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Rare diagnosis in disorders/differences of sex development

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DSD (Disorders/Differences of Sex Development) comprise a rare group of diseases and abnormalities that can present on different ages and have very variable presenting symptoms. Prenatally discrepancy between genotypic sex and ultrasound phenotype or an abnormal genital as seen by ultrasound. Neonatal or pediatric symptoms can be a genuine ambiguous genital, inguinal (ovotestes) in a girl detected during inguinal hernia surgery, small stature in a 45,X/46 XY girl etc. Others are only detected at puberty or when trying to get pregnant.There is consensus that patients should be evaluated by multidisciplinary teams and that it is important to involve patients or caretakers in the process of diagnosis and management. Psychological, socio-cultural and economic-organizational aspects play an important role.Within the group of diagnosis that fall under the term DSD, there are well-known syndromes as Turner syndrome and variants, CAH, or proximal hypospadias, which can have many causes and can present as ambiguous genitalia. Other causes are very rare and may not be easily recognized. Some of these will be presented here. The experience and organization of our team and DSD care in the Netherlands and the place and timing of NGS based diagnostics will be discussed. For the future it is important to realize the importance of choosing the right words to communicate about the condition, especially also for professionals who have little or no experience with DSD but who are frequently the first to see the patient. Transition from prenatal care to postnatal professionals, from a peripheral clinic to a specialized clinic or from adolescent to adult specialists often offers room for improvement. We should not neglect that in these exciting times of growing diagnostic possibilities.

Recent Publications

1. Van Bever Y, Wolffenbuttel KP, Brüggenwirth HT, Blom E, de Klein A, Eussen BHJ, van der Windt F, Hannema SE, Dessens AB, Dorssers LCJ, Biermann K, Hersmus R, de Rijke YB, Looijenga LHJ. Multiparameter Investigation of a 46,XX/46,XY Tetragametic Chimeric Phenotypical Male Patient with Bilateral Scrotal Ovotestes and Ovulatory Activity. Sex Dev. 2018;12(1-3):145-154.

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3. Brosens E, Marsch F, de Jong EM, Zaveri HP, Hilger AC, Choinitzki VG, Hölscher A, Hoffmann P, Herms S, Boemers TM, Ure BM, Lacher M, Ludwig M, Eussen BH, van der Helm RM, Douben H, Van Opstal D, Wijnen RM, Beverloo HB, van Bever Y, Brooks AS, IJsselstijn H, Scott DA, Schumacher J, Tibboel D, Reutter H, de Klein A. Copy number variations in 375 patients with oesophageal atresia and/or tracheoesophageal fistula. Eur J Hum Genet. 2016 Dec;24(12):1715-1723.

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Biography

Yolande van Bever is a clinical geneticist with expertise in congenital anomalies and syndromes and with a special interest in the field of DSD. She worked in various academic hospitals in the Netherlands and abroad. Since 2004 she is an active team member not only in the clinical genetic staf, but also in various multidisciplinary teams such as the follow up team on surgical congenital anomalies, the neurofibromatosis team and the DSD team. In view of the rapid growing diagnostic genetic possibilities it is important to focus on communication with patients and their caretakers. .

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