients reared as female, Prader Stage 0 (normal female) and Prader Stage 1 were considered typical; Prader Stage 5 and Prader Stage 6 (normal male) atypical. Likewise, for patients reared as males, Prader 5 and normal male were considered typical; normal female and Prader 1 atypical. Prader 2, 3 and 4 were grouped as atypical for both.
Statistical analysis was carried out using the SPSS statistical package (SPSS for windows V.16, SPSS, Chicago, IL, USA). To evaluate the effect of different variables on the terminology families used, comparisons were made using logistic regression analysis after transforming the data into dichotomous variables. *Hosmer-Lemeshow goodness of fit* test was used to assess model fit. A 5% type-I error level was used to infer statistical significance.

**Results**

In total 121 patients were evaluated in monthly meetings of DSD council in the study period. Twenty-five patients with diagnoses like MRKH, severe hypospadias, bilateral undescended testes (whose families were presumably not familiar with any of the DSD terminology) were excluded from the study. Among the rest, 9 parents could not be reached and 4 parents had two affected offspring both followed in our institution. Therefore, 79 inquiries were completed.

Median age at diagnosis was 1 year (0 -16 years) and 41% of the patients were diagnosed in the newborn period. Median follow-up was 5 years (1-19 years). Follow up period was longer than five years in 56%. Reason for admission at the time of diagnosis were atypical genital appearance in 55 (70%), delayed puberty in 12 (15%), inguinal hernia in 7 (8%), short stature in 3 (4%), symptoms of salt depletion in 1 (1%), and incidentally during the evaluation of a syndromic child in 1 (1%).

Seven patients (9%) had a history of sex reassignment and 6 of these also had their names changed. Four families had more than one affected children and 9 parents including these 4 had a family history. Forty-seven parents (41%) admitted to another center before referral to our institution. The majority of the parents (73%) pointed endocrinology as the department mostly involved in the management of their children. Fifty-six (71%) children had atypical external genitalia for the gender they were reared.

Sixty (75%) parents admitted to having enough knowledge about the disease and 27 (34%) parents expressed that their child knows what his/her disease was. It was noted that parents were comfortable while using the terms hypospadias or congenital adrenal hyperplasia (CAH); however, they avoided using the word “sex” during the questionnaire. An interesting observation was, some parents only said CAH when they were asked the names they know and used only CAH during the entire questionnaire. At the end, when they were asked about their knowledge of the remaining nomenclature, they first explained the pathology in the adrenal gland in detail and that the genital abnormality was secondary to it.

When asked about the terms they can recall about the disease, 40 parents said specific disease names mostly with Latin origin (which were hypospadias, adrenal insufficiency, testicular feminization, androgen insensitivity, congenital adrenal hyperplasia and 5-alpha reductase deficiency), 14 said chromosomal abnormality, 11 used the word ambiguous, 7 referred to the name of the syndrome the child has, 5 said intersex and only 2 parents mentioned DSD. Seven parents said that they do not know any (figure 1). The parents who referred the disease as a chromosomal abnormality were the parents of children who have chromosome-gender mismatch. One parent said an old Turkish word with Arabic origin (hünsâ; khunsa) which is not in daily use (3). When they were asked if they ever heard of the terms commonly used; 42 approved intersex, 40 DSD, 39 dubious genitals and 36 ambiguous (figure 2).

There were also questions from which we couldn’t collect any comparable data. Parents were asked which terms they use when talking to their spouse, to the doctor, and to relatives. Majority replied as they do not use any terms while talking to their spouse or with the doctor because everybody knows what the issue is. They also stated that they do not talk to their relatives or friends about the disease. They were also asked for any ideas for a new terminology but none of the parents made a suggestion.

When the state of knowledge about each term was evaluated with the independent parameters, statistically significant difference was revealed between the following pairs: the term “chromosomal abnormality” or “expression of a specific disease name” and the department mostly involved in the management; the term “dubious genitals” and the diagnosis in the newborn period.

Fourteen parents (24%) whose children were mainly followed by the endocrinology department stated the disease as a chromosomal abnormality while none of the parents did who were followed by pediatric urology (p=0.024). Expression of a specific disease name was also found relevant to the department mostly involved in
the management (p=0.048). Twenty three parents (41%) whose children were mainly followed by the endocrinology department used a specific disease name while 16 parents (80%) who were followed by pediatric urology.

Twenty-two of 32 parents whose children were diagnosed in the newborn period knew the term dubious genitals (69%) while 17 of 47 (36%) whose were diagnosed later (p=0.046). No statistically significant difference was found between the remaining parameters.

Discussion

Gender is one of the major aspects of personality. Construction of a scientific terminology about a disease that interferes with gender which is not pejorative but definitive is difficult. As Feder and Karzakis perfectly describe, there is probably no terminology that can eradicate the stigma and no nomenclature that can situate this condition into the usual way of medicine (4). Unfortunately, parents’ perception of the terminology has a direct impact on their perception of the disease which designates how they and their child cope with the disease.

Changing the terminology as DSD with the consensus statement in 2006 received widespread acceptance among the clinicians (5); however, its perception was not the same for everyone. Linguistic, religious and cultural factors influence how the lexicon is understood. One major criticism about DSD was the disturbing effect of the word “disorder” (6). Besides the worldwide debates around it, the Turkish equivalent of the word disorder in DSD is probably more disturbing than the English version like in German (7). It has a meaning closer to failure or defect than disorder. It also does not have a widespread use in the medical terminology. Not only naming, but also perception and management of DSD are prone to intercultural differences (8). Some cultures do not allocate sex at birth with the belief that it can change later; however, gender is the major determinant of a human’s entire life in many eastern countries. Islam has a comprehensive attitude towards DSD including prayers, obligations and gender roles in society (3). Turkey is a multicultural country where the majority of inhabitants have a social life influenced both by modern European society and the Islamic rules. In our country, any problem related to sex will cause shame, can hinder a marriage and even affect one’s work life; therefore, nomenclature of DSD gains more importance to prevent stigmatization. Our study confirmed the importance of this issue by revealing the parents’ tendency to avoid the word sex during the interview.

Doctors and activists play the main roles in constituting the terminology (1,4,6,9). The people (children with the disease and their families) who are in the center of the arguments are not a part of decision-making. Few studies have been conducted to consider the perspective of families (5,10–12).

Davies, who is one of the proponents of the new terminology, evaluated the acceptability of the new terminology among 19 parents of children with DSD. The majority stated to prefer DSD over intersex; however, few found it adequate (5). Lin-su et al. (11) interviewed a larger group (128 CAH patients, 408 parents) and stated that majority of the patients didn’t like the new terminology and that it caused negative connotations. An activist, Davis, made interviews with patients and clinicians and argued that the patients do not like the term and the doctors’ insistence on the DSD terminology was a reassertion of their medical authority (12,13). She says the patients who embrace the new terminology are the ones who are not happy with themselves and who find themselves abnormal (13). Ellie Magritte (10), the mother of a child with DSD, used the acronym DSD when referring to the disease writing both forms (disorder/difference of sex development) and emphasized how disturbing ambiguity was.

Our study was consistent with the earlier ones showing a lack of acceptance of the term DSD by the families despite the worldwide use of it among clinicians (14). Half of the families admitted knowing it and only 2 parents recalled it before being reminded by the interviewer.
Lin-Su (11) thought that health professionals did not use the term in their daily routine with their patients. This probably has an effect but even the families who admitted knowing it did not use the term. Most of the families in our study tended to use specific disease names mostly with Latin origin. This supports Karzakis’ ideas (15) who emphasizes the importance of recasting diverse diagnoses rather than keeping them as types of people whose care is directed at correcting sexual difference. Davies (5) and Dreger (16) also recommended temporary usage of the term DSD until specific diagnoses have been made.

There is no consensus on the terminology for DSD in Turkey. International Classification of Diseases (ICD-10) still refers the disease as hermaphroditism. Doctors use intersex or DSD while talking to each other, prefer to use çift cinsiyet while talking to media, and mostly avoid to use any term while communicating with their patients. They can also use specific disease names or some jargons like kuşkulu genital yapı. There is no common patient-oriented language. We have discussed and decided to use DSD in our multidisciplinary council; however, our study revealed probable lack of compliance with this decision and reflected the effect of doctors’ use of terminology on parents. When they were asked, they expressed that their doctors did not use any names for the disease; however, the department mostly involved in the management was a factor affecting their preferences.

Eighty percent of patients who named the disease as hypospadias were mainly followed by pediatric urology, and all the parents who referred the disease as a chromosomal abnormality were mainly followed by endocrinologists. This can be attributed to the need of fewer endocrinology consultations for the ones without chromosome to gender mismatch but also reflects the preferences of the doctors.

We believe, specific disease names with Latin origin provide acceptance of the subject as a medical problem, not a social one. We understand that this may not be desired by an adult with DSD; however, families’ perception and therefore attitude towards the disease designates the adult that child would be. None of the current terms are adequate and a terminology covering the will of both patients and families is yet to be developed. 

Another interesting finding of our study is the significant alteration about knowing the term dubious genitals (kuşkulu genital yapı) if they were diagnosed as a newborn. After referral to DSD council, families probably do not hear this term again; however, half of the families who were diagnosed in the newborn period recalled it. This not only shows the effect of doctors’ preferences but also emphasizes the persisting impact from their first contact with the disease.

Genital atypicality, sex of rearing, and gender reassignment process were reported to cause more stigmatization of parents (2). Therefore, the effect of these variables on parents’ choice of terminology was also analyzed; however, no relation could be identified. Unlike other studies (5), parents stated to be satisfied with their level of knowledge about the condition. This may be due to close communication with their doctors or less expectation due to cultural motivations. DSD is a hard-to-talk subject in our country. Long explanations of the DSD council given by each department individually may be more than enough for the families who have never heard of and willing to forget the subject.

**Study Limitations**

This study tried to evaluate the parental perception about the nomenclature of DSD; however, it was performed as a single center study in Western Turkey. Therefore, it may not reflect the opinion of all population. Also, there were semantic losses while translating the study to English. The authors tried to cover these shortcomings with a detailed methods section.

**Conclusion**

Introduced with the hope of defeating the discomfort of patients and families, the term DSD does not seem to find use among the parents of patients. Parents of our DSD patients avoid using any word containing “sex” and prefer the specific disease names mostly with Latin origin instead. Their preferences were also found to be influenced by their doctors. Each country has its own social norms; therefore, local studies reflecting the linguistic and cultural differences and their uniform usage by doctors are mandatory to avoid negative connotations in the families’ minds.
Conflict of Interest
The authors declare no conflict of interest.

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Author Contribution
All of the authors declare that they have all participated in the design, execution, and analysis of the paper and that they have approved the final version.

References
Appendix 1: Questionnaire

Name:      DOB:
Hospital ID#:     Phone Number:
Diagnosis:

1. When and how was your child diagnosed?
2. With what gender your child was raised before admitting? Was it changed? Was also the name changed?
3. When?
4. Do you have any other child affected? Anyone else in your family?
5. Which institutions were you referred to for your child’s condition? If you changed your doctor in the
   process, was it your choice?
6. Do you think you have enough knowledge about your child’s disease?
7. Does your child know about his/her condition?
8. Do you talk to your child about the condition?
9. There are many terms used to refer this condition. Which ones do you know?
10. Which department is the one most involved in your child’s management? Do you know the terms your
    doctors use?
11. Do different doctors use different terms?
12. Which term do you prefer to use when talking to your husband/wife?
13. Do you have a suggestion for a more proper term?
14. Did you ever heard of the terms I will say?

Legends for figures
Figure 1: The terms parents expressed (answer of question #8)

Figure 2: The terms parents were familiar with (answer of question #15)