Article:
Law, J.M., Amin, N., Ferguson, E.C. et al. (2026) A national survey of paediatric Turner syndrome services in the UK: current practice and variability in care. Hormone Research in Paediatrics. ISSN: 1663-2818

https://doi.org/10.1159/000550412

Hormone Research in Paediatrics

Research Article

Horm Res Paediatr
DOI: 10.1159/000550412

Received: October 6, 2025
Accepted: December 29, 2025
Published online: January 9, 2026

A National Survey of Paediatric Turner Syndrome Services in the UK: Current Practice and Variability in Care

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Keywords
Turner syndrome · Paediatric endocrinology · Multidisciplinary care · UK survey · Guideline adherence · Transition to adult care

Abstract

Introduction: Turner syndrome (TS) is a complex genetic condition requiring lifelong, multidisciplinary care. International consensus guidelines exist, but the organisation of paediatric TS services in the UK has not been systematically explored. ***Methods:*** A structured electronic survey was distributed to paediatric endocrinology centres across the UK with responses collected from June 2023 to February 2024. The survey collected information on service configuration, staffing, multidisciplinary team (MDT) composition, transition pathways, use of consensus guidelines, and engagement with patient registries and support societies. ***Results:*** Responses were received from 20 UK tertiary centres. Six out of 20 centres operated a dedicated TS clinic. MDTs were limited in most centres to paediatric endocrine consultants and nurse specialists, and shared care models for outreach patients were common. Transition practices varied, with 45% of centres using TS-specific pathways, 45% using general endocrine transition pathways, and 10% without a transition pathway. Awareness of international TS guidelines, the Turner Syndrome Support Society, and the i-TS registry was high, but active engagement varied. ***Conclusion:*** Significant variability exists in UK paediatric TS service models. Centres without dedicated clinics were generally smaller with fewer patients. Geographic challenges may exacerbate inequalities for outreach patients. While some centres offer best practice examples, improvements in MDT availability, transition planning, and registry engagement are needed to align more closely with international care recommendations.

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Plain Language Summary

Turner syndrome is a lifelong condition that affects girls and women when one of their X chromosomes is missing or changed. It can lead to short height, fertility difficulties, heart and kidney problems, and challenges with hearing and learning. Because it can affect many parts of the body, guidelines recommend that care for women with Turner syndrome should be provided by several different specialists working together. We wanted to understand how care for girls with Turner syndrome is organised across the UK. To do this, we sent a national survey to children's hospitals that look after patients with hormone-related conditions. We asked about how many patients they cared for, which health professionals were involved, how often girls were seen, and what happens when they move from children's to adult services. Twenty hospitals replied. Only six had a separate clinic just for girls with Turner syndrome. In most centres, patients were seen by a hormone-specialist doctor and a specialist nurse, but many did not have other team members, such as psychologists or gynaecologists. Some hospitals had well-organised plans for helping young people move to adult care, but others had no formal pathway. Travel distance and limited staff were common barriers, especially for families living further away. The survey showed that services differ across the country. Some centres offer excellent, team-based care, while others have gaps that may affect long-term health. More consistent services could help every girl with Turner syndrome receive the same high-quality support.

Introduction

Turner syndrome (TS) is a chromosomal condition characterised by partial or complete monosomy of the X chromosome [1, 2], affecting approximately 1 in 2,000 live-born females [3]. It is associated with a broad and complex spectrum of clinical manifestations, including short stature, ovarian insufficiency, congenital heart defects, renal anomalies, hearing difficulties, and various neurocognitive and psychosocial challenges [4–6]. These lifelong, diverse health needs require coordinated multidisciplinary medical care, beginning in childhood and continuing into adulthood.

Given the range and progression of health concerns in TS, the importance of structured, lifelong medical surveillance and multidisciplinary team (MDT) involvement has been recognised consistently in consensus guidelines [2, 3, 7]. In 2017, the Turner Syndrome Consensus Group published updated international clinical practice guidelines, providing detailed recommendations for the diagnosis, surveillance, and management of TS from infancy through adulthood [2] that were further updated in 2024 [3] subsequent to the survey reported here being undertaken by respondents. These guidelines advocate for MDT involvement, including endocrinologists, cardiologists, psychologists, audiologists, fertility experts, and others, as well as dedicated care pathways and planned transition to adult services.

Despite these recommendations, previous studies have identified considerable variability in TS care provision both internationally and within the UK [8, 9]. While the NHS provides universal healthcare access, there is no nationally standardised TS service model for children and young people, and limited published data exist on how services are structured across regions, contributing to regional differences in care quality, access to specialist teams, and transition planning. A 2019 survey by the Turner Syndrome Support Society (TSSS UK, https://tss.org.uk) highlighted disparities in access to dedicated TS clinics, variable transition support, and inconsistent referral to psychological and fertility services. However, a comprehensive overview of service organisation, professional roles, guideline adherence, and participation in national and international initiatives has not been previously published.

To address this gap, we conducted a UK-wide survey of tertiary paediatric endocrinology centres providing care for girls with TS. The survey sought to capture the current landscape of service provision, including patient numbers, clinic structures, MDT composition, implementation of the 2017 guidelines, signposting of the TSSS, transition pathways to adult care, and participation in the international i-TS registry (sdmregistries.org/i-ts).

This study presents the results of that survey, providing the first comprehensive national overview of paediatric TS services in the UK. Our findings aim to inform clinicians, policymakers, and advocacy groups in efforts to promote more consistent, evidence-based, and equitable care for girls and women with TS.

Methods

Survey Development and Content

A structured electronic survey was developed. An iterative approach was adopted by the team to refine questions ensuring high readability, clarity of meaning, and relevance (online suppl. Material 1; for all online suppl. material, see https://doi.org/10.1159/000550412).

Table 1. Domains covered in the survey of TS services

Domain	Description
Clinic size	Number of consultants, WTE consultants, patients seen
Consultant involvement	Number of consultants specifically managing TS patients
Clinic configuration	Dedicated TS clinic status and reasons for absence
Review frequency	By age group and by clinic type (TS-specific or general endocrine)
MDT composition	Disciplines present in clinic
Local vs. regional service differences	Perceived and described differences in care models
Transition	Presence of formal pathway, referral sites after transition
Best practice and guidelines	Use of care pathways, consensus guidelines, i-TS registry, and TSSS referral
Barriers and needs	Free-text questions on care limitations and research priorities

Questions were reviewed by the TSSS and approved by the British Society for Paediatric Endocrinology and Diabetes (BSPED) Clinical Committee prior to circulation. The survey comprised both quantitative and qualitative items and aimed to capture a comprehensive picture of current UK paediatric TS services. Topics included service size, consultant and MDT staffing, presence and structure of dedicated TS clinics, frequency of clinic attendance by age group, alignment with the 2017 International Turner Syndrome Consensus Guidelines, use of clinical pathways, signposting to the TSSS, transition arrangements to adult services, and awareness or participation in the international i-TS registry (Table 1). Free-text questions invited respondents to share barriers to care, examples of best practice, and priorities for future research.

Participants and Recruitment

All tertiary paediatric endocrinology centres in the UK were eligible to participate. Dissemination was through the BSPED newsletter, and direct invitations were sent by email to service leads, with follow-up contact if required. Responses were collected between June 2023 and February 2024. Response rate was calculated using the BSPED list of paediatric endocrine centres as the denominator [10]. Participation was voluntary, and participants did not have to respond to all questions. Responses were pseudonymised by assigning random centre IDs for data handling and presentation.

Data Handling and Analysis

Survey responses were collected through an online survey tool (www.jotform.com) and exported to Microsoft Excel. Quantitative data were analysed and summarised using Microsoft Excel. Numerical variables are presented as medians with ranges (for consultant numbers) or IQR. Categorical data are summarised as frequencies and percentages. Free-text responses were analysed to identify recurring themes relating to service organisation, barriers to care, and opportunities for improvement. Where centres did not provide a response, they were excluded from analysis of that question. Therefore, the number of respondents varied between questions.

Terminology

Tertiary centres are defined according to the BSPED list of paediatric endocrine centres [10]. Local patients are defined as those for whom the tertiary centre is based at their local hospital. Regional patients are those for whom the tertiary centre is based remotely compared to their local hospital.

Results

Clinic Size and Staffing

Responses were received from 20 tertiary paediatric endocrinology centres across the UK (online suppl. Material 2), representing over 90% of UK tertiary centres [10]. Centres employ between 2 and 12 individual consultants in paediatric endocrinology (median 5, 20 centres), with 2.7 (1–7) whole time equivalent (WTE) consultants (18 centres). In 19 centres that responded, the number of consultants directly involved in seeing patients with TS ranges from 1 to 8 (median 4), representing the majority of consultants (77%). In 10/19 centres, patients with TS are seen by all consultant paediatric endocrinologists, while in 2/19 centres,

Table 2. Frequency of clinic visits (per year) by age group and whether the centre has a dedicated TS clinic

	Clinic visits per year	Under 5s	5–11s	12 and over
TS clinic	2	2/4	4/5	5/6
	3 or more	2/4	1/5	1/6
No TS clinic	2	9/12	10/12	10/12
	3 or more	3/12	2/12	2/12

Values are reported as number of centres with this frequency/total number of centres responding to the question.

patients with TS are seen by one consultant (representing 20% and 25% of consultants in those two centres).

Sixteen centres gave either precise (5 centres) or estimated numbers (11 centres) of patients with TS under their care, with 4 unable to provide numbers. Reported patient numbers range from 10 to 138 (median 30, IQR 23.5–44). The number of patients per consultant varies widely with a median of 5.5 (IQR 4.3–10.3) or 10 (IQR 5–20) per WTE consultant.

Clinic Structure and MDT Composition

Six centres have a dedicated TS clinic, while 14 do not. Centres with a dedicated TS clinic care for 45 (IQR 23.5–91.5) patients with TS per centre (13.6 [IQR 7.2–32.7] per WTE consultant), while those without care for 30 (IQR 22.8–35) (8.8 [IQR 4.9–15.9] per WTE consultant). In centres without a dedicated clinic, the most common reason cited for this is limited clinical capacity (8/14). Other reasons include small patient numbers (4/14), not desired by clinicians (3/14), financial constraints (2/14), geographical constraints, patient preference/ability to travel (2/14), or logistical difficulties (2/14).

A recurring theme was difficulty accessing members of the MDT. Common limitations included the following:

- Psychology services: Six centres reported no dedicated psychology input or inconsistent access as a key barrier to optimal patient management. The value of in-clinic psychology was strongly emphasised.
- Gynaecology, ENT, dietetics: Gaps in timely access to gynaecology and ENT services were common. Some centres relied on sporadic or remote referrals.
- Examples of positive MDT practice: Some centres reported success through the integration of adolescent gynaecology, adult endocrinology, cardiology, genetics, and fertility teams into joint or cohort clinics.

All centres (20/20) have a paediatric endocrinologist present in their clinic. A paediatric endocrine clinical nurse specialist is present in clinic in all centres with a specialised TS clinic, and 11/14 without. Other team members such as adult endocrinologists (1 centre), gynaecologists/fertility specialists (1 centre), general paediatricians with an interest (2 centres), psychologists (2 centres), and dietitians (1 centre) were available in some centres, with no centre having more than one such further member of the MDT present in clinic.

Centres usually see patients twice a year. This happens usually in a dedicated clinic, where available, with some clinics seeing patients more frequently (Table 2).

Local versus Regional Care Variation

Several models of care for regional patients were described, including the following:

- Tertiary-centric models: Some centres offer annual reviews in a tertiary centre with interim care delivered locally.
- Outreach and shared care clinics: Others operate through specialist outreach clinics involving paediatric endocrinologists and local general paediatricians with an interest in endocrinology.
- Clinic type variability: A mix of dedicated TS clinics and general endocrine clinics were reported. Variations extend to access to ENT and cardiology follow-ups, which are sometimes conducted locally.

The majority of respondents felt the care given to patients with TS who live outside the local area does not differ from that given to local patients (7/20), but 4/6 respondents from centres with a dedicated clinic felt it did. Several centres reported that patients outside the local area were seen annually by a tertiary endocrinologist, with review by a local paediatrician with an interest in between. It was commented that other members of the MDT would be less likely to be at outreach clinics and patients may need to travel to the tertiary centre to be seen by them.

Transition and Adult Care

Regarding transition of patients with TS, 9 centres used a generic endocrine transition pathway, 9 centres had a pathway specific to patients with TS, and 2 had no

transition pathway. Those with a dedicated clinic were more likely to have a TS-specific transition pathway (2 general, 4 specific) compared to centres without a dedicated clinic (7 general, 5 specific, 2 no pathway).

Once under adult services, patients with TS are cared for in a variety of settings, with some receiving care in more than one setting. Thirteen of 20 centres could refer to a specific adult TS clinic, 9 to a general endocrine clinic, and 2 to an adult gynaecology (one complex gynaecology service, one service with paediatric and adolescent gynaecologists and adult endocrinologists).

Implementation of Best Practices

All respondents were aware of the TS International Consensus Guidelines and reported working to them. Similarly, all centres knew about the TSSS and routinely informed families about it. Seven centres reported having a departmental clinical care pathway for patients with TS, while 13 did not. Centres shared innovative practices with potential for replication.

- Specific TS-cohorted clinics to foster continuity and peer support.
- Structured transition models, including joint clinics with specialists such as cardiology across the lifespan.
- Clinical methods, such as early AMH assessment, ovarian tissue cryopreservation, calm-setting BP measurements, and centralised GH prescribing, were felt to optimise patient care by individual centres.

Nineteen of 20 centres were aware of the i-TS registry (https://sdmregistries.org/i-ts/), but only 5 were already participating, with a further 11 planning or considering participation. Reported barriers included lack of administrative support, time constraints, and challenges obtaining consent and navigating governance frameworks.

Several centres expressed interest in feedback and centre-specific benchmarking to compare service models and outcomes. Key barriers to optimal care included the following:

- Lack of research infrastructure: Update of the i-TS registry was hindered by time constraints and lack of administrative support (cited by 9 respondents). Time constraints for consenting patients within clinics were cited by 2 centres, and local research governance issues were cited by 2 centres.
- Clinic capacity: Overbooked clinics and limited clinician availability reduced appointment time, and service quality was referred to by 13 centres. A lack of psychology support was specifically mentioned by 9 centres.
- Medication and intervention access: Challenges were noted in accessing pubertal induction medications and weight management interventions.
- Travel and geography: Families in large geographic regions faced cost and logistical challenges in accessing tertiary care.

Research Priorities

Respondents identified several key areas for future research.

- Fertility: A key theme, mentioned by 10 centres, was research around fertility in patients with TS, including optimising fertility prospects and techniques.
- Puberty: Optimal timing and method for puberty induction were cited as a key research question by 6 centres.
- Cardiovascular health: There was recognition by 4 centres of the need for research into long-term strategies to manage BMI and cardiovascular risk.
- Mental health and learning: Better understanding of cognitive and psychosocial impacts, including karyotype correlations by 4 centres.
- Service evaluation: Long-term outcomes of patients managed through dedicated TS services were cited as an area for future research by one centre.

Discussion

This national survey highlights significant variation in the organisation and delivery of paediatric TS services across the UK. Although the current study was undertaken prior to the revised 2024 consensus guidelines being published, the most recent guidelines further strengthen the importance of both the MDT approach and high-quality transition processes [3].

While some centres have developed structured, multidisciplinary models of care in line with international guidelines, many do not have a dedicated TS clinic. These centres typically care for smaller patient populations and have more limited consultant and MDT resources.

In most centres, the clinical team is limited to a paediatric endocrinologist and paediatric endocrine clinical nurse specialist, with few including additional professionals such as psychologists, gynaecologists, or dietitians. This contrasts with the International Turner Syndrome Consensus Guidelines [2], which advocate for a comprehensive, coordinated approach to care involving multiple specialties. The limited access to MDT input, particularly in general endocrine clinics and in the outreach setting, may limit access to the holistic support that patients with TS require, although outreach clinics are an important factor in providing equitable healthcare, particularly in remote areas [11]. The frequency of patient reviews varied across centres and age groups, with most attending clinic twice

per year. Some centres offered more frequent visits, particularly during adolescence.

Patients living at a distance from tertiary centres may be reviewed less frequently by endocrinologists and have reduced access to other team members. Although shared care models with local paediatricians exist, the effectiveness of such arrangements in providing guideline-concordant care would benefit from further review.

It has been shown in several areas that clear, coordinated transition plans improve clinic attendance and health outcomes, including in type 1 diabetes and congenital adrenal hyperplasia [12–15] yet many services still lack formalised pathways. In TS specifically, structured transition involving both paediatric and adult endocrinologists, alongside reproductive and psychological support, is recommended but infrequently achieved in practice [16]. Similarly, in our study, transition practices were highly varied, with some centres able to provide exemplary models of care through dedicated TS transition pathways and joint clinics with a broad range of relevant adult services. Others had limited or no transition pathways in place, potentially leaving young people vulnerable during a critical developmental period [17]. This variability reflects challenges seen across endocrine and chronic conditions, where structured transition remains inconsistently implemented [18, 19].

Examples of good practice – such as cohort clinics, inclusion of adolescent gynaecology, and early fertility discussions – were reported by several centres. However, substantial barriers persist, including lack of administrative support for the i-TS registry [2], clinical capacity issues, limited access to puberty induction therapies, and significant travel burden for families.

While nearly all centres reported adherence to the International Turner Syndrome Consensus Guidelines and routinely informed families about the TSSS, uptake of formal care pathways and registry participation was limited. There is a clear opportunity to improve national alignment with guideline-based care, ensure equity of access to MDT services, and expand best practices more widely.

Using the BSPED definition, responses were obtained from most paediatric endocrine centres and provide a broad representation of services across the UK. Services not involving paediatric endocrinologists were not identified due to the distribution and contact methods adopted. Given being based on a survey, our results rely on self-reported data from clinicians, which may be subject to reporting bias or inaccuracies. Furthermore, responses were obtained from a single person at each centre. While this was someone with comprehensive knowledge of the service, such as the head of service or lead for TS, their views may not fully reflect the practices or opinions of the rest of the team. To maximise response rates, a minimal subset of care questions was made mandatory, which inevitably led to some missing data. It was not feasible within the current work to enquire about adherence to specific recommendations within the Consensus Guidelines or to explore whether particular characteristics of centres were associated with closer alignment to them. Similarly, given the number of variables investigated and the small numbers of certain responses (such as wider MDT composition), it was not feasible to draw wider inferences.

These results provide a snapshot of reported clinical services for patients with TS seen in tertiary paediatric endocrine services across the UK. Further work is needed to provide the patient perspective on services and identify if there are gaps between clinician and patient and carer/parental perceptions. This survey underscores the need for national benchmarking, clearer care standards, and investment in MDT infrastructure to reduce variability and support girls with TS in achieving optimal long-term outcomes.

Acknowledgments

ChatGPT (OpenAI, San Francisco, CA) [20] was used to assist with drafting and editing portions of the manuscript. All content was reviewed, verified, and finalised by the authors.

Statement of Ethics

The survey was classified as a service evaluation and did not require formal research Ethics Committee approval in accordance with NHS Health Research Authority guidance. Written informed consent to participate was not directly obtained but inferred by completion of the questionnaire.

Conflict of Interest Statement

James M. Law has received sponsorship to attend conferences from Kyowa Kirin, Pfizer, and Novo Nordisk. Nadia Amin has received speaker fees from Pfizer and sponsorship to attend conferences from Novo Nordisk, Kyowa Kirin, Merck, and Pfizer. Elspeth Ferguson has no conflicts of interest. Jan Idkowiak has received grants from NIHR, ESPE, and Birmingham Children's Charity; and consultancy fees from Novo Nordisk and Egetis Pharma. Sasha R. Howard has received grants from the NIHR, Wellcome Trust, Barts Charity, British Council, Rosetrees Trust, and ESPE and speaker fees from Sandoz, Pfizer, Novo Nordisk, and Merck. Harshini Katugampola has received grants from

MRC, BSPED, and Rosetrees Trust. Nils Krone has received speaker fees from Neurocrine Biosciences, Sandoz, and Novo Nordisk; consultancy honoraria from Neurocrine Biosciences; royalties from Wolters Kluwer; and grants from International Funding CAH, ESPE, NIHR, and Neurocrine Biosciences (investigator-initiated trial).

Funding Sources

This study was not supported by any sponsor or funder.

Author Contributions

J.M.L. designed the work, acquired, analysed and interpreted the data, drafted the work, and approved the final version. N.A. and E.C.F. designed the work, acquired, analysed and interpreted the data, reviewed the work, and approved the final version. J.I., S.R.H., and H.K. designed the work, acquired the data, reviewed the work, and approved the final version. N.P.K. designed the work, reviewed the work, and approved the final version.

Data Availability Statement

All relevant data generated and analysed during this study are included in the published article. The survey was undertaken as a national service evaluation, and respondents did not provide consent for public data sharing. The full dataset from the national survey contains identifiable information about clinical centres which cannot be shared publicly for reasons of confidentiality. The dataset is stored securely by the corresponding author. An anonymised version of the dataset will be made available to individuals or groups upon reasonable request to the corresponding author, subject to review and in accordance with data sharing on a case-by-case basis.

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