

Background

Men with Klinefelter Syndrome (KS) often present with infertility. However, the optimal management of this condition can involve many different specialties. This often results in major gaps and delays to the patient pathway, often with a lack of coordination in areas such as genetic & psychosexual counselling.

Aim

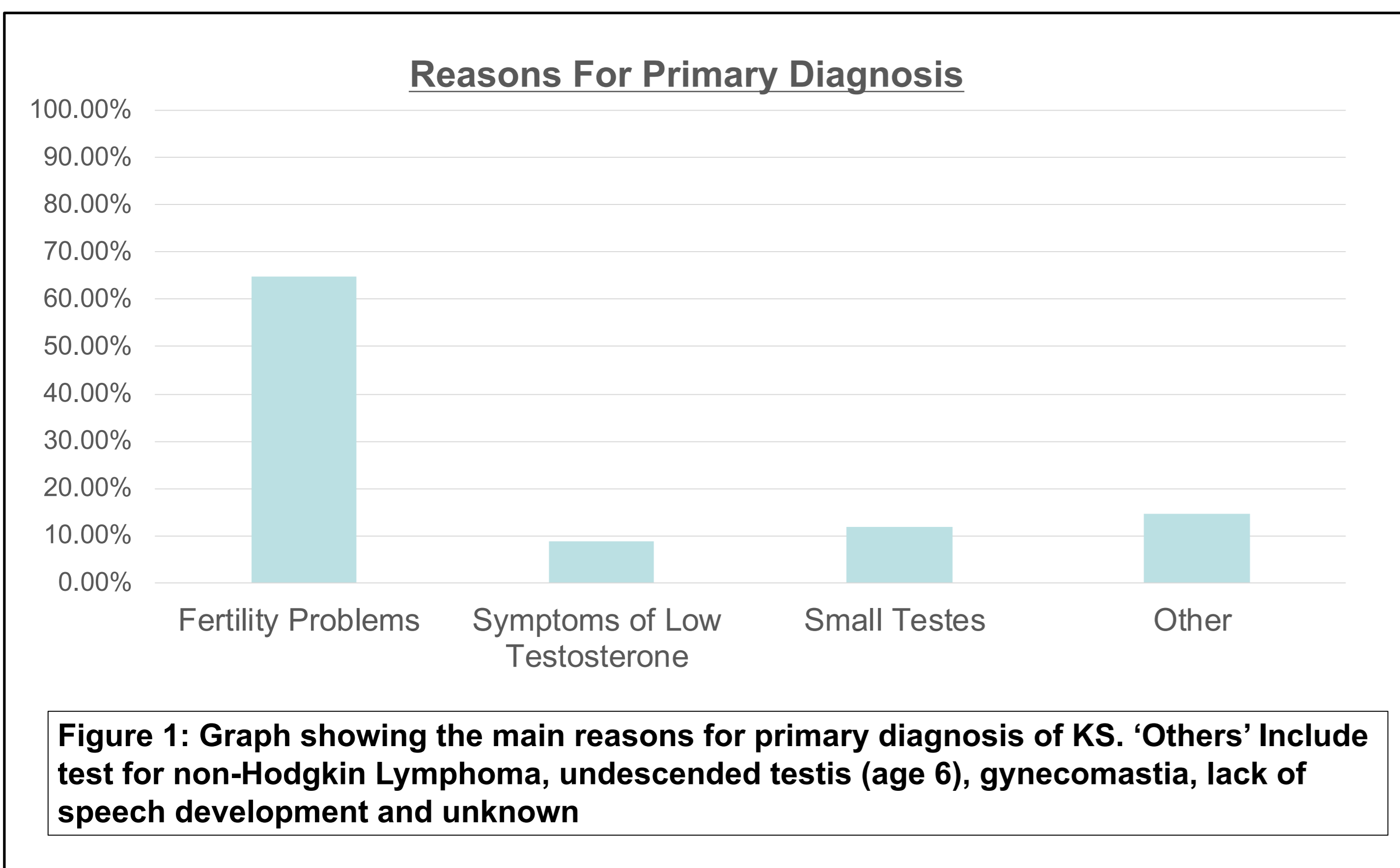
- National survey to assess needs of adult KS patients
- To set up one-stop multidisciplinary team (MDT) clinic model for KS

Methods

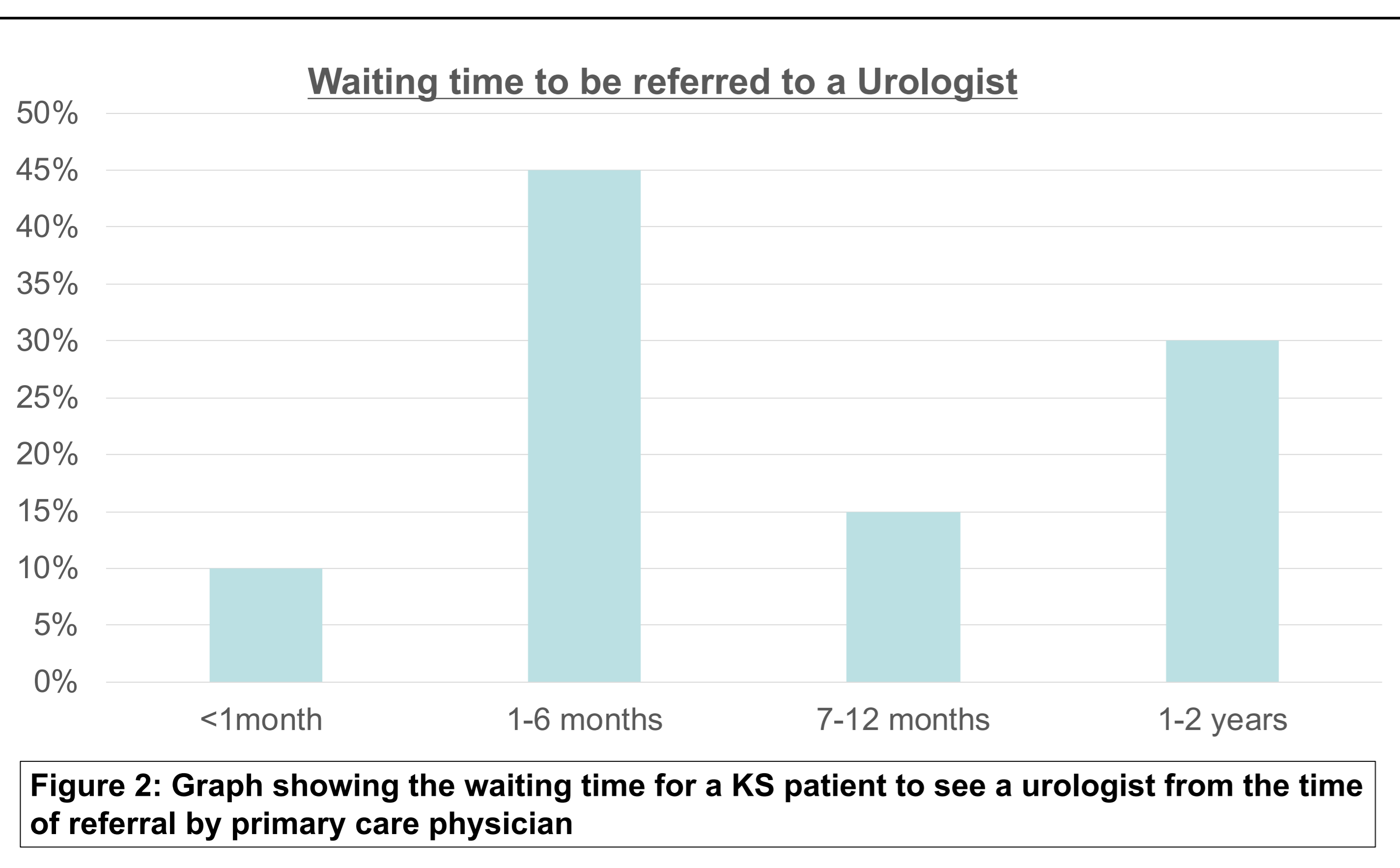
- Men with KS over age 18 who were members of the Klinefelter Association of the UK as well as current KS patients in three large teaching hospitals were invited to participate in the study.
- A detailed, anonymized online questionnaire was used to collect data on initial presentation, delays, and gaps in management and outcomes.
- Separately, an expert panel involving 7 specialties (Urology, Endocrinology, Genetics, Radiology, Reproductive Medicine, Embryology, Psychosexual Medicine), patient representatives, and allied specialties (pharmacy & nursing) was set up to identify the feasibility and benefits of an MDT clinic.
- Ethics approval was obtained from our local NHS board.

Results

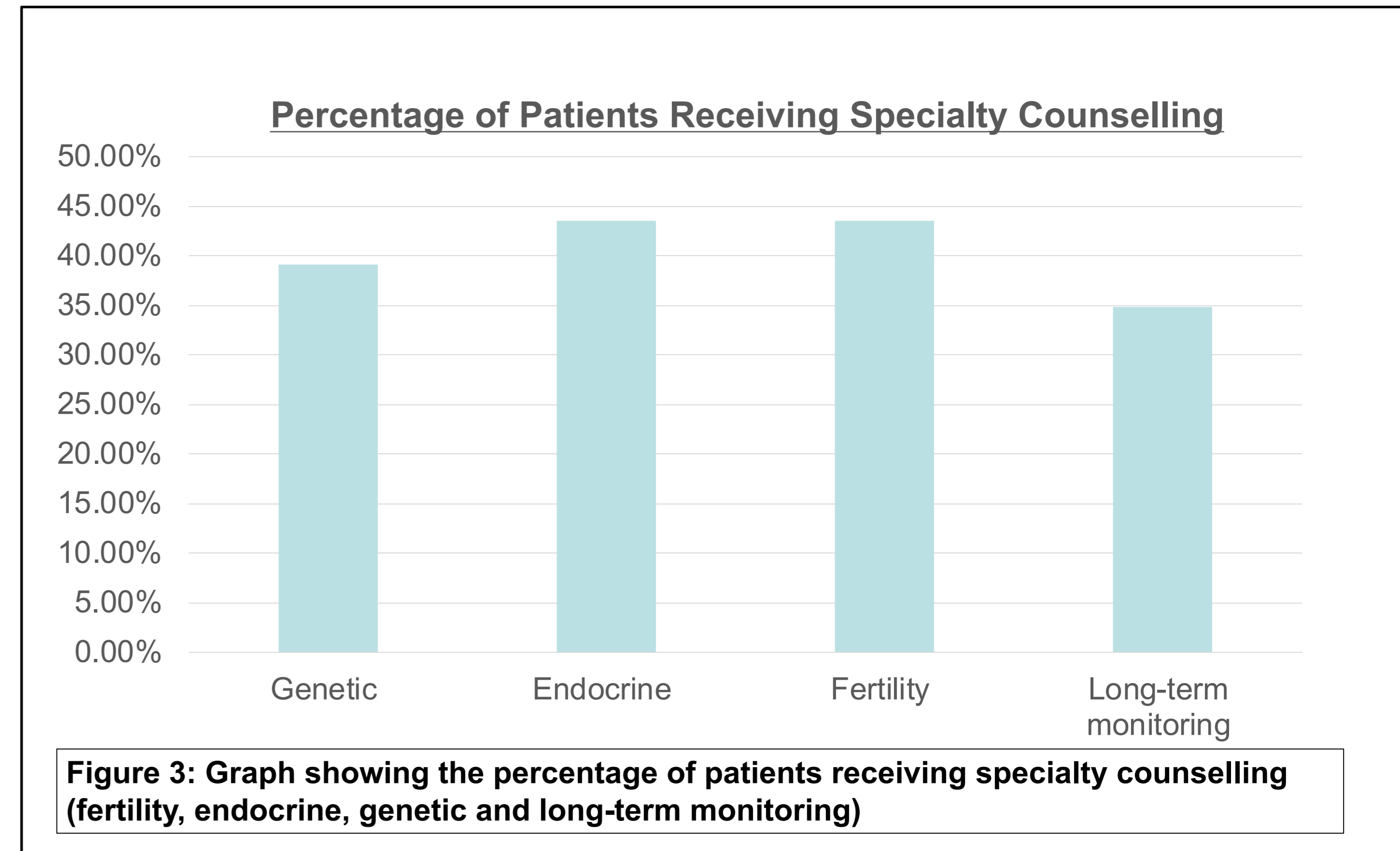
- 92 patients in total completed the online questionnaire



- most (64.7%) presented with infertility for further investigation
- Uncommon presentations include undescended testes, gynecomastia, lack of speech development, and 1 patient who was being investigated for Non-Hodgkin Lymphoma.



- 45% of patients waited a year or more in primary care before eventually being referred to a urologist, with most patients waiting 6 months to see a urologist from referral (Figure 2) with definitive fertility plan only started a mean of 1.5 years after initial diagnosis.



- Only 39% of patients had formal genetic counselling, whilst 35% had no long-term endocrine input.
- 87% were aware of surgical sperm retrieval, only 30% were offered Microsurgical testicular sperm extraction (microTESE).
- 89% felt that they would have benefited from seeing an endocrinologist (and 61% for psychologists) at diagnosis,
- Other key findings from our survey indicate that almost half of the cohort had noticed signs of KS by the age of 16, but the median age of diagnosis was 26-30 with more than 20% diagnosed after the age of 35 and only 14% diagnosed before the age of 16. This means that there is a significant delay from the initial presentation of KS to the diagnosis.
- With 86% strongly supporting an MDT approach. An MDT clinic approach would also decrease visit frequency, ensure a standard of care, and potentially reduce delays to definitive fertility management from 18 to 3 months.

Conclusions

The valuable information gathered about the delays and issues faced by adult men with Klinefelter Syndrome has led to the setting up of a one-stop MDT clinic encompassing 7 specialties with pharmacy & patient liaison services.

To date, 38 men with KS have now been seen by the MDT clinic, 70% of whom had primarily fertility issues. 12 clinicians involved in the running of the clinic were also interviewed. The clinic was very positively viewed based on patient and clinician feedback. All patient responded 'Yes' to the 5-point questionnaire:

- Did you feel the clinic was beneficial to your understanding of Klinefelter Syndrome?
- Did you feel the clinic was beneficial to the understanding of the management of Klinefelter Syndrome?
- Did you leave with a management plan that you clearly understood?
- Did you think the consultations were long enough?
- Did you feel the clinic addressed all your queries related to Klinefelter Syndrome?

The delays in seeing a fertility team from initial primary care presentations were reduced from 20 to 3 months, with the time to seeing the complete MDT team reduced from 32 to 3 months.

For Further Information

The Klinefelter Clinic:
<https://theklinefeltersyndromeclinic.com/>

The Klinefelter Association:
<https://www.ksa-uk.net/>

