Journal of Pediatric Urology (2014) 10, 955—961

Journal of Pediatric urology

Impact of laparoscopy for diagnosis and treatment in patients with disorders of sex development

K. Moriya*, **K. Morita**, **T. Mitsui**, **T. Kitta**, **M. Nakamura**, **M. Kon**, **K. Nonomura**

Department of Renal and Genitourinary Surgery, Hokkaido University Graduate School of Medicine, North-15, West-7, Kita-Ku, Sapporo 060-0824, Japan

Received 6 November 2013; accepted 24 March 2014
Available online 12 April 2014

KEYWORDS
Disorders of sex development;
Diagnosis;
Treatment;
Laparoscopy

Abstract *Objective:* To review laparoscopy in patients with disorders of sex development (DSD) in order to clarify its usefulness in diagnosis, devising subsequent therapeutic strategies and managing patients with various conditions.
Patients and methods: Between April 1992 and December 2012, 29 laparoscopic surgeries were performed in 25 DSD patients. Among them, ten were diagnostic laparoscopy including gonadal biopsy, and 19 were therapeutic laparoscopy. Surgical procedures and complications were evaluated.
Results: For diagnostic laparoscopy, laparoscopic gonadal biopsy was performed in three patients. Inspection, with or without open gonadal biopsy, was performed on four out of seven patients with 46XY DSD or mixed gonadal dysgenesis (MGD). Additional surgery was planned and performed based on diagnostic laparoscopic findings in six out of seven patients. In the three patients with ovotesticular DSD, the gonadal pathology was diagnosed as: testis/ovary in one, testis/ovotestis in one and ovary/ovotestis in one — this was from the laparoscopic inspection and/or gonadal biopsy. However, the final diagnoses were bilateral ovotestis in two patients and ovary/ovotestis in one patient.

For therapeutic laparoscopy, surgical procedures were: gonadectomy in 17 patients (bilateral in 13, unilateral in three, partial in two); hysterectomy in two patients; orchiopexy in one; and sigmoid vaginoplasty in one patient (included multiple procedures). There were no severe perioperative complications. In the four patients with a history of diagnostic laparoscopy, no severe intra-abdominal adhesions that would disturb therapeutic laparoscopic surgery were observed.

Abbreviations: US, ultrasonography; MRI, magnetic resonance imaging; DSD, disorders of sex development; hCG, human chorionic gonadotropin; hMG, human menopausal gonadotropin; MGD, mixed gonadal dysgenesis.

* Corresponding author. Tel.: +81 11 716 1161x5949; fax: +81 11 706 7853.
E-mail addresses: k-moriya@med.hokudai.ac.jp (K. Moriya), kenordic@mac.com (K. Morita), mitsui68@med.hokudai.ac.jp (T. Mitsui), kitta@fb3.so-net.ne.jp (T. Kitta), tamago929@yahoo.co.jp (M. Nakamura), k-masafumi@mri.biglobe.ne.jp (M. Kon), k-nonno@med.hokudai.ac.jp (K. Nonomura).

http://dx.doi.org/10.1016/j.jpurol.2014.03.006

Conclusion: While diagnostic laparoscopy was helpful in devising a therapeutic surgical strategy in most of the patients with DSD who were suspected as having complex gonadal status or Müllerian duct derivatives, attention must be paid to precisely diagnosing the gonadal status in ovotesticular DSD. On the other hand, therapeutic laparoscopic surgeries were valuable procedures in treating DSD patients, even with a history of previous diagnostic laparoscopy.

Introduction

Disorders of sex development (DSD) are rare congenital anomalies with atypical chromosome, gonadal, or anatomical sex organ development. The incidence of DSD is reported as two in 10 000 live births [1,2]. To diagnose DSD, chromosomal, biochemical and endocrinological investigations, and various types of imaging such as ultrasonography (US) and magnetic resonance imaging (MRI) are required. The therapeutic strategy is devised depending on the type of DSD, anatomy of the external and internal genitalia, biochemical and endocrinological findings, and the sex assignment. However, since these kinds of disorders are rare and have many anatomical variations, individual care is necessary, especially regarding surgical management.

Recently, laparoscopy has gained wide acceptance in pediatric urology [3]. Laparoscopy is also reported to be a useful tool for diagnosing and treating DSD because of its minimal invasiveness and favorable cosmesis [4—7]. However, reports of evaluation and management for large numbers of DSD patients are limited.

In this study, laparoscopy in DSD patients was reviewed in order to clarify its usefulness in diagnosing, devising the subsequent therapeutic strategies, and managing the patients with various conditions.

Patients and methods

Between April 1992 and December 2012, after chromosomal and endocrinological analysis studies, 29 laparoscopic surgeries were performed in 25 DSD patients. The age at laparoscopic surgery ranged from 3.3 months to 21.1 years.

Among them, diagnostic laparoscopy including gonadal biopsy was performed on ten patients. In this institution, diagnostic laparoscopy is aggressively indicated for DSD patients who are suspected as having complex gonadal status or Müllerian duct derivatives with or without ambiguous genitalia. Genetic and endocrinological analysis is initially performed in order to devise strategies for therapeutic management. The gonadal biopsy was performed in the same manner as reported by Yu et al. [4]. Briefly, after the gonadal appearance was observed carefully by laparoscopy, the gonad was held by atraumatic grasping forceps and small specimens were obtained with toothed biopsy forceps. If heterogenic appearance was identified in the gonad, biopsy was performed from each portion of heterogenic appearance. Hemostasis with electrocauterization was performed thereafter.

Nineteen therapeutic laparoscopy surgeries were performed in 19 patients; this included four patients with a history of prior diagnostic laparoscopy. Impacts of diagnostic laparoscopy, the surgical procedure and the complications of laparoscopic surgery in patients with DSD were retrospectively evaluated.

Results

Diagnostic laparoscopy

The age for diagnostic laparoscopy ranged from 3.3 months to 20.7 years. The patients' demographic data are shown in Tables 1and 2. In one patient (Case 3) with 46 XY, who had perineal hypospadias with a left peeping gonad and a right non-palpable gonad, endocrinological tests showed a mild rise in testosterone but no rise in estradiol responding to hCG-hMG stimulation. Since bilateral gonads were confirmed as immature testis by diagnostic laparoscopy with gonadal biopsy, he was finally diagnosed as having partial gonadal dysgenesis. Final diagnoses after diagnostic laparoscopy in these ten cases were 46XY DSD in four, MGD in three, and ovotesticular DSD in three.

Among the four cases with 46XY DSD, simultaneous transinguinal gonadectomy just after laparoscopic inspection of gonadal status or Müllerian duct derivatives was performed in two patients. These two patients had completely female-type external genitalia and were already assigned as females. Since their bilateral gonads were diagnosed as testes by karyotype, preoperative endocrinological examination and laparoscopic homogeneous appearance, simultaneous gonadectomy was performed. In the remaining two patients with 46XY DSD, laparoscopic gonadal biopsy was performed. Pathological examination confirmed that these gonads were testes. In one patient, who was assigned as a female, bilateral gonadectomy through the labial incision at the time of subsequent feminizing genitoplasty was performed. In the other, who was assigned as a male, an open hysterectomy with bilateral Fowler Stephens orchiopexy was performed at the time of subsequent urethroplasty.

Among the three with MGD, laparoscopic inspection revealed the existence of a uterus in two of them. Laparoscopic gonadal biopsy was carried out in the one who was assigned as a female with complex mosaicism of 47XYY/45X/46XY. Since immature testes were suspected from pathological examination, a laparoscopic bilateral gonadectomy was indicated as the therapeutic procedure thereafter. In the one with male gender, who had bilateral gonads in the scrotum confirmed by physical examination and laparoscopic inspection, unilateral open gonadectomy by a genital incision for atrophic gonads was performed at