**Diagnostic approach to expanding aetiologies of DSD**

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Disorders or differences of sexual development (DSD) represent an umbrella term for a wide and diverse range of conditions. Thus, DSDs present with diverse clinical features and pathophysiology. When including all changes, the birth prevalence of individuals born with atypical genitalia might be as high as 1 in 300 live births. However, the prevalence of DSD conditions requiring further expert examination based on genital ambiguity has been estimated to be approximately around 1 in 5,000 live births. The diversity of all conditions classified as DSD, however, requires highly specialized and personalized care provision for individuals when medical health care provision is required and desired. However, the differential diagnosis of DSDs remains highly complex and challenging. This diagnostic pathway requires an integrated strategy consisting of thorough clinical, hormonal and genetic workups. Key to optimizing diagnostic processes and hopefully outcomes is the standardized and structured clinical assessment and the development of a diagnostic and management plan covering all aspects. The technological revolution towards genomic medicine appears to result in a complete overhaul of the diagnostic pathway. The traditional more stratified clinical pathway from clinical phenotyping via biochemical analysis towards stratified genetic analysis is likely to be replaced by a multidisciplinary approach in which the information on clinical phenotyping is considered in parallel with the biochemical data and genetic results in an integrative manner. Overall, this presentation will discuss the clinical sensitivity and usefulness of established test and provide an update on recent developments in the diagnostic approach to DSD.