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Changing the Nomenclature/Taxonomy for Intersex: A Scientific and Clinical Rationale

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ABSTRACT

We explain here why the standard division of many intersex types into true hermaphroditism, male pseudohermaphroditism, and female pseudohermaphroditism is scientifically specious and clinically problematic. First we provide the history of this tripartite taxonomy and note how the taxonomy predates and largely ignores the modern sciences of genetics and endocrinology. We then note the numerous ways that the existing taxonomy confuses and sometimes harms clinicians, researchers, patients, and parents. Finally, we make six specific suggestions regarding what a replacement taxonomy and nomenclature for intersex should do and not do, and we call for the abandonment of all terms based on the root "hermaphrodite".

KEY WORDS

intersex, hermaphrodite, hermaphroditism, true hermaphroditism, pseudohermaphroditism, nomenclature, taxonomy, ambiguous genitalia, disorders of sexual differentiation

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INTRODUCTION

We present scientific and clinical problems associated with the language used in the existing division of intersex types, in order to stimulate interest in developing a replacement taxonomy for intersex conditions. The current tripartite *division* of intersex types, based on gonadal tissue, is illogical, outdated, and harmful. A *new* typology, based on phenotypic presentation, as well as karyotype, gonadal histology, and etiology, could improve diagnosis and management.

The present taxonomy for congenital sexual anatomies divides humans into five types:

1. *Females:* defined as presenting only standard female sexual anatomy.
2. *Males:* defined as presenting only standard male sexual anatomy.
3. *Female pseudohermaphrodites:* defined as presenting some mixture or blurring of standard female and male sexual anatomy with the presence of ovaries (and not testes or ovotestes) and of an 'XX' chromosomal complement.
4. *Male pseudohermaphrodites:* defined as presenting some mixture or blurring of standard female and male sexual anatomy with the presence of testes (and not ovaries or ovotestes) and of an 'XY' chromosomal complement.
5. *True hermaphrodites:* defined as presenting at least one ovary *and* at least one testis, or at least one ovotestis. (The definition of true hermaphroditism does not depend on the presentation of other sexual anatomy or the chromosomal complement.)

Textbooks and clinicians today use this taxonomic system, developed approximately 125 years ago, before the development of modern genetics and endocrinology, and well before the current diagnostic techniques and scientific knowledge of sexual anatomy. The system outlined above was first presented by Theodor Albrecht Edwin Klebs in 1876, in the *Handbuch der Pathologischen Anatomie*[1]. Although Klebs did not recognize the existence of ovotestes or chromosomes, he was the first to sort hermaphroditisms primarily according to the nature of the gonads. Klebs divided hermaphroditisms into three basic types: female and male pseudohermaphroditism, defined as above but without the chromosomal criteria, and true hermaphroditism, defined as presenting with at least one ovary *and* at least one testis.

Klebs' system was widely accepted and disseminated among the biomedical professions after George F. Blacker and Thomas William Pelham Lawrence published their text on the subject in the *Transactions of the Obstetrical Society of London* in 1896[2]. Blacker and Lawrence, recognizing the existence of ovotestes and critically reviewing the record of supposed cases of true hermaphroditism, endorsed Klebs' basic division into the five anatomical types.

Clinicians thereafter rallied around this gonadal division. They came to sort all patients into categories according to gonadal anatomy, regardless of their phenotypic presentation and regardless of the functionality of their gonads.

THE CONTINUED PRIVILEGED ROLE OF GONADAL ANATOMY IN THE EXISTING SYSTEM

One might presume that the present system, as outlined earlier, differs from Klebs' or Blacker's and Lawrence's systems, as it seems to recognize, at least in the cases of female and male pseudohermaphroditisms, the importance of chromosomes. However, the discovery and understanding of 'sex chromosomes' did not substantively alter the earlier taxonomic systems. Indeed, the existing division does not depend on chromosomes, because it still defines 'true' hermaphroditism as the presence of ovaries and testes, or ovotestes, regardless of chromosomal complement.

THE EXISTING SYSTEM IS NOT BASED ON 'NATURAL KINDS'; ALTERNATIVES ARE AVAILABLE

The current system, like Klebs', is based primarily on the anatomy of the gonads. Even before and for 20 years after Klebs' taxonomy, scientists and clinicians offered alternative taxonomies which focused on the degree and type of mixture of male and female parts. For example, Isidore Geoffroy Saint-Hilaire divided hermaphrodites into sexually ambiguous people with extra parts and sexually ambiguous people with the usual number of parts[3]. These alternative systems did not favor the use of gonadal anatomy above all else for demarcation purposes as Klebs did.

In other words - and this point is critical - the current taxonomy does *not* represent a division into what philosophers of science call 'natural kinds'. Nature does *not* tell us the existing system is the one and only way to view sexual anatomies. Instead, the existing, quite arbitrary system was developed and adopted for *pragmatic* reasons. It seemed to sort confusing and sometimes disturbing anatomies into clear types, and thereby seemed to make sense of confusing presentations[4]. It gave a central position to gonadal histology, at the time a fashionable (though vague) explanation for gender and sexual behavior. The existing taxonomic system may once have been useful, but it does not represent the only, the most logical, or even the most natural taxonomy.

WHY THE EXISTING SYSTEM NEEDS TO BE REPLACED

A system that emphasizes gonadal anatomy above all else suffers from two major deficits. First, it is scientifically questionable, because it relies on the anatomy of the gonads (functioning or not) more than any other considerations. Second, it provides little clinical help, often confusing and harming the patient, and sometimes also the physician.

SCIENTIFIC PROBLEMS WITH THE EXISTING SYSTEM

Why has such an outdated system not been discarded and replaced? The scientific understanding of sexual development has progressed tremendously in the last 125 years, but the existing taxonomy does not reflect that progress. Scientists and clinicians now recognize that the structure of the gonads does not correlate simply with genotype, phenotype, physiology, diagnosis, or gender identity. The anatomy of testicular tissue in women with androgen insensitivity syndrome (AIS) is quite similar to the anatomy of testicular tissue in non-intersex males, yet their physiologies, phenotypes and gender identities differ markedly. Many people with so-called true hermaphroditism have ovotestes, yet their genotypes, phenotypes, physiologies, and gender identities vary considerably more than their single categorization implies. To continue to use rhetoric suggesting that gonadal anatomy is the most important marker or is a simple marker of sex type denies the full breadth of our current scientific knowledge.

In fact, many more physiologically-specific diagnoses of intersex conditions have been developed since Klebs' system was proposed 125 years ago. 'AIS' is a more scientifically specific and useful diagnosis than 'male pseudohermaphroditism', the blanket term into which AIS is still fitted. 'Gonadal dysgenesis' is a scientifically specific and useful intersex diagnosis that does not even fit into the five-item categorization scheme in current use.

When researchers and clinicians use the broad categories first described by Klebs, instead of more specific diagnoses, such as AIS and gonadal dysgenesis, accurate research and literature searching become much more difficult. This is not a minor issue. If researchers want to conduct sound studies and clinicians hope to use evidence generated by such research, the nomenclature must allow sorting patients into diagnostically meaningful groups; the present system simply does not achieve this end.

CLINICAL SHORTCOMINGS OF THE EXISTING SYSTEM

When they developed and disseminated it, 19th century thinkers apparently expected the taxonomy of Klebs to function clinically and socially, as well as scientifically. They assumed that sexually ambiguous patients with testes ('male pseudohermaphrodites') could be scientifically, clinically, and even socially labeled male. Indeed, the system was designed to clear up social problems caused by sexual ambiguity through offering a method to separate males from females according to gonadal anatomy. However, modern diagnostic techniques and understanding of conditions such as AIS show that, for example, in AIS, a woman with unremarkably feminine appearance could have testes.

As early as 1915, clinicians began to recognize that Klebs' taxonomic system sometimes *caused* clinical and social problems, rather than solving them. In increasing numbers of cases, the sex assignment suggested by the gonadal taxonomic system conflicted with the external phenotype and the social gender assignment. Clinicians then began an awkward habit of trying to categorize patients according to two, often conflicting, systems: based on the patient's gonads, they would categorize the patient in Klebs' 'sex' system; based on the patient's phenotype, they would categorize the patient in a gender system. Often a patient wound up 'scientifically' labeled a 'male pseudohermaphrodite' but socially labeled 'girl' or 'woman', or vice versa.

This conflict of labels - a conflict caused at least in part by the gonadal taxonomy - put clinicians in an awkward position. Many realized that differences between supposed 'sex' (male pseudohermaphroditism, female pseudohermaphroditism, or true hermaphroditism) and gender could cause distress for patients. Some clinicians tried to avoid the conflicts by not revealing to patients their true diagnoses. In a practice now widely criticized, they withheld diagnostic information or actually deceived patients in the belief that patients would be harmed by knowing of the conflict of labels[5]. Others believed that early cosmetic surgeries were necessary to 'resolve' the conflict, to bring the anatomical sex as much in line with the gender assignment as possible. This led to many early-

childhood surgeries that patients (and then many clinicians) later regretted[6].

Despite the fact that the 5-sex taxonomy is not based on natural kinds, clinical practice has often followed it, creating conflicts and confusion that may produce unintended ill effects. The mismatch between taxonomy and modern science and the clinical misdirection that flows from continued use of the outmoded taxonomy strongly suggest the need to develop an accurate classification system for intersex that avoids labeling conflicts and permits clear and sound communication with families and patients.

ADDITIONAL RHETORICAL PROBLEMS WITH THE EXISTING SYSTEM

First, the rhetoric of the existing system centers around the term 'hermaphrodite', a term which originally signified - and still signifies to many people - a person with two full sets of genitals and sex organs, male and female. This continued reference to what only exists in mythology frightens and confuses many non-professionals, including patients and their families. In addition, it attracts the interest of a large number of people whose interest is based on a sexual fetish and people who suffer from delusions about their own medical histories. This unwanted attention can rise to a level that interferes with the work of support groups and clinicians.

Second, the system labels people as male or female simply based on gonadal anatomy. This distresses many patients who hear science labeling them as a sexual type with which they do not otherwise identify. One of the authors (AS) has had the experience of having to calm an adult patient after an internal medicine resident announced to her that she was 'really' a man, because he had found testes in the patient. What use is there in calling a woman with AIS a 'male', when her external phenotype and her gender identity are female? Neither patients nor clinicians benefit when the clinician has to try to explain to a woman with AIS, "You are a male pseudohermaphrodite but you're really a woman." The implications of nomenclature do not end with the pediatric endocrinologist's or urologist's diagnosis. Most patients with intersex conditions are confronted with social and sexual issues at many developmental stages over the course of their lives; a patient's understanding of her condition will be strongly affected by labels she encounters in her own medical record or in medical journals and texts.

Third, the use of a Klebsian system has typically led to the labeling of the whole person according to the condition ('male pseudohermaphrodite'), rather than naming a condition a person has ('male pseudohermaphroditism'). This subtle but critical distinction is similar to the difference between labeling a person a hemophiliac and labeling him a person with hemophilia. The former reduces the person to the condition; the later recognizes the condition as but one aspect of the person.

Finally, the division into 'pseudo-' and 'true' forms of hermaphroditism implies a hierarchy of authenticity, whereby one person has a fake form of intersex and another a real form. This pseudo/true division is a hangover from the gonadal demarcation system. However, a technical 'true hermaphrodite' may present far less ambiguity than a technical 'pseudohermaphrodite'. It is unhelpful and harmful to patients to use loaded prefixes 'pseudo-' and 'true'. The same problem pertains with terms like 'sex reversal', which implies an original 'true' sex, and can confuse physicians, parents, and patients alike.

TOWARDS A NEW TAXONOMY

The clinical approach to intersex should aim to use methodologically sound evidence to facilitate the development of healthy and happy patients. Continued use of the existing system for dividing intersex types ignores modern science and, as currently used, inadvertently undermines the central goals of the clinical treatment of intersex. A new system is needed.

What should the new system look like? The answer to this question will depend on what patients and clinicians decide they need. If physicians feel that karyotypes help guide the diagnostic work-up, then chromosomal analysis might form the basis for taxonomical division; unfortunately, karyotyping has the same tendency to confuse physicians and patients, many of whom do not

understand the relationship between 'sex chromosomes' and clinical phenotype and gender identity.

We propose that clinicians should work to articulate their needs and to develop, using scientific evidence and patients' experiences and advice, a taxonomic system which specifically and effectively addresses those needs.

As a starting point, we suggest that the replacement taxonomic system:

- *should* enhance, not complicate, the use of medical informatics in research and clinical practice;
- *should* recognize that diagnosis and taxonomy inform, but do not determine, gender assignment and/or gender identity (thus, should avoid the words 'male' and 'female');
- should *not* include the words 'hermaphrodite', 'hermaphroditism', 'sex reversal', or other easily misunderstood terms;
- *should* label the condition rather than the person;
- should *not* confuse physicians and patients;
- *should* make clear that diagnosis does not simply dictate therapy.

In conclusion, we suggest the language of 'hermaphroditism' and 'pseudohermaphroditism' be abandoned. One possible alternative to the procrustean 5-sex approach is to use instead specific etiology-based diagnoses (such as AIS, 5α-reductase deficiency, etc.) and the umbrella term "disorders of sexual differentiation". Such an approach would have the salutary effects of improving patient and physician understanding and reducing the biases that are inherent in the use of the current language of 'hermaphroditism'.

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