Diagnostic laparoscopy in a twelve year old girl with right iliac fossa pain: A life changing diagnosis of complete androgen insensitivity syndrome

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**Abstract**

**Introduction:** Right iliac fossa (RIF) pain is one of the most common presenting complaints faced by general surgeons in the emergency department. Correct diagnosis and appropriate surgical intervention can often pose a challenge. While haematological and radiological investigations can aid in diagnosis of patients with RIF pain, they can frequently be normal in the presence of significant pathology. In this case report, we present a twelve year old girl with RIF pain, who was incidentally found to have complete androgen insensitivity syndrome when undergoing diagnostic laparoscopy for acute appendicitis.

1. Introduction

Right iliac fossa (RIF) pain is one of the most common presenting complaints faced by general surgeons in the emergency department. Correct diagnosis and appropriate surgical intervention can often pose a challenge. While haematological and radiological investigations can aid in diagnosis of patients with RIF pain, they can frequently be normal in the presence of significant pathology. In this case report, we present a twelve year old girl with RIF pain, who was incidentally found to have complete androgen insensitivity syndrome when undergoing diagnostic laparoscopy for acute appendicitis.

2. Case report

A 12-year-old girl presented to the emergency department with a four day history of initially central acute abdominal pain, now localised in the RIF. She described the pain as constant and associated with anorexia. She denied any urinary symptoms and is pre-menarchal.

Her background surgical history consisted of a left inguinal hernia repair at the age of 11 months. She was on no medications and had no allergies.

Her family history revealed her to be one of five sisters, and their mother known to be a carrier of androgenic insensitivity syndrome. The oldest sister aged 21 years had been diagnosed with complete androgen insensitivity syndrome (CAIS). The second oldest sister aged 17 year is well and has normal menstrual periods. The third sister aged 14 years has primary amenorrhoea but has not been tested for CAIS. The youngest sister aged 11 years is also pre-menarchal and has not been tested for CAIS.

On examination she was afebrile and noted to have mild RIF tenderness without any guarding. There was no abdominal...
Distension and bowel sounds were present. She was phenotypic female, weighing 51.1 kg and her height measured at 1.67 m. Her breast development was at Tanner stage II (breast buds formed, with small area of surrounding glandular tissue).1

Bloods tests showed normal range of white blood cell count and C-reactive protein concentration. The urine dipstick was clear. She went on to have an ultrasound scan of her abdomen and pelvis which did not visualise the appendix. No free fluid was noted, however her uterus was noted to be small sized, and her ovaries could not be visualised.

She was admitted to the hospital for observation. On examination the following morning her RIF tenderness was more marked and associated with guarding. After discussion with the patient and her parents about the possibility of acute appendicitis and its risks, a diagnostic laparoscopy was performed.

During laparoscopy, the following findings were made: macroscopically dilated appendix, right and left gonads (Figs. 1 and 2 respectively) at the internal opening of the inguinal canal (appearances consistent with intra-abdominal testes), empty pelvis (Fig. 3) with a rudimentary uterus on the right side (Fig. 4). No evidence of fallopian tubes or connection of uterus to the vagina and broad based, non-inflamed Meckel’s diverticulum (Fig. 5).

An intra-operative pelvic examination by a gynaecology consultant confirmed a short, blind-ending vagina with a mid-line band and the absence of a cervix.

Laparoscopic appendectomy was performed, with the histology confirming acute appendicitis.

3. Discussion

Androgen insensitivity syndrome (AIS) is a disorder of hormone resistance characterised by a female phenotype in an individual with an XY karyotype and testes producing age-appropriate normal
concentrations of androgens. The presence of the Y chromosome directs testicular development through a switch gene present on its short arm, called the SRY gene. CAIS is the most common condition leading to an XY females with an estimated incidence of 1:40,000–60,000 births.

In CAIS, there is no potential for virilisation, the atypical chromosomes are usually identified in adolescence during investigations for primary amenorrhoea and/or delayed puberty. In this condition, there is a defect in the androgen receptor (AR) leading to a complete absence of function, and consequently no androgenic action on target cells. Phenotypically, there is normal breast development but minimal pubic and axillary hair. Testes are normally developed but can be located anywhere in their line of descent, usually leading to the formation of a unilateral or bilateral inguinal hernia. CAIS diagnosis is based on the following criteria:

1. The presence of testes along with normal female external genitalia in a 46, XY individual,
2. Identification of an androgen receptor (AR) mutation,
3. Spontaneous feminization (with primary amenorrhoea) at puberty before gonadectomy with no virilisation despite normal or high male levels of testosterone,
4. Markedly decreased or absent post pubertal axillary and pubic hair growth.

In females with one mutated X chromosome, each pregnancy has a 1 in 4 chance that a daughter is carrier only and can pass on the mutated gene to her children.

The diagnosis of children with disorders of sex development (DSD) requires a karyotype and different biochemical and radiological investigations in the context of a multidisciplinary team.

The application of laparoscopic surgery in infants and children has expanded tremendously in recent years. However, its feasibility remains somewhat controversial. Most reports on the feasibility of laparoscopy in children are restricted to specific procedures such as appendectomy, pyloromyotomy, or fundoplication. Minimally invasive surgery has however revolutionised many aspects of paediatric surgery, and the indications for laparoscopic surgery have gradually expanded in children.

For patients with DSD the diagnostic laparoscopy has become, in many centres, a standard procedure. Laparoscopy provides an excellent view of the intra-abdominal gonads and internal genitalia by magnification, maintaining a wide space, easy access to the pelvic cavity and adequate illumination, and is helpful for correct diagnosis of these patients.

In a recent study comparing the diagnostic value of laparoscopy and ultrasonography (US) in the assessment Mullerian structures in children with complex DSD, pelvic US was found to have significant limitations with only 54% sensitivity and 50% specificity. Although laparoscopy requires a general anaesthetic, it allowed excellent visualisation of pelvic structures and gonads in children with complex DSD. Laparoscopy provides the further benefit of allowing for tissue sampling and surgical intervention as well as a diagnostic tool.

The non-infamed Meckel Diverticulum (MD) which was incidentally discovered was not operated on in this patient. Thought to be one of the most common congenital anomalies, affecting 1–3% of the population. MD is formed when there is incomplete obliteration of the vitelone duct and is associated with an increased risk of other congenital anomalies, including cardiovascular, neurological, and gastrointestinal. To our knowledge, this is the first reported case of MD in association with CAIS incidentally diagnosed during laparoscopic appendectomy.

In most cases, a MD is an incidental finding, with only approximately 16% ever becoming symptomatic. The diagnostic value of laparoscopy for MD is well documented, however little evidence exists in support of prophylactic resection in cases of incidental finding where other pathology is confirmed. Where available such data is based on small number case series from single centres. Comparing outcomes of laparoscopy vs laparotomy for resection of MD, laparoscopy with stapled diverticulectomy or small bowel resection appears to have comparable and in some cases better outcomes than laparotomy.

Because of the potential for germ cell tumours in dysgenetic gonads and intra-abdominal tests in patients with Y chromosome material and gonadal dysgenesis as well as the various inter-sex disorders, these patients have been routinely advised to undergo gonadectomy. A gonadectomy was not performed on this patient for several reasons, foremost among these was that the patient presented as an emergency with RIF pain which was caused by acute appendicitis and her diagnosis of CAIS was unknown prior to the laparoscopy. While gonadectomy is recommended in phenotypic females with CAIS, this should be performed after appropriate discussion with the patient, their family and allow for sufficient counselling. There remains controversy regarding the ideal timing of gonadectomy for patients with DSD, however in CAIS postpubertal gonadectomy is recommended to allow for complete feminization and development of breasts.

This case report highlights the advantage of laparoscopy as a diagnostic and treatment tool in a twelve year old girl with multiple intra-abdominal findings. While the ultimate diagnosis responsible for her symptom of RIF pain was acute appendicitis, the additional diagnosis of CAIS and incidental Meckel’s would have otherwise likely gone undetected.

Conflict of interest
None.

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Ethical approval
Written informed consent was obtained from the patient and their guardian (next of kin).
A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author contributions
Babak Meshkat: Author of introduction, case report and conclusion.
Melania Mat covici and Claire Buckley: Literature review and authors of discussion section.
Muhammad Salama: Formatting, corrections and feedback on early drafts.
Haresh K Perthiani: Senior author, operating surgeon during case, consultant surgeon primarily looking after patient.

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Key learning point

• Case of incidental finding of androgen insensitivity syndrome. Diagnosis and management of complete androgen insensitivity syndrome.

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