Rev Int Androl. 2013;11(3):100–106

Revista Internacional de Andrología

www.elsevier.es/andrologia

REVIEW

Disorders of sex development

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Received 11 January 2013; accepted 26 February 2013
Available online 17 June 2013

KEYWORDS
Intersex;
Ambigus genitalia;
Disorders of sex development

Abstract
Background: Disorders of sex development (DSD), which has the possibility of the risk of life-threatening endocrinologic emergencies of the newborn, require a careful multidisciplinary approach.
Objectives: The aim of our study is to consolidate the proposed classification, evaluation and management of DSD.
Materials and methods: The literatures related with DSD were reviewed to find the best approach for this disease.
Results: The detailed history, systemic physical examination of the patient, particular laboratory and imagine evaluations are needed for the urgent treatment of life-threatening abnormalities and the gender assignment.
Conclusion: The gender should be assigned depending on the definitive diagnosis, fertility potential, genital appearance, surgical options, and the parents' opinion.

PALABRAS CLAVE
Intersexualidad;
Ambigüedad genital;
Trastornos del desarrollo sexual

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Resumen
Antecedentes: Los trastornos del desarrollo sexual (TDS), que tienen la posibilidad de poner en riesgo vital las emergencias endocrinológicas del recién nacido, precisan un cuidadoso enfoque multidisciplinar.
Objetivos: El objetivo de nuestro estudio es consolidar la propuesta de clasificación, evaluación y tratamiento de los TDS.

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1698-031X/$ - see front matter
http://dx.doi.org/10.1016/j.androl.2013.02.006

Materiales y métodos: Se revisaron las literaturas relativas a TDS, a fin de hallar el mejor enfoque para esta enfermedad.
Resultados: Se precisan la historia clínica detallada, la exploración física sistémica del paciente, el laboratorio concreto y las evaluaciones de imágenes para tratar urgentemente las anormalidades con riesgo vital y la asignación de género.
Conclusión: Deberá asignarse el género dependiendo del diagnóstico definitivo, el potencial de fertilidad, el aspecto de los genitales, las opciones quirúrgicas y la opinión de los padres.

Introduction

Sex development disorder occurs in 1–2 of every 10,000 newborns.[1] Ambiguous genitalia are a serious concern for the family and the first physician examining the child. This is due to not only ambiguous genitalia, but also the risk of life-threatening endocrinologic emergencies of the newborn. Therefore, ambiguous genitalia require a careful multidisciplinary approach. Evaluation and management should be performed in a center with a multidisciplinary team, comprised of pediatric specialists in surgery, typically in urology, endocrinology, neonatology, nursing, psychology, genetics, and medical ethics.[1–5]

Objectives

Designations such as hermaphroditism or pseudohermaphroditism, which are previous terminologies, are confusing to patients and their relatives, and also constitute a humiliating situation. Therefore, the term ''disorders of sex development'' (DSD) is proposed to indicate congenital conditions with atypical development of chromosomal, gonadal, or anatomic sex. This terminology should be well-grounded, open to new information, and explain the genetic etiology and phenotypic changes, comprehensible by the physician, patient, and the parents.

This study was designed to consolidate the proposed new classification, and to facilitate the evaluation and the management of DSD.

Materials and methods

The literatures related with DSD were reviewed to find the best approach for this disease. An extensive research was in the Pubmed. The MeSH terms, such as ''hermaphroditism'', ''pseudohermaphroditism'', ''disorders of sex development'', ''congenital adrenal hyperplasia'' and ''androgen insensitivity syndrome'', were used in research of the articles without the limits of publication date.

Results

In 2006, under the sponsorship of the European Society for Pediatric Endocrinology (ESPE) and the Lawson Wilkins Pediatric Endocrine Society (LWPES), with the participation of 50 experts in Chicago, the DSD results were evaluated and published as a consensus.[6] Afterwards, the DSD consensus was reviewed with the current practical applications of 60 centers from 23 European countries and the following topics were emphasized: the necessity of the evaluation of patients by skilled professionals, avoidance of gender assignment in the newborn without the assessment of an expert, the need for a multidisciplinary team, psychosocial support for the patients, surgical procedures performed by the surgeons specialized in this field, assessment of the functional and the cosmetic surgical procedures all together, and more accurate information for the parents and the patients, which will lead to a definitive final decision. In order to define the disease more accurately and clearly, today, new nomenclature and classification are used (Tables 1 and 2).

Normal sexual development

The male and female embryos are phenotypically identical until the sixth gestational week in the normal conditions. The gonads are bipotential at first, forming as a mesodermal thickening in the urogenital ridge. There are both Müllerian and Wolffian ducts. In the presence of 46, XY karyotype bipotential gonad differentiates into testis. If the karyotype is 46, XX the differentiation progresses in the favor of

Table 1 Old and new nomenclatures.

Previous	Revised
İntersex	Disorder of sex development (DSD)
Male pseudohermaphrodite	46, XY DSD
Undervirilization of an XY male	
Undermasculinization of an XY male	
Female pseudohermaphrodite	46, XX DSD
XX female with overvirilization	
XX female with Masculinization	
True hermaphrodite	Ovotesticular DSD
XX male or XX sex reversal	46, XX testicular DSD
XY sex reversal	46, XY complete gonadal dysgenesis