Confusion in gonadal dysgenesis terminology persisting beyond Chicago Consensus Statement on disorders of sexual development

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Abbreviations:

DSD: Disorder(difference) in sexual development

SO: Streak ovary

DT: Dysgenetic testis

GD: Gonadal dysgenesis

OT-DSD: Ovotesticular disorder(difference) in sexual development

A consensus on DSD classification and terminology was published in 2006 under the auspices of the Lawson Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology. Terminology in this group of disorders has social, psychologic and even political and philosophical intricacies, and the medical community should be cautious about any trace of stigmatization or social unfairness. Preceding literature has adopted a confusing approach toward the terminology of gonadal dysgenesis with inconsistent emphasis on either gonadal histology or karyotype. Descriptive words "partial," "complete," and "pure" have been used diversely and sometimes interchangeably.
We try to show some inconsistencies below in enumeration:
1) The phrase "partial gonadal dysgenesis" is used as a discriminative term vs mixed gonadal dysgenesis. Since MGD denotes a streak gonad on one side and a dysgenetic testis on the other, the term "partial gonadal dysgenesis" has been used to denote a condition of bearing dysgenetic testes on both sides.

2) A condition of two streak gonads with XX karyotype and without somatic features of Turner
syndrome is called 46XX pure gonadal dysgenesis. The word "pure" is presumed to convey a non-mosaic karyotype (46XX). This is somewhat obscure as there are cases of two streak gonads and 45X/46XY karyotype that are mosaic and evade the quoted applicability of the word "pure". This condition is not covered by the 2006 Chicago statement (1).

3) Complete gonadal dysgenesis has been used to denote cases of bilateral streak ovaries and hypergonadotropic hypogonadism (2). Specifically, Swyer syndrome with 46XY and bilateral streak ovaries has been called 46XY-CGD in the 2006 Consensus Statement (3). Confusingly, cases of pure gonadal dysgenesis have also been called CGD in some essays, sparing the DSD Consensus Statement (2, 4). There are cases of 45X/46XY bilateral SO that have been proposed to be called CGD (4) and 46XY-Pure gonadal dysgenesis in some dissertations (2). Although the DSD Statement attempted to solve the confusion by attributing any terminology to a specific situation, the words partial, complete, and pure are still non-descriptive and difficult to understand regarding the assigned condition.

One does not find a clear explanation why the word "partial" is presumed to represent two dysgenetic testes and the word "complete", two streak ovaries.

It is clear that much confusion remained in the 2006 Consensus about the terminology of gonadal dysgenesis conditions as an important subclass of DSD. Confusion in terminology appears to be due to uncertainty in shifting emphasis between karyotype and gonadal histology in each disorder.

Any proceedings in terminology must be scientifically well sought and bear significant consequences. Such decisions must be adopted and coined by multidisciplinary conventions.

A terminology in any of gonadal dysgenesis conditions should convey these specifications:

a) It is one of the gonadal dysgenesis subtypes of DSD.
b) Types of dysgenetic gonads: streak ovaries, dysgenetic testes, ovotestis, or undifferentiated
c) Karyotype: It must be mentioned if specifically addressed in the definition of the condition, e.g. Swyer syndrome. If the condition is not specifically attributed to a non-

mosaic karyotype, it is adequate to declare it as a type of gonadal dysgenesis; this simply denotes that the karyotype may have any combination.

We attempted to propose a more descriptive blueprint terminology in Table 1. We aimed to demonstrate this study's message in a clearer exemplary fashion. There is no doubt that a newer statement on DSD is necessary after fifteen years.

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Table 1. Confusion in current terminology and proposal of a blueprint

	Condition	Proposed terminology	Traditional or frequently cited		Consensus 2006(1)
1	GD:SO-DT	Asymmetrical gonadal dysgenesis	Mixed gonadal dysgenesis		MGD
2	46 XX-bilateral DT	46XX- symmetrical testicular GD	46-XX sex reversal	De la Chapelle	46-XX testicular DSD
3	46XX-bilateral SO	46 XX symmetrical ovarian GD	46XX-pure gonadal dysgenesis (2)	46-XX complete gonadal dysgenesis (2, 4)	46XX-pure gonadal dysgenesis
4	XO/XY-bilateral SO	Mosaic (XO/XY)Symmetrical ovarian gonadal dysgenesis	Complete gonadal dysgenesis (2)		Not found
5	GD: bilateral DT	Symmetrical testicular gonadal dysgenesis	Partial gonadal dysgenesis	Dysgenetic male Pseudohermaphroditism	Partial gonadal dysgenesis
6	46XY bilateral SO	46 XY symmetrical ovarian gonadal dysgenesis	46XY-sex reversal or XX female	Swyer syndrome	46-XY complete gonadal dysgenesis
7		OT-DSD	True hermaphrodite		OT-DSD