THE “EXTERNAL GENITALIA SCORE”
A EUROPEAN MULTICENTER VALIDATION STUDY

S. van der Straaten1, A. Springer2, D. Hebenstreit3, A. Zecic1, U. Tonrhofer2, A. Gawlik3, M. Baumert4, K. Szelliga2, S. Debulpaep1, A. Desloovere1, L. Tack1, K. Smets1, M. Wasniewska5, D. Coric6, M. Calafiore7, M. Lindhardt Lubicic8, A. Siegfried Busch1, A. Juul9, A. Nordenström9, J. Sigurdsson10, C. E. Fluck1, T. Haamberg11, S. E. Hannema11, K. P. Wiffenbutter11, S. Faisal Ahmed11, M. Cools1
University Hospital Ghent, Dept. of Pediatrics, Div. of Pediatric Endocrinology and Dep. of Neonatology1, Medical University of Vienna, Dept. of Pediatric Surgery2, Medical University of Silesia, Katowice, Dept. of Pediatrics and Neonatology and Pediatric Endocrinology3, University of Messina, Dep. of Human Pathology of Adulthood and Childhood4, Copenhagen University Hospital, Dep. of Growth and Reproduction5, Karolinska University Hospital Stockholm, Dep. of Women’s and Children’s Health and Department of Pediatric Endocrinology6, University Children’s Hospital of Bern, Dep. of Pediatrics, Division of Pediatric Endocrinology and Diabetology7, Erasmus MC Sophia Children’s Hospital, Dept. of Pediatric Endocrinology and Pediatric Urology, Rotterdam, University Hospital of Glasgow, Dep. of Pediatric Endocrinology, COST Action BM1303: “A systematic elucidation of differences of sex development”

INTRODUCTION
For the initial work-up of a child with DSD a detailed description of the external genitalia should be recorded. Until now the Prader Scale (PS) and External Masculinization Score (EMS) are used. The “External Masculinization Score” (EMS) is an objective method to describe undervirilized genitalia in male infants. A standardized genital assessment that captures the appearance of the genitalia across the phenotypic spectrum from female to male is needed at diagnosis and during follow up and can be used in established international registries enabling clinical research studies. The “external genitalia score” (EGS) was developed in group 1 of COST Action BM1303. The ano-genital distance (AGD) is a sensitive index of androgen action during fetal development.

OBJECTIVES
Validation of the External Genitalia Score. Presentation of normal values for premature and term infants, babies up to 2 years and in babies with atypical genitalia.

METHODS
EGS was compared to Prader Score and EMS. Ano-genital distances were measured using the TIDES protocol and video training. Intra- and inter-observer variability of the different scoring systems and AGD’s were studied in infants with typical and atypical genitalia. Repeat measurements for each distance were done. In a multicenter validation study, cross-sectional reference data were obtained.

RESULTS
Inter-observer reproducibility of EGS in typical and atypical genitalia is excellent, being 1 and 0.98 respectively (95%RI 0.97-0.99).

The ano-genital ratio in male and females followed a normal distribution. The ano-genital ratio does not correlate with anthropometric variables or gestational age. AGDratio as/ap in 46,XY DSD correlates positively with the EGS (Spearman’s r=0.19, p<0.05) AGDratio as/ap in 46,XY DSD correlates positively with the EGS (Spearman’s r=0.47, p<0.05) AGDs/ap in 46,XY typical genitalia is significantly different from AGDs/ap in 46,XY atypical genitalia (t=-3.9, p<0.05; CI 0.03-0.08).

CONCLUSIONS
The EGS can describe the phenotypic spectrum from female too male in premature and term infants up to 24 months. The AGD-ratio correlates positively with EGS in male infants.

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REFERENCES
1. The role of the clinical score in the assessment of ambiguous genitalia. SF Ahmed. 2000
2. Anogenital distance and penile width measurements in the Infant Development Environmental Study (TIDES), Methods and predictors. Shealet Sathyarayanara et al. 2015