



The time of first presentation at the department of paediatric endocrinology of patients with 46,XY DSD.

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Background

The atypical appearance of the external genitalia in a neonate, defined as the external masculinization score (EMS) less than 11, should incline the clinicians to perform diagnostic procedure ideally managed by a multidisciplinary team in a tertiary centre. Among the patients with disorders of sex development (DSD), the most challenging subgroup in terms of aetiology, is the subgroup with 46,XY karyotype.

Objective and Hypotheses

To study the time of first presentation at the department of paediatric endocrinology of 46,XY DSD patients with atypical genitalia after birth and to assess whether there is a correlation between the time of first presentation and the EMS in those raised as males.

Methods

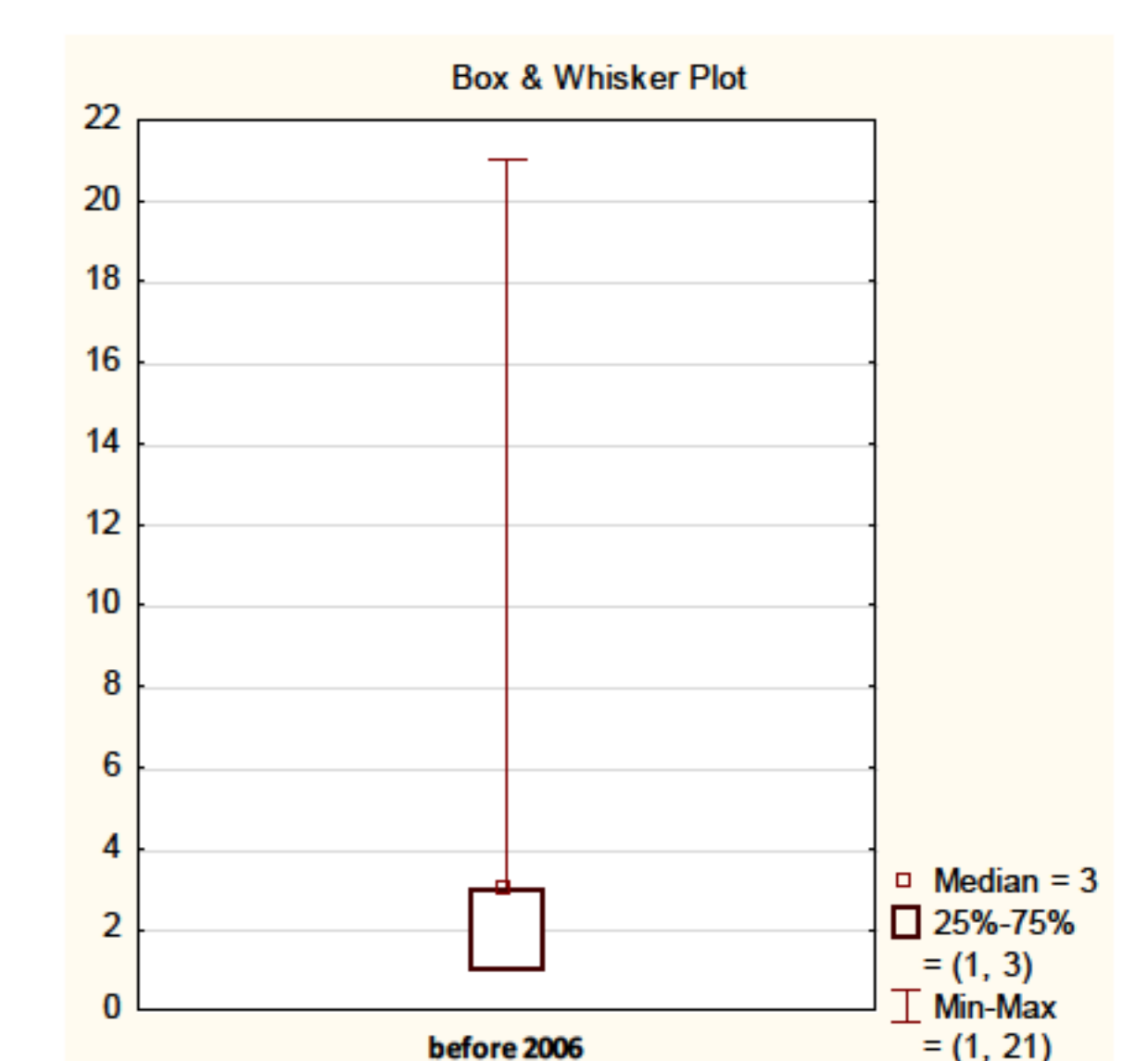
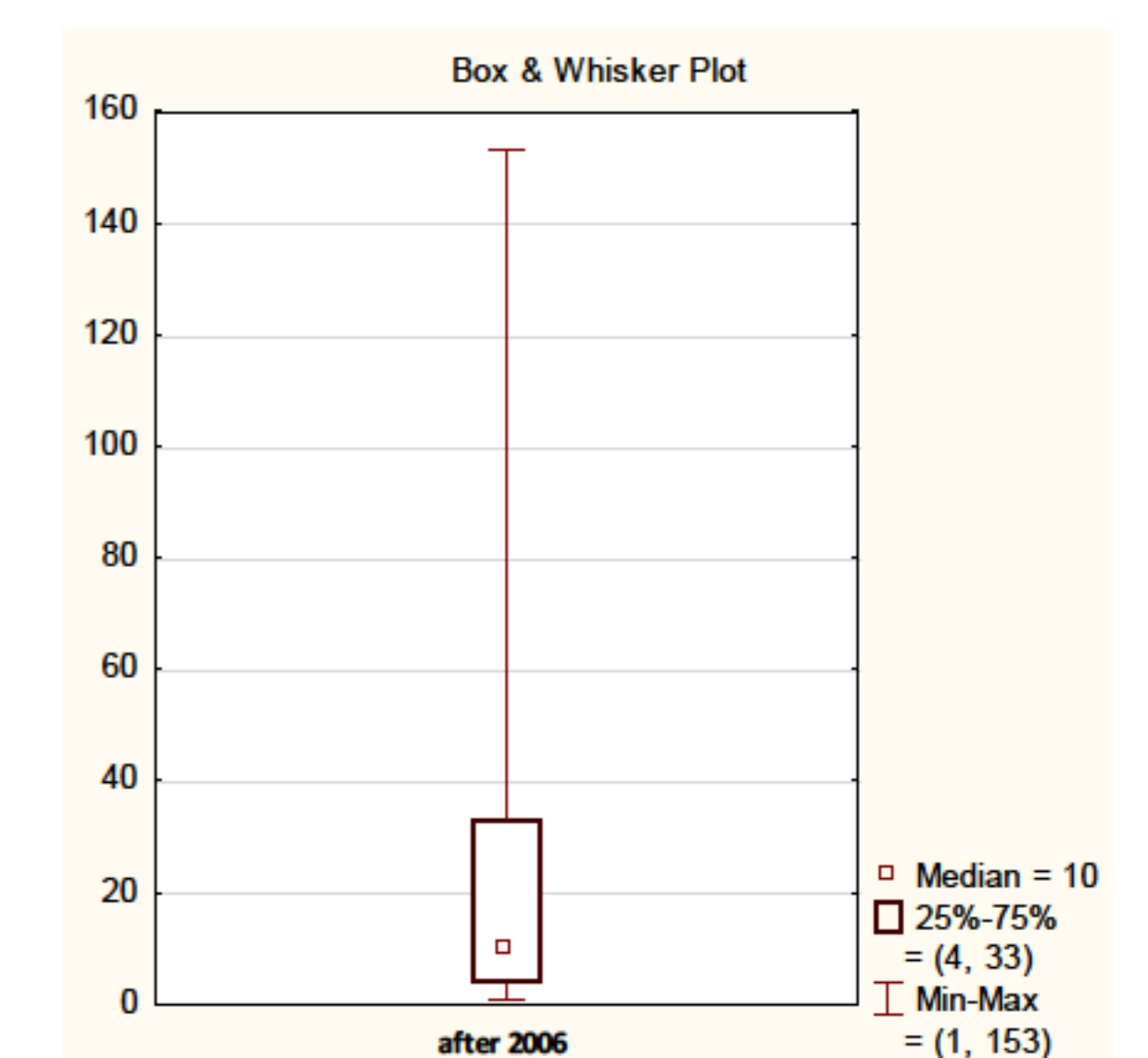
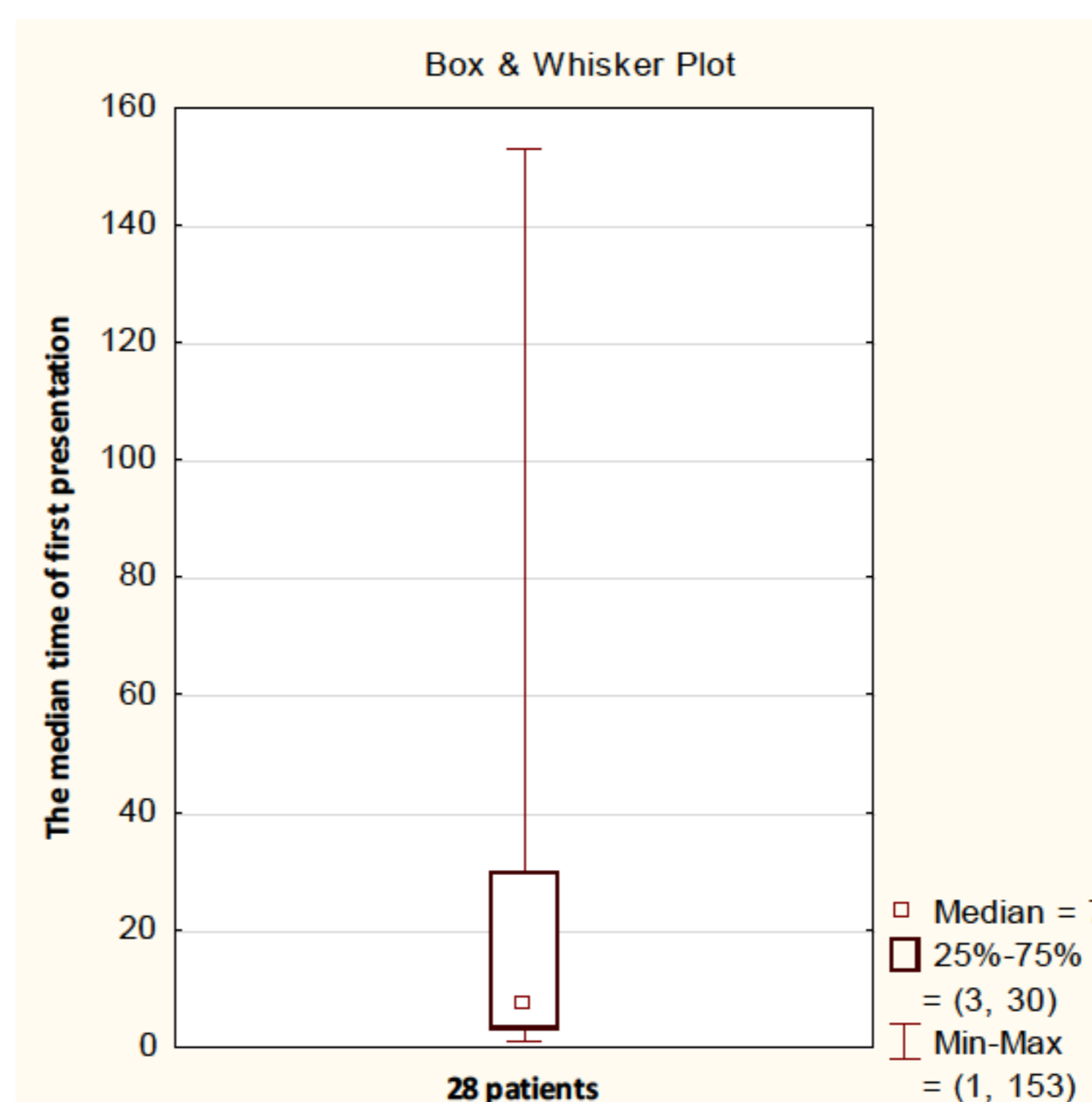
We performed a retrospective analysis of 28 patients (25 raised as males, 3 raised as females) with 46,XY DSD that presented for the first time at our department over the last 15 years. We excluded those patients where the reason of undescended testis (UDT) was obvious (i.e. CHARGE syndrome, prune belly syndrome, omphalocele) or the condition of UDT was acquired. We also excluded girls with female external genitalia (i.e. with complete androgen insensitivity syndrome or complete gonadal dysgenesis) in whom the initiation of diagnostic procedure was done due to primary amenorrhea or delayed puberty. The analysed group was not deprived of selective bias as this cohort had been selected for further genetic study.

Literature

- Hughes IA, Houk C, Ahmed SF, Lee PA, Lawson Wilkins Pediatric Endocrine Society (LWPES)/European Society for Paediatric Endocrinology (ESPE) Consensus Group. *Consensus statement on management of intersex disorders*. Arch Dis Child. 2006 July; 91(7): 554–563. Published online 2006 April 19. doi: 10.1136/adc.2006.098319
- Ahmed SF, Khwaja O, Hughes IA. *The role of a clinical score in the assessment of ambiguous genitalia*. BJU Int. 2000 Jan;85(1):120-4
- Zofia Kolesinska MD, S Faisal Ahmed MD, Marek Niedziela MD, Jillian Bryce PhD, Marta Molinska-Glura PhD, Martina Rodie MRCPCH, Jipu Jiang MSc, Richard O Sinnott PhD, Ieuan A Hughes MD, Feyza Darendeliler MD, Olaf Hiort MD, Yvonne van der Zwan MD, Martine Cools MD, Tulay Guran MD, Paul-Martin Holterhus MD, Silvano Bertelloni MD, Lidka Lisa MD, Wiebke Arlt MD, Nils Krone MD, Mona Ellaithi PhD, Antonio Balsamo MD, Inas Mazen MD, Anna Nordenstrom MD, Katherine Lachlan MD, Mona Alkhwari MD, Pierre Chatelain MD, Naomi Weintrob MD. *Changes Over Time In Sex Assignment For Disorders Of Sex Development*. Pediatrics. 2014 Sep;134(3):e710-5. doi: 10.1542/peds.2014-1088. Epub 2014 Aug 4.

Results

The median time of first presentation at the department of paediatric endocrinology was 7 months (range: 1-153). The time of first presentation was not correlated with the degree of undermasculinization expressed by EMS in those raised as males ($r_s=0.0$). The consensus on DSD issued in 2006 does not seem to influence the time of first presentation as the median time was 3 months (1-21) in the group that was diagnosed before 2006, and 10 months (1-153) after 2006, respectively.



Conclusions

There is a strong need to study the reasons of those patients with late presentation to the paediatric endocrinologist and to promote the knowledge on DSD management in regional neonatal departments.

