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The challenges in diagnosis and gender assignment in disorders of sex development presenting to a pediatric surgical unit in a developing country: The role of laparoscopy and simple tests for gender identity

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Abstract *Objective:* We aimed to assess how the diagnosis and determination of gender identity of disorders of sex development (DSD) is different in a developing country from Western medicine, and whether a pediatric surgery department can determine the underlying diagnosis and use simple tools to determine the likely gender identity (GI).
Material and methods: We reviewed the records of DSD patients admitted to the Department of Pediatric Surgery, Chittagong Medical College & Hospital (CMCH), Chittagong, Bangladesh, from January 2006 to December 2012 and performed a cross-sectional study on GI and gender-related behavior in these patients during the year 2012. DSD boys and girls answered a GI interview and had their gender role behavior assessed by observations of structural toy play and analyzed for differences in scores.
Results: This cohort of DSD patients presented in mid-childhood (6 months—16 years, mean 6.9 years) rather than infancy, and 30% came from consanguineous unions. Congenital adrenal hyperplasia (CAH) constituted only 11 of 50 (22%) of the DSD cohort, and not all families had access to steroid hormone replacement. A simple assessment of GI and gender-related behavior allowed effective gender assignment, as there was significant difference between DSD boys and girls in GI and gender-related behavior score.
Conclusions: DSD management in Bangladesh provides some unique challenges because of limited resources. A national reference laboratory for biochemical and genetic testing and

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development of a quaternary referral center for DSD patients will be helpful. Continued use of the GI interview and gender-related behavior study will enable effective interim decisions about diagnosis and management.

Introduction

The management of disorders of sex development (DSD) is complicated, and pediatric surgeons in developing countries are often confronted with extra challenges that do not occur in the developed world. Though there has been progress in diagnosis and management of DSD, many issues remain to be resolved [1]. Analysis of clinical management strategies has focused on gender assignment but the determinants of gender identity (GI) are much less known, and most likely multiple, such as exposure to androgens or social/cultural factors [2]. During a study of DSD patients that was carried out in the Department of Pediatric Surgery, Chittagong Medical College & Hospital (CMCH), there were a number of apparent differences between what we were finding and the current norms in the developed world [3]. This inspired us to consider more formally the challenges facing the diagnosis and gender assignment of DSD in Bangladesh, with the aim of providing some solutions to these problems.

Methods

We reviewed the records of DSD patients admitted to the Department of Pediatric Surgery, CMCH from January 2006 to December 2012 and performed a cross-sectional study on GI and gender-related behavior in these patients during the year 2012. Patients were asked to provide oral answers about GI in a questionnaire with the help of one of the authors and their parents (Appendix 1). After informed, written consent, patients were included if they were between 2 and 16 years of age. All patients completed a GI interview, observations of gender-role behavior with toys and games, and the child game participation questionnaire (CGPQ) and parent report on CGPQ [3–7]. A questionnaire with 13 questions on GI was used with each answer scored on a 3-point scale ranging from 0 to 2 (0, answer normal for a girl; 1, ambiguous response, kept silent or could not answer; 2, answer normal for a boy) [3–5]. Gender role behavior was observed by toy play over 10 min with three types of toys (boys: cars, pistols, aeroplanes; girls: dolls, doll clothing, kitchen supplies; neutral: books, sketchbook, marker pens) [8,9]. A score of 1 was assigned for each minute of play with a boy's toy. Toy to keep was scored by the type of toy the child chose to keep from a bag containing a pistol, ball, book, marker pen, and a doll with scoring from 5, 4, 3, 2, and 1 respectively. Children and their parents were asked about the type of game the children liked, with each masculine game assigned a score of 1 (Appendix 2). A composite score was then calculated by first converting all the scores into the same denominator by taking a least common multiple, and then by taking the means of the numerators after converting the numerators accordingly.

The Student *t*-test was used to compare the means of scores between the DSD boys and girls. The GI and gender role behavior tests were assessed to observe whether the assigned gender was in accordance with their GI. During the review, we evaluated the records of all the features in the presentation, investigations, and management, and the patients who underwent laparoscopy were presented in a tabulated form. Observation was done on how the presentation and diagnostic workup were different from what normally occurs in a developed country.

Results

Of the 50 DSD patients, 22% had 46, XX DSD with congenital adrenal hyperplasia (CAH), 64% with 46, XY DSD, 8% with mixed gonadal dysgenesis (MGD), and 6% with ovotesticular DSD. A higher (86%) percentage of DSD patients were from middle class families while only 12% came from poorer classes and 2% from higher classes. Parents of 15 (30%) were blood related, with seven of them first cousins. Consanguinity occurred in two of 11 (18%) of 46, XX DSD, 12 of 32 (38%) of 46, XY DSD, and one of four (25%) of MGD.

The ages when the DSD patients presented first ranged from 6 months to 16 years (mean 6.5 $\pm$ 3.9 years), and 2–16 years (mean 8.7 $\pm$ 4 years) when the GI interview was taken. While DSD boys had a mean age of 6.8 $\pm$ 4.2 years at presentation and 9 $\pm$ 4.1 years during GI interview, the mean age of DSD girls was 5.8 $\pm$ 3.3 years at presentation and 7.6 $\pm$ 3.9 years during the GI interview. Only 19 out of 50 DSD patients were less than 5 years old, and 31 out of 50 were greater than 5 years old at presentation. Thirty patients underwent karyotyping, which included eight of 11 patients with 46, XX DSD; 19 of32 patients with 46, XY DSD; and two patients with each of MGD and ovotesticular DSD. Other patients were included into the karyotype-based classification on the basis of the results of Barr body analysis to avoid previous terminology that was not patient friendly. Barr body analysis was done in 22 patients. It was present in the three 46, XX DSD patients, and absent in the other 19 patients. On clinical examination, the length of the genital tubercle (GT) (i.e. the stretched penile length) ranged from 2.5 mm to 5 cm, with 46, XX DSD patients having an average length of 2.6 cm (0.5–5 cm range). This was larger than the average (GT) length in 46, XY DSD patients of 1.9 cm (range: 0.25–4 cm). The majority of the patients had a Prader score of 4 (46%) with Prader 3 (28%), 2 and 5 (10% each), Prader 1 (6%), and none had Prader 0.

Cystoscopy was carried out on 22 patients. In two of the three patients with 46, XX DSD, it showed urogenital sinus