

EDITORIAL

Disorders of Sexual Development - Still a Big Challenge!

It is always difficult to have a child born with ambiguous genitalia, not only for the family and relatives, but also for the medical team. Not all disorders of sex development come with ambiguous genitalia, but whenever ambiguity is present, it needs immediate action, to try to clarify the diagnosis and to decide the best choice for the sex of rearing.

In the last 30 years, our group has followed up more than 400 patients with disorders of sexual development (now the preferred term to 'intersex'), and we have witnessed the difficulties for the patient and for the family in supporting the many hardships imposed by the diagnostic work-up and the choice of sex of rearing.

As there is a period of time during which the diagnosis has not yet been made, the whole team caring for the patient and the family must be aware that the terminology employed has to be carefully chosen not to give any connotation of the sex of the infant, which may eventually be different from the sex of rearing chosen for that particular patient, when the final diagnosis has been reached. Thus, the use of terms such as 'gonads' instead of testis or ovary, 'internal ducts' instead of uterus, tubes, vas deferens, is mandatory and requires a lot of practice.

Discussion with the family about the findings in the patient and the possibilities involved in choosing male or female sex of rearing helps to clarify the many doubts so frequent in this situation. Some conditions are very clear and then the choice of the sex of rearing is relatively easy, while there are situations in which we do not have clear-cut options, and either male or female choice may pose problems for the child in the future.

Since the terminology used for patients with ambiguous genitalia dates back to 1876, proposed by Theodor Albrecht Edwin Klebs, it is nice to see a group of specialists come up with a new nomen-

clature on disorders of sex development (DSD), since terms such as 'intersex', 'pseudohermaphroditism (male or female)' and 'hermaphroditism' may give a confusing connotation to the patient's family². Although a wonderful job has been done, I would like to disagree with certain options taken by the authors. To start with, to put the karyotype in the name of the disorder is not adequate, since it gives a connotation of the sex. Everybody knows what 46,XX or 46,XY means.

With regard to the terminology for certain diseases, such as 'ovotesticular DSD' for true hermaphroditism and '46,XX testicular DSD' for XX male, also seems inadequate. We all know that many patients with true hermaphroditism are reared as females, and it is weird to see a girl with a condition called ovotesticular DSD. It is similar to the situation of 'feminizing testes', used before it was known that this syndrome resulted from androgen receptor resistance.

As the majority of XX males go into male sex of rearing, '46,XX testicular DSD' would not be so complicated, although to have 46,XX in the name of the disease for a child who will be reared as a male is confusing. On the other hand, if we remember that 20% of these patients have ambiguous genitalia and some may be reared as females, again the name of the disease becomes inadequate for the social reality of the patient. Maybe 'reversal DSD' would be a better option, since it does not include the karyotype in the name and does not give the name of the gonad, leaving either option, male or female, not stigmatized by the name of the condition.

Taking into consideration the great effort made in trying to improve the way we talk about problems of sexual development, to make minor modifications in the proposed nomenclature could help to attain one of the main objectives of the Consensus of Chicago, namely, to avoid stigmatiza-

tion of patients already suffering the heavy setback of not having the sex determined at first, at the moment of birth.

*Durval Damiani, M.D., Ph.D.
Pediatric Endocrinology Unit
São Paulo University Medical School
Brazil*

REFERENCES

1. Dreger AD, Chase C, Sousa A, Gruppuso PA, Frader J. Changing the nomenclature/taxonomy for intersex: a scientific and clinical rationale. *J Pediatr Endocrinol Metab* 2005; 18: 729-733.
2. Hughes IA, Houk C, Ahmed SF, Lee PA; LWPES Consensus Group; ESPE Consensus Group. Consensus statement on management of intersex disorders. *Arch Dis Child* 2006; 91: 554-562.