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**“Imperforate hymen: retrospective review from a single tertiary centre of presenting symptoms and diagnostic process”**

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**“Imperforate hymen: retrospective review from a single tertiary centre of presenting symptoms and diagnostic process.”**

**Abstract:**

**Aim:**

To review presentations, diagnostic processes and time to diagnosis of presentations of imperforate hymen at one tertiary paediatric hospital over a 10-year period. The aim is to

improve knowledge, enable targeted education and to reduce unnecessary tests and diagnostic delay for this congenital obstructive disorder of the female reproductive tract.

#### Methods:

A pragmatic, retrospective, observational study of all female patients, aged less than or equal to 18 years of age, presenting to the Royal Children's Hospital, Melbourne, over an 18-year period. Presenting symptom(s), time to diagnosis, investigations ordered, and number of healthcare professionals consulted prior to diagnosis.

#### Results:

27 patients met eligibility criteria. 85% presented with pain (23/27). Pain was frequently associated with other symptoms (15/23). Over three quarters of patients consulted more than one healthcare professional prior to diagnosis (22/27). 19/27 had imaging undertaken prior to diagnosis.

#### Conclusions:

Paediatric and adolescent females managed at our tertiary paediatric hospital are experiencing diagnostic uncertainty and the unnecessary ordering of imaging prior to a diagnosis of imperforate hymen. Targeted education is recommended to health professionals managing female paediatric and adolescent patients.

### **Brief points:**

- **What is already known of this topic:** Imperforate hymen is an uncommon congenital obstructive disorder. Common presenting symptoms include primary amenorrhoea, abdominal and/or back pain and “rare” presentations with other body system symptoms. Diagnosis is clinical and does not require imaging to be ordered.
- **What this paper adds:** Although pain is the most common presenting symptom of imperforate hymen, the majority of presentations involve multiple symptoms from more than one body system. It is not uncommon to present with other symptoms. The majority of patients are consulted by multiple health professionals and undergo diagnostic imaging to achieve diagnosis.

### **Key words:**

#### MeSH terms

- Hymen
- Hymen, Imperforate
- Amenorrhoea
- Gynaecology
- Female, adolescent
- Delayed diagnosis
- Emergency medicine
- Urinary retention
- Constipation

## Introduction

Imperforate hymen is an uncommon congenital obstructive disorder of the female reproductive system. The literature reports an incidence of 0.014 – 0.02% in children and 0.05% in gynaecological patients.<sup>1</sup> It occurs when the hymen, an embryological remnant of mesodermal tissue, fails to perforate during the later stages of embryo development. Clinical presentation can occur in infancy with inter-labial swelling, or more commonly in adolescent girls presenting with primary amenorrhoea, a bulging hymenal membrane, cyclical abdominal pain and / or a pelvic mass. A review of the literature reveals that there are numerous of case reports of “rare” presentations with other presenting symptoms including urinary retention,<sup>2-8</sup> acute kidney injury,<sup>9</sup> constipation and tenesmus,<sup>1</sup> back pain<sup>10</sup> and dyspareunia.<sup>11</sup>

Imperforate hymen is a clinical diagnosis. A history of symptoms as described above, in a female of menarchal age (normal range 8-14 years) is suggestive. Clinical examination involves a general examination, and in particular a focused abdominal and genital examination. The latter should be performed in a private space, free from interruptions. It is recommended that a trusted caregiver be present for the examination of all prepubertal girls.<sup>12</sup> Consent to conduct the examination should be sought from the patient and the caregiver. The process of the examination should be explained to the patient to reduce fear and anxiety, and to increase the patient’s sense of control. The genital examination of a prepubertal girl is usually performed in the supine frog-legged position. While supine, the feet are placed together and the knees apart. The labia majora can be visualised. Separation of the labia minora allows for visualisation of the hymen, urethra and clitoris. It is not recommended, nor necessary, to perform an internal vaginal examination for the diagnosis of an imperforate hymen. Internal vaginal examinations are seldom required in patients prior to the onset of sexual activity and specialist input is recommended if this is required.

Clinical examination findings include normal vital signs, height and weight measurements. Abdominal examination may reveal a palpable mass, which may represent urinary retention, haematocolpos and / or haematometra or constipation. Genital examination reveals normal female external genitalia. The hymen is usually a rim of tissue around the lower end of the

vaginal orifice. When it is imperforate, it is a thin membrane which covers the vaginal opening. The membrane may 'bulge' due to haematocolpos and may be bluish in appearance due to retained old blood within the vagina. Variations to a complete imperforate hymen that do not result in obstruction can sometimes be found, which include microperforations or septate hymen. The main differential diagnosis is a transverse vaginal septum. This is where a horizontal band of tissue forms within the vagina, obstructing the vaginal canal. The septum can occur at various levels of the vagina. Classically a low transverse vaginal septum (blockage close to the hymen) is the main distinction from an imperforate hymen. In these cases, the hymenal ring is patent, and the septum is found internal to the hymenal ring. The septum may be thin and mimic a membrane; however it is usually a thicker band of tissue. This differentiation can occasionally be difficult on first examination. If there is a high index of suspicion for a transverse septum, an ultrasound performed by an experienced gynaecology sonographer can distinguish between these diagnoses. The treatment for an imperforate hymen involves an examination under anaesthetic and a small cruciate incision made in the centre of the hymen. Retained blood is evacuated. There are no known long term consequences of an imperforate hymen.

Many patients are undergoing imaging (ultrasound [USS], abdominal x-ray [AXR], computerised topography [CT], magnetic resonance imaging [MRI]) to make the diagnosis. Given the diagnosis can be made clinically, ordering imaging is likely to delay diagnosis and management.

As the standard teaching is presentation with lower abdominal pain and primary amenorrhoea,<sup>8,13</sup> we wished to explore the frequency and range of other presentations as well as the investigative approach, diagnostic delay, and the number of health professional reviews prior to diagnosis that occur in these adolescents who are managed in a tertiary children's hospital. Our purpose in undertaking this audit is to improve knowledge, and potentially use this to enable targeted education to relevant medical professionals.

## **Materials and method**

This audit of the care of adolescents with an imperforate hymen was a pragmatic retrospective observational study. All female patients, less than or equal to 18 years of age, managed at RCH from January 2000 to January 2018, for imperforate hymen were identified through the operating theatre database under the term hymenectomy. Given the code for hymenectomy also included correction of other hymenal anomalies, these were excluded. Medical records of all patients fulfilling inclusion criteria were reviewed.

Medical records were reviewed to determine patients' presenting symptom(s), time to diagnosis from first presentation to a health professional, investigations ordered to make a diagnosis and the number of presentations to a health professional prior to diagnosis. Data was extracted from RCH records, collated and de-identified.

HREC permission to undertake this audit was obtained.

## **Results**

During the study period, 29 paediatric and adolescent females were managed at RCH for an imperforate hymen. Two were excluded from final analysis given their diagnosis had occurred prior to pubertal age from an incidental finding on imaging. The mean age at presentation was 12yrs, range 2 months –to 18 years. The number of presentations per year ranged from 1-5 (figure 1).

Eighty-five percent of patients presented with pain (23/27); abdominal pain was significantly more common than back pain (n = 16 vs. 4 and an additional 3 patients having both abdominal and back pain). Pain was frequently associated with other symptoms; with gastrointestinal (GIT) symptoms (n=4), GIT and urinary symptoms (n=2), GIT and mass (n=1), urinary symptoms (n=4), urinary symptoms and mass (n=1), mass (n=2) and fever (n=1). Of the four patients presenting without pain, 2 patients presented with primary amenorrhoea and 2 patients presenting with GIT symptoms. Figure 2 illustrates symptoms present at diagnosis.

Greater than 80% of patients presented to more than one health professional prior to diagnosis (22/27). This included presentations prior to presenting to our hospital (e.g. from general practitioners). The number of presentations to healthcare professionals prior to diagnosis is illustrated in figure 3.

Approximately 70% of patients had imaging to diagnose imperforate hymen (19/27). Pelvic USS was the most common imaging modality ordered (15/19); two of these patients had multiple imaging ordered (USS / AXR / MRI and USS / CT respectively). One patient underwent a pelvic MRI alone, and 2 patients had an AXR alone. Figure 4 illustrates this. Two thirds of patients were diagnosed on the day of presentation (18/27). There were two outlying cases that took 1 and 2 months respectively from initial presentation to diagnosis. Excluding these, the mean time to diagnosis was 1.4 days (range 1-7 days).<sup>2</sup> The presenting symptoms of patients that took 1 or more month to diagnosis was (a) primary amenorrhoea, which was initially worked up by a GP without complete examination (b) abdominal and back pain without a definite cyclical nature. No examination was undertaken at initial presentation. Time to diagnosis is illustrated in figure 5.

## **Discussion**

The majority of patients presenting to RCH with a diagnosis of imperforate hymen had symptoms from multiple organ systems. This is in comparison to the literature, whereby symptoms from single organ systems are frequently discussed. Over half of the patients presented with the combination of pain and another symptom (14/27). The awareness and pattern recognition of the common combinations that the diagnosis of imperforate hymen presents with, may reduce diagnostic uncertainty, reduce diagnostic delay and decrease the ordering of unnecessary investigations.

Unfortunately, 70% of patients required a visit to more than one healthcare professional prior to diagnosis. Healthcare professionals predominantly included general practitioners (GP), emergency physicians, gynaecologists, or any combination of these. It is likely that omitting an adequate gynaecological history or examination in female paediatric and adolescent

patients presenting for care, is the predominant cause of failure to diagnose an imperforate hymen. Given the unique area of paediatric and adolescent gynaecology, which spans general practice, paediatrics, emergency medicine and gynaecology, it is a common area whereby inadequate education and knowledge, exacerbated by limited experience, contributes diagnostic uncertainty.

It is likely that inadequate experience and / or knowledge is further illustrated by the fact that three quarters of our patients underwent diagnostic imaging to make the diagnosis of imperforate hymen. Imaging including pelvic ultrasound, CT scan, AXR, MRI, with 1 patient having multiple imaging modalities ordered. Imaging is unnecessary in the diagnosis of an imperforate hymen, which can be a clinical diagnosis based on history and examination. Importantly, this is a concerning finding from both a patient viewpoint due to the ensuing anxiety and risks associated with ionizing radiation if AXR and CT are utilised, but also from a healthcare expenditure viewpoint of the unnecessary costs and human hours involved. Eighty six percent of our patients were diagnosed within 1 week of presentation (n = 25). All of our patients were within three months of symptom onset. This is reasonable given the cyclical exacerbation of menstrual symptoms. Nevertheless, there was a considerable delay in presentation by young women with symptoms being present for several months prior to presentation. Therefore, despite the diagnostic uncertainty in making a diagnosis of imperforate hymen, it does not appear to significantly delay patients receiving treatment. It is unable to be determined from our retrospective review and small numbers, whether this is an accurate representation of a larger cohort of patients, or whether patients seen in other centres from RCH are enduring delay from presentation to definitive management.

Strengths of our analysis is that this is the first paper, to our knowledge, to analyse multiple presentations of imperforate hymen across almost 2 decades. This enabled identification of symptoms patterns, and in particular the multiple concomitant symptoms present at diagnosis, which appears more common than isolated organ system symptoms. This is also the first paper to analyse diagnostic uncertainty and unnecessary investigation ordering, which is likely the result of inexperience and inadequate knowledge in paediatric and adolescent gynaecology.

Finally, medical records were examined by one researcher and all relevant data was able to be collected from the records thereby eliminating the risk of bias due to incomplete records.

There are several limitations to a retrospective observational study, including the quality of data analysed being limited to what has been documented in the medical records, change in knowledge of paediatric and adolescent gynaecology in our emergency department over this study period, absence of data from patients presenting at other hospitals which our data may not be representative of and the small numbers of patients that may limit generalizability of the results.

### **Conclusion**

Paediatric and adolescent females managed at our tertiary paediatric hospital are experiencing diagnostic uncertainty and the unnecessary ordering of imaging prior to a diagnosis of imperforate hymen. Targeted education is recommended to health professionals managing female paediatric and adolescent patients, in order to reduce diagnostic uncertainty and costly medical investigations.

**Word Count:** 1803 1 words

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## Figures

Figure 1: Number of presentations to RCH with imperforate hymen over the study period

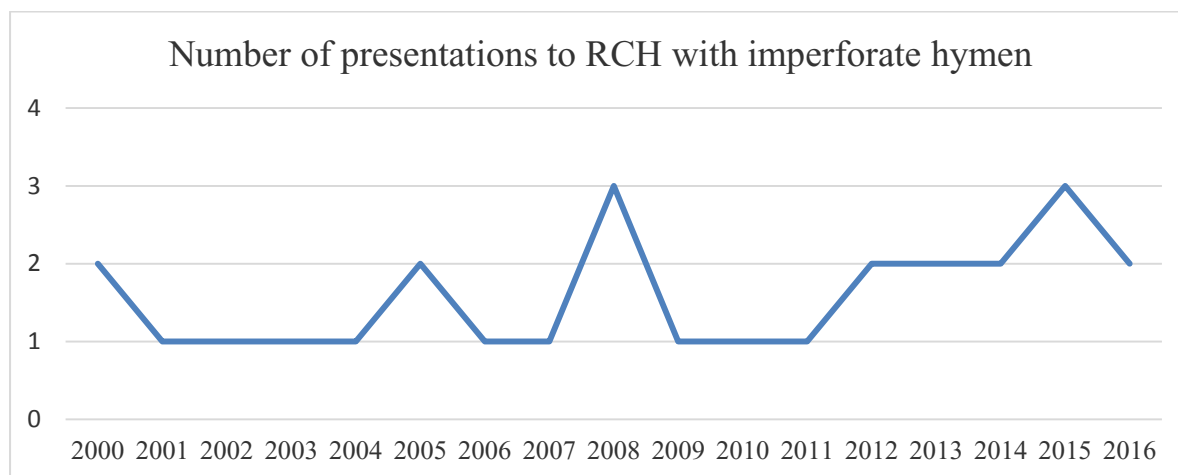


Figure 2: Symptoms present at the time of presentation to RCH

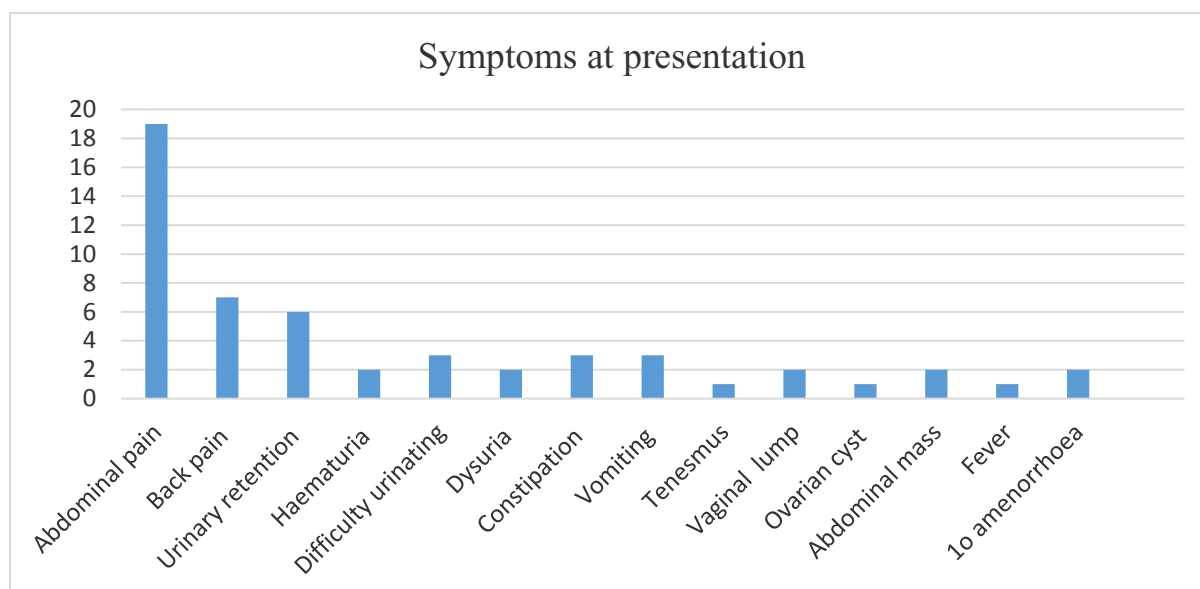


Figure 3: Classification of symptoms at presentation

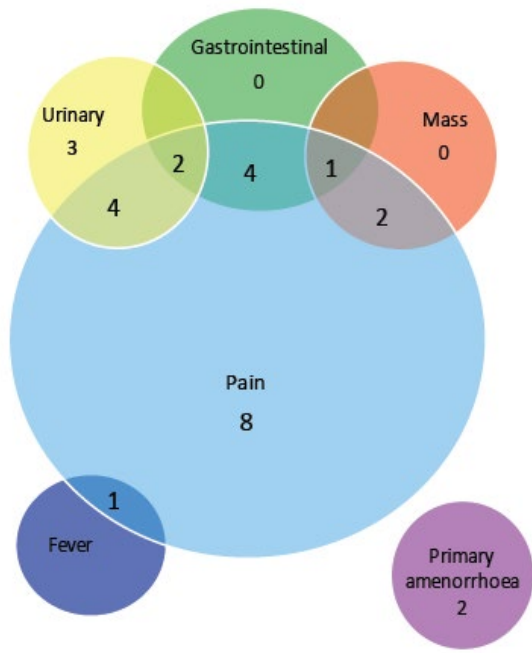


Figure 4: Number of presentations to a healthcare professional prior to diagnosis

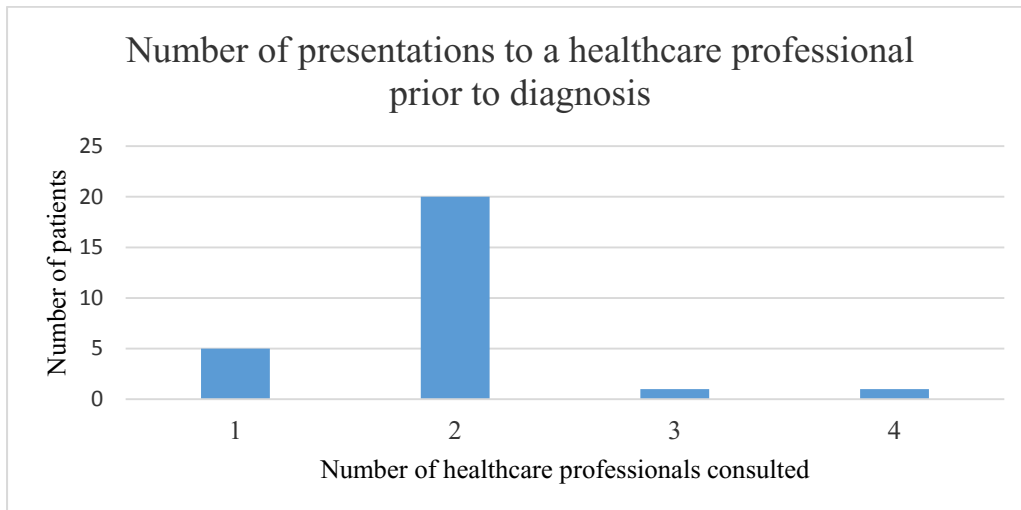
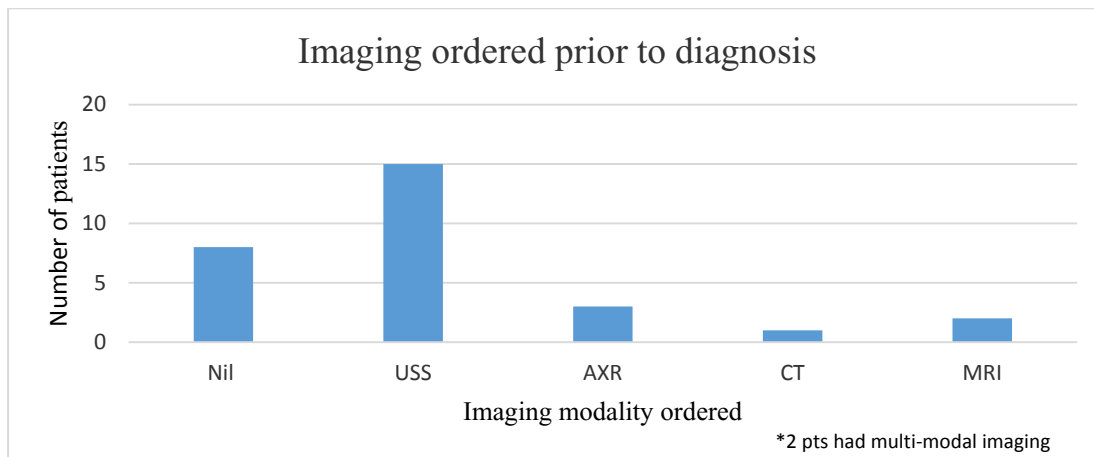


Figure 5: Imaging ordered prior to diagnosis



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