



## Endrology &amp; Infertility

## Massive tubular ectasia of the rete testes in a patient with Marfan syndrome

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## ABSTRACT

This case study explores massive tubular ectasia of the rete testes in a patient with Marfan syndrome. Marfan syndrome is a connective tissue disorder and has no known association with tubular ectasia in published research. Despite the unclear pathophysiology of tubular ectasia, potential causes include mechanical obstruction and congenital deformity. The patient, a male in his 40s, presented with bilateral testicular pain and swelling up to a volume of 200 cc over two years. Tubular ectasia was diagnosed on ultrasound and MRI, no obstructive aetiology was identified. Initial conservative management was chosen, but further enlargement led to reconsideration of treatment options.

## 1. Introduction

Tubular ectasia is a benign condition characterised by dilatation of the rete testis.<sup>1</sup> This case study presents a patient with Marfan syndrome who developed tubular ectasia of the rete testes, highlighting the clinical presentation, investigations and management.

Marfan Syndrome is diagnosed clinically using the Ghent nosology.<sup>2</sup> It is a connective tissue disorder affecting fibrillin, crucial for tissue elasticity.<sup>3</sup> Mutations in fibrillin lead to Marfan syndrome,<sup>4</sup> characterised by skeletal, cardiac, and ocular features.<sup>5</sup>

Tubular ectasia of the rete testes involves cystic dilatation of seminiferous tubules in the testicle.<sup>6</sup> The pathophysiology of the condition is unclear but may include mechanical obstruction, congenital deformity, or androgen deficiency.<sup>7</sup> Observational studies suggest other potential causes including post-vasectomy status, inguinal hernia, or epididymal cysts.<sup>8</sup> Ultrasound shows hypoechoic coarse cystic lesions, and MRI shows low T1 and high T2 signal intensity. These pathognomonic imaging findings are diagnostic, requiring no further investigations for this benign condition.<sup>9</sup>

A literature search was conducted searching the key words Marfan-Marfan disease or syndrome, and testicular ectasia - cystic ectasia of the rete testis - rete tubular ectasia-cystic transformation of rete testis. Databases PubMed, Embase/Ovid and Google Scholar were searched for relevant English-language publications from any date and including any age group. No results were identified which included both key terms.

## 2. Case presentation

A male patient in his forties, presented to the emergency department in 2022 with scrotal pain and swelling on the left side, along with left iliac fossa pain.

The medical history includes Marfan's syndrome with a previous mechanical aortic valve replacement, for which the patient is on warfarin, as well as trauma related right hip osteoarthritis. There is no previous history of cryptorchidism, vasectomy or inguino-scrotal surgery. The patient has previously conceived two children. The patient is full time employed, does not smoke and drinks a moderate amount of alcohol.

Upon presentation there was no reported dysuria, frequency, discharge, or haematuria. On initial examination, the scrotum was unremarkable apart from the swelling with no evidence of hernia. Although the urine analysis was not suggestive of infection a clinical diagnosis of epididymo-orchitis was made and the patient was treated with a course of antibiotics. The patient next presented a year later with increased swelling and discomfort and was referred to the Urology department for assessment.

Initial investigations included a negative urinalysis and sexually transmitted infection testing, normal blood tests, including kidney, liver, thyroid function, sex hormones, haematology and inflammatory markers. An initial ultrasound scrotum (Fig. 1a) revealed classical appearances of tubular ectasia measuring up to 5cm in both testes with the

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right testicular volume estimated at 26 cc and the left testicular volume estimated at 37 cc.

A follow up ultrasound five months later showed enlargement of the tubular ectasia which measured up to 8cm in the right testes and 7cm in the left testis (Fig. 1b). A further follow up ultrasound 10 months after his second ultrasound (Fig. 1c) showed significant interval enlargement of the tubular ectasia with the right testicle measuring 222 cc and the left measuring 119 cc with the tubular ectasia progressively occupying most of the testicular volumes. Additional MRI investigation (Fig. 2) showed the testicles with tubular ectasia and no identified ductus obstruction.

After discussion at the urology multi-disciplinary team meeting (MDT), the patient was initially managed conservatively and reassured about the benign nature of the condition. However, one year later, the patient was re-referred due to further increase in size and discomfort. Imaging confirmed the persistence of tubular ectasia with no new lesions or concerning features. The case was discussed at the MDT meeting again, and options of trial of aspiration or bilateral orchidectomy with testosterone replacement were considered. The patient was seen for clinical consultation and opted for continuation of conservative management.

### 3. Discussion

Tubular ectasia is typically benign with no known clinical sequelae. Obstructive causes were ruled out in this case. The epithelial histology of the rete testes consists of multiple cell types on the luminal surface. Peripheral to the basal lamina, there are layers of fibroblasts, collagen and elastic fibres<sup>10</sup>. In Marfan syndrome encoding mutations in the

FBN-1 gene lead to alterations in production of fibrillin-1. Fibrillin microfibrils form a peripheral mantle surrounding elastic fibres, contributing to the maintenance of the shape and function of connective tissues<sup>11</sup>. It is plausible that changes in fibrillin affect the elastic fibres in the rete testis epithelium, thereby reducing the structural integrity of the connective tissue scaffolding. Variations in the FBN-1 gene sequence may result in different phenotypic manifestations of gene expression<sup>12</sup>. Genetic mutation screening could identify a subtype of the gene responsible for a fibrillinopathy that specifically affects the elastic fibres specific to the rete testis.

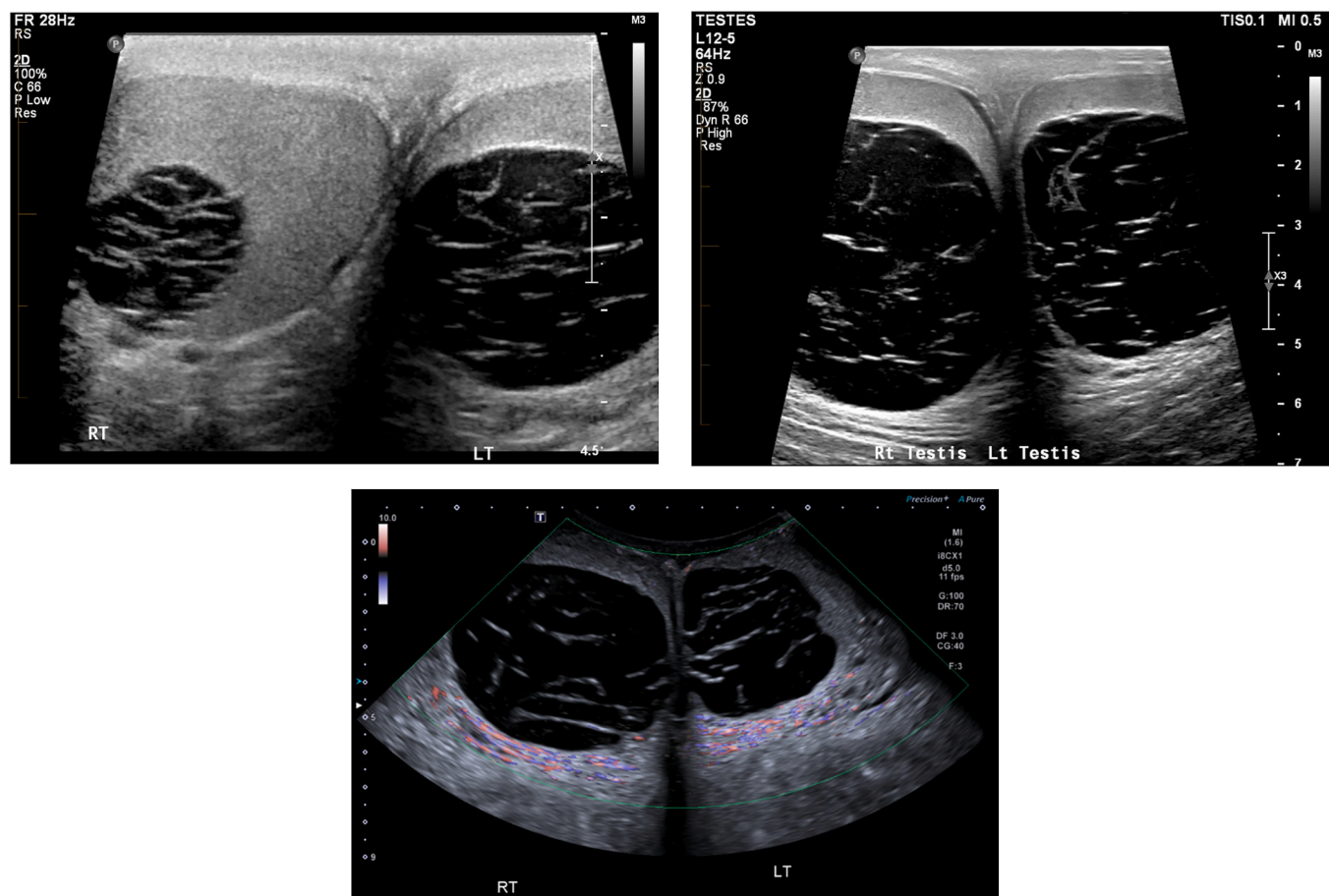
While Marfan syndrome might have contributed, no literature supports an association between Marfan syndrome and tubular ectasia of the rete testes. The pathophysiology remains unclear, suggesting a need for further research.

### 4. Conclusion

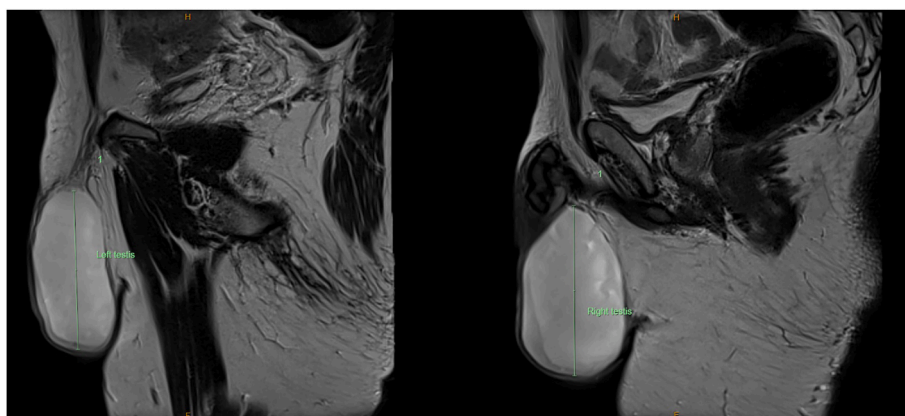
This case highlights the importance of thorough clinical evaluation and appropriate imaging in the correct diagnosis and management of tubular ectasia. Reassurance and conservative management is usually appropriate for this benign condition; however, intervention can be considered in severe cases.

### CRediT authorship contribution statement

**Ashley Lee:** Writing – review & editing, Writing – original draft, Visualization, Validation, Project administration, Methodology, Investigation, Formal analysis, Data curation, Conceptualization. **Ronny Low:** Writing – review & editing, Writing – original draft, Visualization,



**Fig. 1. (1a, 1b, 1c).** Ultrasound appearances of bilateral testicles with tubular ectasia.  
Legend: Transverse section of the scrotum at the midline showing the right (RT) and left (LT) testicles.



**Fig. 2.** MRI scrotum

Legend: Left testicle 109 cc Right testicle 200cc

Legend: MRI T2 weighted imaging, appearances of the left and right testicles with tubular ectasia.

cc: cubic centimeter.

Validation, Investigation, Formal analysis, Data curation. **Steve McCombie:** Writing – review & editing, Supervision, Investigation. **Dickon Hayne:** Writing – review & editing, Validation, Supervision, Resources, Project administration, Conceptualization.

## Ethics

Reporting a review of patient clinical information as a case study or case series is considered “anecdotal” and usually involves retrospective data collection and analysis. In SMHS, case reports and case series are not classified as research, and therefore do not require SMHS Human Research Ethics Committee (HREC) approval to proceed.

## Footnote

Departmental approval was sought prior to publication and written informed consent has been obtained from the patient.

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## Glossary

**Tubular ectasia:** Tubular ectasia of the rete testes involves cystic dilatation of seminiferous tubules in the testicle

**Marfan syndrome:** A connective tissue disorder which is characterised by skeletal, cardiac, and ocular features

**Rete testes:** Collection of tubules in the hilum of the testicle linking the seminiferous tubules to the epididymis

**Massive::** Significantly above normal for the population

**MRI:** Magnetic resonance imaging

**Ductus:** Ductus deferens, duct which carries sperm from the testicle to the ejaculatory ducts

**MDT:** Multi-disciplinary meeting of urologists, radiologists, pathologists, oncologists, allied health and nursing staff