Diagnostic Disclosure in Turner Syndrome: A Qualitative Exploration of the Experiences and Needs of Individuals living with Turner Syndrome and Family Members.

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Diagnostic Disclosure in Turner Syndrome: A Qualitative Exploration of the Experiences and Needs of Individuals living with Turner Syndrome and Family Members.

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Declaration

I hereby state that the work presented in this thesis is my own and reference to other work has been cited with full details. No part of this thesis has been submitted for any other qualification at another university.

Miss Emma Clarke

Signature: E. Clarke Date: 12/12/2022

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Contents

General abstract

Background: Approximately 7,000 genetic conditions affect 1-2% of the UK population. Studies on the biological causes abound in the literature, whereas research on the psychosocial impact remains scarce. Diagnostic disclosure has been gaining attention due to the significant impact it can have on those affected by genetic conditions. This thesis focuses on the diagnostic disclosure of genetic conditions and places a particular focus on Turner Syndrome (TS), which affects approximately 1 in 2,500 live female births. The author of this thesis is an individual living with TS and carried a special interest in exploring this area topic.

Aims and objectives: First, we aimed to map current knowledge on experiences of diagnostic disclosure of genetic conditions, from the perspectives of those affected by diagnosis. We focused broadly on genetic conditions due to the significant paucity of studies in TS. Second, we aimed to explore the experiences and needs of individuals living with TS and their parents around diagnostic disclosure. The final aim of the study was to provide recommendations for future researchers by critically reflecting on lessons learnt from conducting a study on a condition of which the author has lived experience.

Methods: A mapping review was conducted through a systematic search for peer-reviewed studies in 6 electronic databases. Semi-structured interviews were then carried out with 16 individuals living with TS and 8 parents. Data were transcribed verbatim and analysed using inductive thematic analysis. Finally, the author provided critical reflections on her role as a researcher with lived experience of TS and how this may have influenced the research processes of the original study. Emphasis was placed on the author's positionality, prior knowledge and experience of disclosure, and the emotional impact of studying her own condition.

Results: Findings from 12 studies included in the mapping review showed that openness and gradual information sharing facilitated disclosure and adapting to living with a genetic condition. Collaborative approaches between healthcare professionals and service recipients enabled positive experiences of disclosure, while fear of stigma acted as a barrier. Analysis of interviews yielded 3 major themes representing

participants' experiences and needs: 'Guardianship of disclosure', 'Coping with infertility' and 'Awareness of Turner Syndrome and its impact'. Individuals with TS preferred to learn about the diagnosis and take ownership of disclosure early on. Fear of the impact of infertility prevented parents from disclosing the diagnosis to their children. Strengths-based approaches may prevent stigma and enable those affected to receive empowering advice and support. The author's reflections indicate that the 'total insider' position may need to be negotiated with participants before being established. Reflexivity and PPI input are necessary to ensure rigor and representativeness of findings. Pastoral and peer support may facilitate maintaining the researchers' emotional wellbeing when conducting research into one's own condition.

Conclusions: Our findings provide novel insights into the condition-specific context of TS diagnostic disclosure and can inform recommendations for healthcare professionals and third-sector organisations. The lessons learnt can be useful for researchers conducting qualitative research in a condition of which they have lived experience.

General introduction

Genetic conditions are defined as medical conditions which result from mutations or changes within the DNA of an individual. The prevalence of genetic conditions is estimated to be 1-2% in the UK, with 1 in 12 individuals affected by a diagnosis (Public Health Action Support Team, PHAST, 2020). Down Syndrome and Cystic Fibrosis are two well-known examples of genetic conditions, which are approximately 7,000 in total (Milunsky and Milunsky, 2015; Genetic Alliance UK, 2016). During the last few decades, scientific advances in the use of more effective diagnostic tools and processes have resulted in improved diagnostic rates, showing that genetic conditions are more frequent than once thought.

This thesis is focused on Turner Syndrome, which is a genetic condition affecting approximately 1 in 2,500 live female births (Hutaff-Lee et al., 2019). The condition is caused by a genetic abnormality which is characterized by the complete or partial absence of a X chromosome (Ackermann and Bamba, 2014). Common symptoms of Turner Syndrome include short stature and webbed neck, with the possibility of co-morbidity alongside other medical or health conditions, such as heart or kidney diseases (Reimann et al., 2018). Similar to other genetic disorders, studies on the genetic causes and biological mechanisms involved in the development of Turner Syndrome abound in the literature. Notwithstanding the significance of the relevant knowledge, there is a noticeable paucity of studies on the psychosocial impact of the condition, particularly diagnostic disclosure (Gallo et al., 2005; Sandberg et al., 2019; Sutton et al., 2006). Currently, approximately 91% of published literature focuses on the genetic aetiology of the condition, whereas only 9% explores the psychological impact of living with Turner Syndrome (Sandberg et al., 2019).

Diagnostic disclosure has been gaining attention during the recent years due to reports of the significant impact it can have on those living with the condition and their families (Sutton et al., 2006). It is broadly defined as the process of discussing or sharing information about one's condition with range of people, including healthcare professionals and significant others in a social context (Munro et al., 2015). Previous studies have shown that the experience of diagnostic disclosure can influence the way individuals living with a genetic condition and their parents cope with diagnosis. For instance, children that receive full details about the condition and its symptoms tend to display better coping skills and less psychological challenges (Gallo et al., 2005).

Negative experiences of disclosure, such as lack of empathy or information from healthcare professionals, can negatively influence parents and their decision about when or how to disclose information about the condition to their children (Goodwin et al., 2014). This becomes particularly important when considering the additional impact this may have on treatment adherence (WHO, 2011).

Research in diagnostic disclosure of Turner Syndrome is currently very scarce. There have only been two studies exclusively focusing on this topic area (Sutton et al., 2006; Nisbet et al., 2022) which presented contradictory findings. As a result, there are no condition-specific guidelines for diagnostic disclosure of Turner Syndrome, with professionals and parents making the relevant decisions based on generic guidance on the disclosure of genetic conditions. The latter primarily suggests that diagnostic disclosure should be a gradual process (Middleton et al., 2018) and that the individual's developmental stage should be considered in the process (Gallo et al.,2005). However, the experience of genetic disorders may significantly vary across individuals and families affected by different genetic conditions, which implies that their needs around diagnostic disclosure may be different. Furthermore, the aforementioned guidelines are largely informed by research with parents of children diagnosed with genetic conditions and rarely represent the voices of those living with the diagnosis. It is, therefore, important to acquire insight into the unique experiences of all those affected by Turner Syndrome and ensure that evidence-based recommendations reflect the experiences and needs of those living with the diagnosis.

The current thesis aimed to address current gaps in knowledge by: (a) critically outlining current evidence on the experience of diagnostic disclosure of genetic conditions, (b) exploring in depth the diagnostic disclosure experiences of individuals living with Turner Syndrome and their parents, (c) identifying the barriers and facilitators involved in diagnostic disclosure of Turner Syndrome, from the perspectives of those affected by the condition, and (d) contributing to condition-specific recommendations on diagnostic disclosure of Turner Syndrome. The thesis comprises three chapters to address the aims stated above. In the first chapter, a systematic mapping review was conducted to map current knowledge on the experience of diagnostic disclosure of genetic conditions, from the perspectives of those affected by genetic disorders, and identify potential gaps to inform future research. In the second chapter, an original study was carried out to explore the experiences and needs of individuals living with Turner Syndrome and their parents

around diagnostic disclosure. This study further aimed to identify facilitators and barriers to diagnostic disclosure, from the perspectives of those affected by Turner Syndrome, and contribute to condition-specific recommendations by adding to the evidence base. The last chapter comprises a paper with the author's reflections on lessons learnt from the delivery of the study, to provide recommendations for future researchers who may also be considering conducting studies on a condition of which they have lived experience. As the author of this thesis also lives with Turner Syndrome, it was considered important to critically discuss and reflect on her dual role and how this may have influenced the research processes of the study.

Overall, it is the author's hope that this thesis will contribute to a better understanding of diagnostic disclosure of Turner Syndrome and the development of condition-specific needs-based recommendations, which will raise awareness among healthcare professionals and families affected by the condition and will facilitate positive experiences of disclosure for those diagnosed in the future.

Chapter 1. Diagnostic disclosure of genetic disorders: A mapping review of the experiences of individuals living with genetic conditions and family members

1. Introduction

A genetic condition is defined as a medical condition or disorder caused by changes or 'mutations' to an individual's DNA. Some examples of genetic disorders include Down Syndrome and Cystic Fibrosis (Genetic Alliance UK, 2016). Genetic disorders are quite common with a prevalence of approximately 1-2% in the UK (Public Health Action Support Team, PHAST, 2020). Additionally, 1 in 12 individuals are affected by (whether aware or unaware of) a diagnosis of a genetic condition, including one of over 7,000 known rare genetic disorders (Milunsky and Milunsky, 2015).

Disclosure is defined as the extent to which individuals discuss a diagnosis and the associated thoughts and feelings openly with a range of people (Munro et al, 2015). The experience of disclosure is significant because it can influence how the individual and family cope with the disclosed information and living with the diagnosis (Nisbet et al., 2022). For example, children who receive full information about their condition, including the symptoms and consequences, display better coping skills and less psychological problems than those who do not (Gallo, et al; 2005). In contrast, a diagnosis conveyed in a negative way or without consideration of the individual's needs can negatively impact their self-esteem and self-concept (Krieger, 2001). A negative diagnostic disclosure to the parent may also negatively influence the subsequent disclosure to their child, as parents become confused or unsure of how and when to disclose information about the diagnosis to the individual living with the condition (Goodwin et al., 2014). This indicates the importance of addressing the needs of all those affected by the diagnosis during disclosure by healthcare professionals and parents.

Previous research into the disclosure of genetic conditions has informed the development of relevant guidelines (WHO, 2011). For instance, research into the experience of disclosure for individuals living with dementia highlighted the fear of stigma resulting from diagnosis and its role as a barrier to disclosure (O'Connor et al., 2018). Studies on diagnostic disclosure by parents to their children living with autism highlighted the importance of open communication, disclosure being a gradual learning process, and considering the individual's level of understanding (Crane et al., 2019). The guidelines focus on the needs of those affected by the diagnosis of a genetic condition and emphasise the benefits of a positive needs-based diagnostic

disclosure, which include reassurance about living with the condition and access to needs-appropriate post-diagnostic support (Munro et al., 2015), increased sense of empowerment (O'Connor et al., 2018), and enhanced treatment adherence (WHO, 2011).

Although previous research has provided insights into some aspects of good practice in diagnostic disclosure of genetic conditions such as HIV (WHO, 2011), the needs of individuals and family members affected by other genetic conditions, such as Turner Syndrome, remain largely unexplored. Previous studies have provided general suggestions such as considering the individual's level of cognitive development (Gallo et al., 2005) and the need for a gradual sharing of information about the diagnosis (Middleton et al., 2018). Family members' sense of guilt and their concerns about the psychosocial impact of disclosure to the person living with the condition have also been highlighted (Middleton et al., 2018). However, evidence-informed recommendations on diagnostic disclosure from parents to children remains scarce (Gallo et al., 2005). This review aimed to map current evidence available in the literature regarding the experience of diagnostic disclosure of genetic conditions for family members and individuals living with the condition, to identify potential gaps in knowledge and inform future research.

2. Methods

A mapping review aims to present existing evidence on a selected topic to describe the level of relevant knowledge (James et al., (2016) and highlight gaps that need to be addressed through further research (Clapton et al., 2009). The results of a mapping review summarise current knowledge on a given area topic and provide a useful point of reference for researchers, funders and policy makers, to highlight important issues requiring further attention and inform future research (Bates et al., 2007). The latter constituted our main aim for this review due to the scarcity of evidence on the impact of diagnostic disclosure and the needs of those affected by genetic conditions during disclosure. We purposively selected a systematic mapping review method to enhance the rigour of our findings through the quality appraisal of the studies and evidence reviewed (Grant and Booth, 2009), which is usually optional when conducting mapping reviews (James et al., 2016).

A mapping review was selected compared to, for example, an Umbrella review, due to its suitability to address the review question. An Umbrella review is similar to a mapping review because it shares the aim to address specific questions within a broad problem or condition and provide an overview of the research topic. However, an umbrella review requires an exhaustive search and comparison of systematic reviews within the topic being studied. This was not suitable for the current thesis because, according to the researcher, there were no reviews available within the topic of diagnostic disclosure. Therefore, it was not possible for this approach to be used. (Grant and Booth, 2009).

2.1 Inclusion and exclusion criteria

The review aimed to include studies of any design focusing on the diagnostic disclosure of genetic conditions through the perspectives of parents/family members, individuals living with the diagnosis, and/or healthcare professionals. To be included in this review, studies had to be peer-reviewed and published in English. Grey literature and studies focusing on genetic testing, genetic counselling, ethics of disclosure (if this was the major focus), and general physical conditions were excluded from this review.

2.2 Search strategy

A search strategy was developed by the research team in collaboration with two librarians of Manchester Metropolitan University who had expertise in translating research questions into search concepts. The strategy employed search terms relevant to the aims of the review (i.e., ‘Diagnostic’, ‘Disclosure’ and ‘Genetic Conditions’). A list of alternative search terms was also developed, which are summarised in Table 1.

Table 1. Alternative search terms.

Diagnostic	Disclosure	Genetic Conditions
illness status, diagnosis	discovery, sharing information/ information sharing, convey information, communicate information, communication	genetic disorder, genetic defect, genetic abnormality, genetic disease, genetic syndrome

Throughout the search process, we noticed that the most effective and appropriate search terms were “Diagnostic Disclosure” OR “Disclosure” AND “Genetic Conditions”. Therefore, not all the alternative search terms were used in the final search. Boolean operators/truncation (e.g., “disclos*) and advanced search strategies (i.e., “AND”) were also used.

Literature searching took place in July 2020 and June 2022 to ensure all relevant studies were included. The databases searched were Medline (EBSCOhost), PsychInfo, Scopus, Web of Science, Wiley Online Library, and Manchester Metropolitan University Library Online. Additional reference checking and hand searching of references took place to identify potential studies. Publication date limits were not used to narrow the search.

2.3 Screening process

After removing duplicates, relevance of records was assessed by screening the titles and abstracts of the studies resulting from database and manual searches. Once non-relevant studies were removed, full articles were read in detail to assess their relevance to the aim of the review. Studies not meeting the inclusion criteria were removed and a total of 12 studies were included in the review (see Figure 1 for a summary of the screening process).

2.4 Data extraction

The data extracted from the studies comprised the title and aim(s)/objective(s) of each study, as well as information about the study samples, methods and main findings. Data extraction and entry was completed by the principal investigator (PI). The extracted data are summarised in Table 2.

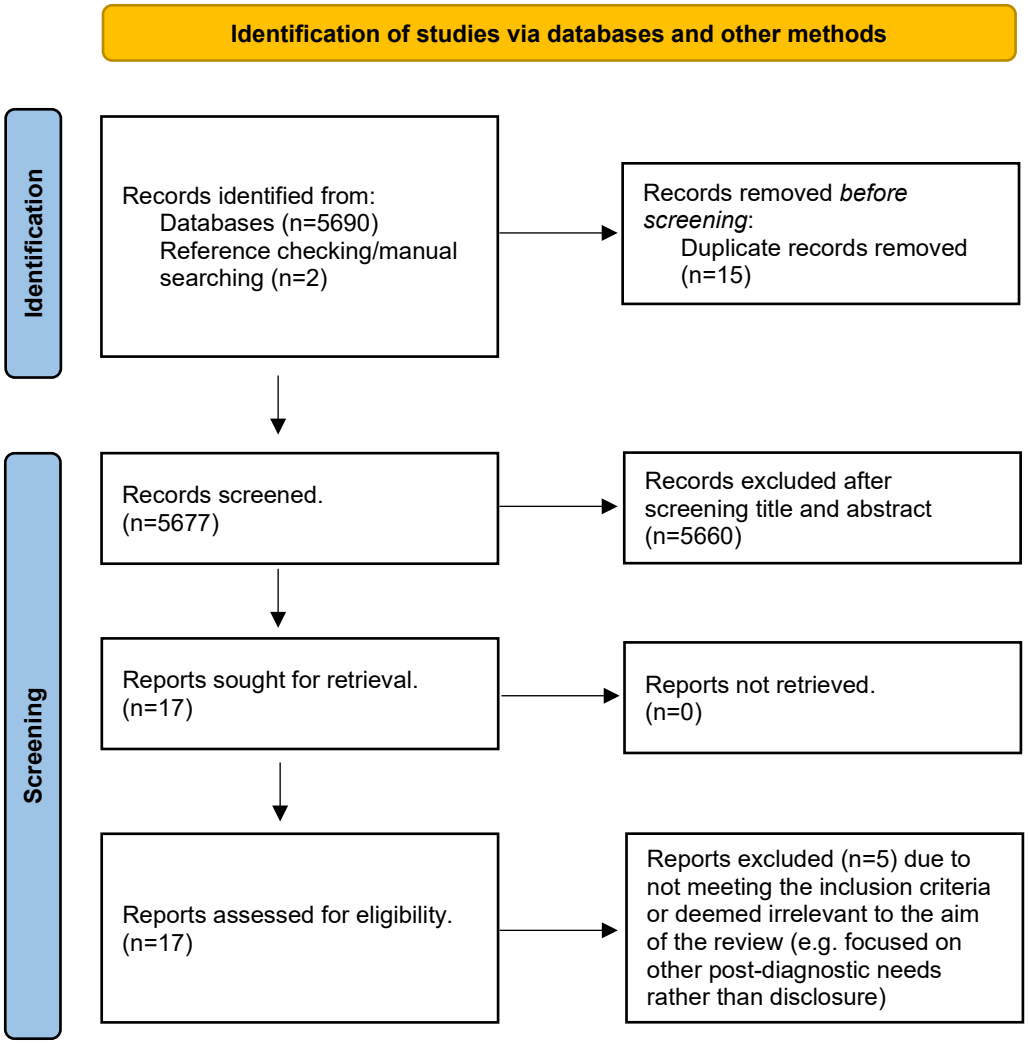
2.5 Quality appraisal

The quality appraisal of the included studies was conducted using the Quality Assessment Tool for Studies with Diverse Designs (QATSDD) (Harrison et al., 2021). The QATSDD appraisal tool has demonstrated good validity and reliability in the assessment of the quality of studies with different methodologies (Sirriyeh et al., 2012), including studies with a mixed-methods design. The QATSDD tool has the advantage of resulting in a quantitative quality end score, which allows making comparisons between studies and identifying potential biases. Each study was individually assessed using the QATSDD criteria which included: explicit theoretical framework, statement of aims or objectives in the main body of the report, a clear description of the research setting, evidence of sample size considered in terms of analysis, representative sample of target group of a reasonable size, description of procedure for data collection, rationale for choice of data collection tool(s), detailed recruitment strategy, statistical assessment of reliability and validity of measurement too(s) (Quantitative only), fit between stated research question and method of data collection (Quantitative), fit between stated research question and format and content of data collection tool such as interview schedule (Qualitative only), fit between research question and method of analysis, good justification for analytical method selected, assessment of reliability of analytical process, evidence of user involvement in design, and strengths and limitations critically discussed (Harrison et al., 2021). Each study was evaluated on a scale from 0 to 3 against each criterion, with 0 indicating that the studies did not meet the relevant criteria at all, and 3 indicating that the respective criteria were fully met in the studies. A total study quality score was then calculated, with a possible maximum score of 48 for mixed method studies and a maximum score of 42 for qualitative and quantitative studies.

3. Results

Database and manual searches resulted in 5,692 records. Once duplicates were removed, the titles and abstracts of 5,677 studies were screened for relevance. The full manuscripts of the remaining 17 studies were then read in detail and screened for relevance, with only 12 meeting the inclusion criteria and included in this review (see Figure 1 for a summary of the screening process). Nine of the studies included in the review employed a qualitative design, two studies used a quantitative design, and one study employed mixed methods. The quality appraisal of the reviewed studies resulted in scores ranging from 10 to 32.5 (23% to 76% of the Maximum Quality Score). Results of the quality appraisal process are presented in the last column of Table 2. We decided to include the studies with lower scores due to the scarcity of research in diagnostic disclosure of genetic conditions.

Figure 1. Flow chart of the screening process.



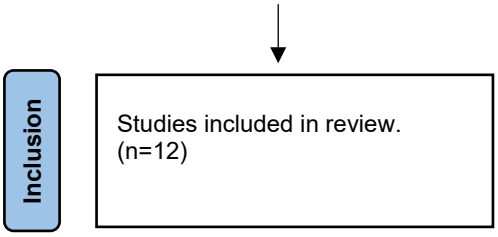

Inclusion
Studies included in review.
(n=12)

Table 2. Data extracted from the reviewed studies.

Study	Location	Aim(s)/Objective(s)	Study sample	Study design/methods	Main findings	Quality appraisal
Ablon (2000)	US	Not clearly stated	18 Families	Qualitative: Interviews (semi-structured)	• Parents recalled shock, fear and subsequent depression on learning of diagnosis. • Four mothers and fathers immediately went to a library to find further information. Reading materials often painted a negative picture. • Mothers typically the only parent present when given the diagnosis. • Families stated that they were given very little understandable information	10/42 (23% of the Maximum Quality Score)
Close et al. (2016)	US	To describe family management challenges for parents who have sons with Klinefelter Syndrome	40 parents (33 Female, 7 Male)	Mixed Methods Qualitative: Interpretative description-interviews Quantitative: Survey	• Parents described need for information, such as understanding features and symptoms of Klinefelter Syndrome (KS), support for disclosing the diagnosis and to create a logical plan for how to provide care for the individual. • Parents dissatisfied with Health Care provider knowledge of KS and sought information from the internet	32.5/48 (76% of the Maximum Quality Score)
Faux et al. (2012)	US	To gain insight into factors related to the decisions of caregivers in disclosing a diagnosis of 22q11 Deletion Syndrome	Primary Caregivers of 8 children aged between 10-17 years old	Qualitative: Interviews (semi-structured)	• Reasons for disclosure: the need to explain things to the child (e.g., why doctors' visits are necessary) • Concerns: didn't want the individual to focus on the diagnosis and use it as an excuse, did not want to scare the individual	17/42 (40% of the Maximum Quality Score)

					• Participants felt reasonably well-prepared for disclosure discussion but may need additional support/resources to increase confidence	
Gallo et al. (2005)	US	To examine parents' beliefs and strategies related to sharing information about a genetic condition with their affected and unaffected children	86 families, 139 parents (Phenylketonuria, Sickle Cell Disease, Cystic Fibrosis, and Marfan Syndrome).	Qualitative: Interviews (semi-structured)	• N=68 openly shared information about the condition and inheritance • N=57 selectively shared information • N=5 used conversations between parent and healthcare professional • Initial focus of discussion was on the management of the condition and maintaining a positive attitude. • Parents considered their child was ready for information when they demonstrated curiosity. • Concern about potential blame and future consequences influenced decision to disclose. • Healthcare professionals need to actively partner with parents to support and find strategies to integrate or share information	29/42 (69% of the Maximum Quality Score)
Goodwin et al. (2014)	Australia	To examine the relationship between the diagnosis experience and the disclosure experience for parents of children with developmental disorders of a known genetic aetiology	559 parents and caregivers • 22q11DS, N=193 • Down Syndrome, N= 122 • Fragile X Syndrome, N=34 • Williams Syndrome, N=48 • Tuberous Sclerosis, N=111 Prader-Willis Syndrome, N=51	Quantitative: Online survey	• Diagnosis experience was stressful, and parents felt they initially had a poor understanding of the syndrome. The amount /quality of the information received from healthcare professionals was unsatisfactory. • Caregiver respondents in the Down Syndrome group disclosed to their individual earlier and felt more prepared to do so than 22q11DS	31.5/42 (75% of the Maximum Quality Score)

Hallberg et al., (2010)	Sweden	To explore and describe parent's experiences of the diagnostic process and of being parents of a child with 22q11 deletion syndrome	12 parents (2 male), with children aged between 2-40 years	Qualitative: Interviews (semi-structured)	• The time before the diagnosis was full of worries regardless of the age of the child. • Parents of those diagnosed at an early age were often unable to process the information and did not want to know all the details of the condition immediately. • Parents of those diagnosed later found it was an affirmation of their suspicions. Repeated meetings with healthcare professionals recommended for information to be learnt gradually	16/42 (38% of the Maximum Quality Score)
Kunmar et al. (2018)	Singapore	• Describe the impact of thalassemia on the affected individual's family, social and professional lives. • Better understand disclosure patterns, what they tell and to whom	30 (16 individuals, 14 parents) 11 females, 14 males	Qualitative: Interviews	Major themes: • Medical management • Impact on social and professional life • Decision making, influential factors: Considered social, normative, and strategic practical factors. 1) Social: Nature and length of relationship 2) Normative: How other people managed decision making, e.g., friends with same condition and families 3) Practical: Impact condition would have on life-stigma • Concerns: physical limitations, demands of medical treatment • Suggestion of more involvement from healthcare professionals in communicating with parents and individuals	21/42 (50% of the Maximum Quality Score)

Middleton et al., (2018)	UK	To explore how parents communicate with children affected by Sickle Cell Disease	12 parents of children aged between 7-14 years old with a diagnosis of Sickle Cell Disease (8 female, 4 male)	Qualitative: Interviews (semi-structured)	• Timing and content of conversations typically started by children. Topics e.g.: genetic inheritance, medical appointments and procedures, normalising and offering hope. • Proactive Sickle Cell Communication - responding to questions as they arise with the aim of empowerment. • Challenges: avoidance due to guilt of passing on the condition, and appropriate communication for individual's developmental stage • Gradual disclosure and 'socialisation' to living with the condition. • Support from healthcare professionals to adapt communication for the child's needs or communicate on their behalf	19/42 (45% of the Maximum Quality Score)
Muggli et al., (2009)	Australia	To explore the experiences of families with a baby with Down Syndrome at the time of diagnosis, and their preferences for information and support in the early period after diagnosis	18 families (mothers, aged between 25-43 years)	Qualitative: Interviews	• Parental coping of unexpected diagnosis influenced by time interval between birth and disclosure, level of certainty of attending physician, and time interval between disclosure of clinical suspicion and confirmation. • Parental needs for support: normalising postnatal care, ensuring privacy, and providing early access to peer support and up-to-date information	15/42 (35% of the Maximum Quality Score)
Nisbet et al. (2022)	UK	• Explore the experiences of diagnostic disclosure and disclosure to others for adolescents with	5 girls with Turner Syndrome and one parent/guardian of each girl	Qualitative: Interviews (semi-structured)	• Three themes identified: Communication and support, Stigmatisation of TS, and Psychological Consequences • Disclosure as a gradual process, with the individual attaching more meaning to the condition over time.	21/42 (50% of the Maximum Quality Score)

		Turner Syndrome and their parents/guardians. • Examine the impact of disclosure on the lives of the individuals and families			• Primary concern of the individual was short stature, until adolescence when this changed to infertility. • Continuous process of acceptance as different challenges faced throughout lifetime. • Desire to conceal diagnosis from others-fear of stigma and judgement. • Anxiety and uncertainty around the future and prognosis of the condition	
Sutton et al. (2006)	Unknown	To ascertain the social, psychological, and medical concerns and challenges experienced by girls and women affected with Turner Syndrome.	97 girls and women living with Turner Syndrome, and 21 parents	Qualitative: Interviews (semi-structured)	• Secret-keeping within Turner Syndrome population is surprisingly common, which can lead to patient's depression, isolation, fear, and general mistrust. • Disclosing infertility is a particular challenge for parents, who perceive themselves to have a lack of knowledge, alongside factors such as stigma, the individual's desire to become a mother, and parental loss of a biological grandchild. • Disclosing diagnosis at a young age helps the individual to adjust and include Turner Syndrome within their self-identity and development. • Overall parental dissatisfaction of disclosure due to the complexity of the information and lack of empathy received from healthcare professionals. • Recommendation for healthcare professionals to discuss with the parents and individuals how much information they would like to receive	10/42 (23% of the Maximum Quality Score)
Waxler et al. (2013)	US	To learn about the experience of receiving a diagnosis from parents of	Parents recruited through the Williams Syndrome Association parents support group- 574 responses	Quantitative: Survey	• 17 families reported receiving out-of-date information. • Nearly 60% of parents stated they were told something inappropriate, unhelpful, or insensitive at the time. • Healthcare providers should:	22/42 (52% of the Maximum Quality Score).

		children with Williams Syndrome			○ Engage with the family in dialogue. ○ Deliver the message accurately and compassionately. ○ Have up-to-date information readily available. ○ Listen to the family and answer questions. ○ Stop and think before giving diagnosis over the phone, showing a picture, or bringing trainees into the room. ○ Involve other knowledgeable professionals. ○ Connect families to support groups and other parents affected by WS. ○ Assist with developing a game plan. ○ Remember that words can have a lasting impact	

The studies included in the review were conducted in a range of locations. The majority of studies were conducted in the United States (US) (Gallo et al., 2005; Close et al., 2016; Waxler et al., 2013; Faux et al., 2012; Ablon, 2000), with the rest conducted in Australia (Goodwin et al., 2014; Muggli et al., 2009), the United Kingdom (Middleton et al., 2018; Nisbet et al., 2022), Sweden (Hallberg et al., 2010), and Singapore (Kunmar et al., 2018). There were a range of genetic conditions explored within the studies included in the review. These were conditions such as Down Syndrome, Williams Syndrome, Klinefelter Syndrome, Sickle Cell Disease, and 22q11.2 Deletion Syndrome. Furthermore, the studies explored disclosure from the perspectives of families or primary caregivers and the individuals living with the diagnosis. Of interest, the majority of studies focused on the perspectives of the family members/caregivers of the individuals living with the condition, apart from three studies which included those living with a diagnosis (Kunmar, et al., 2018; Sutton et al., 2006; Nisbet et al., 2022).

3.1 Protecting the individual.

In three of the twelve studies (Middleton et al., 2018; Gallo et al., 2005; Faux et al., 2012) parents expressed their concerns about the importance of protecting the individual living with the condition. For example, parents wanted to avoid causing unnecessary or preventable panic or worry to the individual living with the condition (Gallo et al., 2005). This was also linked to another aspect frequently reported by parents which was related with disclosure as a gradual process of information sharing (Middleton et al., 2018). The gradual aspect of disclosure was highlighted as a protective factor against overwhelming information and disclosure experiences that would be difficult to process for those living with the condition. Additional emphasis was placed on normalising the condition for the child and empowering them through their active involvement in the management of their condition (Middleton et al., 2018). Parents frequently reported the importance and benefits of open communication, including discussions about the possible limitations resulting from diagnosis and how these could be addressed, to ensure trusting relationships within the family and protect the individual from traumatic accidental discovery of their condition (Middleton et al., 2018; Nisbet et al., 2022; Sutton et al., 2006). This appeared to present a conflict as

on one hand the importance of open communication was emphasised, yet on the other hand parents did not want to unnecessarily burden the child and wished to protect them from knowing about their diagnosis.

3.2 Developmental stage

In two of the twelve studies (Gallo et al., 2005; Faux et al., 2012), a major concern of parents was associated with the developmental stage of the individual and their preparedness to learn about their diagnosis. Typically, parents seemed to assess the child's readiness to receive information by observing the curiosity they displayed about the condition or any questions they asked in relation to emerging symptoms (Gallo et al., 2005). The main aim of the parents behind the needs-appropriate controlled flow of sensitive information towards the individual appeared to be related with their attempts to normalise the diagnosis. Parents initially focused on the practical management and treatment of the condition and then gradually shared information about more complex aspects of the diagnosis, according to their child's needs and preparedness to process the relevant information (Gallo et al., 2005). The latter included topics related with childbirth and disclosure of diagnosis within a relationship (Gallo et al., 2005).

3.3 Adapting to diagnosis and uncertainty around living with the condition.

In four of the twelve studies (Middleton et al., 2018; Close et al., 2016; Hallberg et al., 2010; Sutton et al., 2006), parents discussed how they themselves had to adapt and process the diagnosis, while considering how to disclose it to the individual (Close et al., 2016). Common barriers to disclosure and concerns of family members appeared to be related with their own feelings of guilt that their child had to live with a heritable condition which was passed on by them (Middleton et al, 2018). Furthermore, parents reported a sense of uncertainty in six of the twelve studies. This uncertainty included concerns about how to respond to potential emotional distress experienced by their child at the point of or after disclosure (Middleton et al., 2018) and the prognosis and future progression of the condition (Faux et al., 2012; Close et al., 2016;

Ablon , 2000; Gallo et al., 2005; Middleton et al., 2018; Nisbet et al., 2022). Uncertainty around these aspects acted as potential barriers to disclosure and required personal adjustments and support to promote accepting the diagnosis, acquiring sufficient knowledge about the condition and coping strategies, and alleviating confusion resulting from diagnosis.

3.4 Fear of stigma

An additional aspect influencing disclosure was related with fear of stigma. This seemed to be a concern for family members and individuals living with a genetic condition alike and appeared to constitute a significant barrier to diagnostic disclosure to others (Sutton et al., 2006; Close et al., 2016; Kunmar et al., 2018; Ablon, 2000; Middleton et al., 2018; Nisbet et al, 2022). Another significant barrier to disclosure by parents was related with specific symptoms of the genetic condition, such as infertility resulting from Turner Syndrome, which could elicit stigma and discrimination by peers (Sutton et al., 2006; Nisbet et al, 2022). The fear of stigma and judgement extended to parents who felt guilty for passing on a genetic condition and experienced a sense of loss of much anticipated biological grandchildren. Such barriers to disclosure were primarily the result of family member's lack of knowledge around the condition and how to cope with symptoms and prevented conversations with their children about the diagnosis (Sutton et al., 2006).

3.5 Emotional impact of disclosure

In five of the twelve studies, parents commonly reported a negative diagnostic experience (Goodwin et al., 2014; Close et al., 2016; Waxler et al., 2013, Ablon, 2000; Sutton et al., 2006). They were often left to conduct their own research about the condition (Close et al., 2016) or were provided with out-of-date information (Waxler et al., 2013). Parents also highlighted the need for healthcare professionals to become aware of the impact of disclosure and the way this is conveyed on families affected by genetic conditions (Waxler et al., 2013), as many reported experiences of a blunt disclosure (Ablon, 2000), receiving insensitive, inappropriate or insufficient information

(Sutton et al., 2006; Waxler et al., 2013), and a lack of emotional support at the point of diagnosis (Sutton et al., 2006). This appears to be in contrast with Nisbet et al., (2022), who reported that most participants had a positive experience of diagnostic disclosure by healthcare professionals and highlighted openness and honesty as key factors of a positive disclosure (Nisbet et al., 2022).

Regarding diagnostic disclosure from the parent/family member to the individual living with the condition, Gallo et al. (2005) identified an additional barrier related with fear. Some parents were afraid that their child would respond in an angry manner when learning about the diagnosis, due to the genetic aetiology and heritability of the condition. This consequently influenced parents' decision to disclose the diagnosis, leading to delays in disclosure and sustained emotional distress in parents.

3.6 Support from healthcare professionals

In seven of the twelve studies, the findings indicate the need for additional support from healthcare professionals at the point of diagnosis, to facilitate a positive experience of disclosure, understanding, accepting and coping with the condition, and acquiring access to needs-appropriate post-diagnostic support (Middleton et al., 2018; Gallo et al., 2005; Hallberg et al., 2010, Waxler et al., 2013; Kunmar et al., 2018; Muggli et al., 2009; Sutton et al., 2006). Although parents prefer to lead and be responsible for disclosing the diagnosis to their child in the first instance (Gallo et al., 2005; Hallberg et al., 2010), healthcare professionals may need to be more involved in the process of disclosure and partner with family members in a meaningful manner (Gallo et al., 2005). Middleton et al. (2018) support this finding and argue that healthcare professionals may need to speak with the individuals living with the condition alongside their parents or even on their behalf if needed.

Parents and family members also emphasised the need for a family-oriented approach to address the needs of the whole family at the point of diagnosis (Waxler et al., 2013). For instance, professionals should ask about or assess how much information would be beneficial at the initial stages of diagnostic disclosure (Hallberg et al., 2010), provide sufficient time to family members and individuals living with the genetic condition to ask additional questions and clarifications (Sutton et al., 2006), and arrange essential follow up appointments, as appropriate, for those affected by

the diagnosis (Hallberg et al., 2010). Although some service recipients may feel reasonably prepared for the disclosure process (Faux et al., 2012), there appears to be a general consensus that increased family-focused support from healthcare professionals could facilitate a positive experience of disclosure and building a sense of confidence around living with the condition.

4. Discussion

To the best of our knowledge, this is the first review of evidence on diagnostic disclosure of genetic conditions. We are confident that it may offer an important first step towards a better understanding of diagnostic disclosure of genetic conditions and provide the basis for future research. Although most of the studies were conducted with family members of people living with genetic conditions, our findings reflect important aspects of the disclosure experience. Parents tend to delay disclosure or favour the gradual release of information to the individual, with the primary aim to protect them from unnecessary concerns and social stigma. Our findings also highlight the psycho-emotional impact of diagnostic disclosure, particularly for parents, and the requirement for needs-appropriate family-focused formal support during the diagnostic period, to facilitate positive experiences of diagnostic disclosure and enable families affected by genetic conditions to adjust to living with the diagnosis.

Most studies focused on the perspective of parents and family members. These individuals play an integral role in the process of diagnostic disclosure, as they are often the first ones to find out about the diagnosis and have to make decisions about if, when, and how to share information with the individual living with the condition and others (Middleton et al., 2018). Previous reports have underlined the importance of including individuals living with a disability in important decision-making processes around their condition (WHO, 2022). However, parents withhold diagnostic information from their children to protect them from social stigma and emotional distress, which may eventually jeopardise family relationships and promote feelings of isolation to the individual living with the diagnosis (Sutton et al., 2006, Nisbet et al., 2022). Of interest, we found only three studies that included the perspectives of those living with the condition (Kunmar, et al., 2018; Sutton et al., 2006; Nisbet et al., 2022), which makes it difficult to infer their views on this aspect of disclosure. The findings indicate that

secret keeping may be quite common within families affected by genetic conditions, particularly when there are life-changing implications resulting from symptoms such as infertility (Sutton et al., 2006). Nevertheless, individuals living with the diagnosis appear to benefit from honest communication and disclosure at a young age, which help them to adjust to living with the condition and reduce the impact of diagnosis on self-identity (Nisbet et al., 2022).

The point of disclosure by healthcare professionals seems to place particular challenges and pressure on parents. The lack of needs-appropriate information, advice and support can leave parents in a blindfold as to how they can make sense of and cope with the diagnosis, their sense of guilt for passing on a genetic condition, and making a decision about whether or how to disclose information about the diagnosis to their child (Close et al., 2016; Hallberg et al., 2010; Sutton et al., 2006). These pressures can significantly delay disclosure and may be preventable through condition-specific information and support during medical appointments. In eight of the twelve reviewed studies, participants emphasised the importance of effective informational resources that provide clear information about the condition and how to cope with symptoms, signposting to peer support groups with other families affected by the same condition, and follow-up appointments for further clarifications around diagnosis and support with emerging symptoms and needs. Although these findings are consistent with current guidance on positive support for people living with a disability (WHO, 2022), very few studies provided evidence of relevant good practice, potentially due to limited resources within healthcare systems or limited awareness among healthcare professionals regarding the impact of diagnostic disclosure of a genetic condition.

Of interest, our findings also highlighted fear of stigma as a barrier to disclosure. This fear and associated sense of insecurity may result from the aforementioned lack of awareness of genetic conditions and their impact. Farmer and Macleod (2011) argued that inflexible policies, undermining access to relevant information, and discriminatory behaviours constitute significant barriers to social inclusion of individuals living with a disability. Clark (2015) reported that biases and stereotypical beliefs against individuals living with a disability are often associated with irrational fears, misleading information and lack of exposure. Similar issues were highlighted by the World Health Organisation who underlined the need for effective public educational campaigns and professional training for healthcare practitioners, to promote

awareness of the rights and needs of people living with disabilities and prevent social exclusion resulting from diagnosis (WHO, 2022). It may be reasonable to assume that if put in practice, these measures could alleviate the fear of stigma for parents and individuals living with a genetic condition and facilitate early disclosure of diagnosis within the family and to others.

4.1. Limitations and implications for future research

The quality appraisal of the reviewed studies resulted in scores ranging from 10 to 32.5 (23% to 76% of the Maximum Quality Score). It was decided to include the studies with lower scores due to the scarcity of research on diagnostic disclosure of genetic conditions. The inclusion of the studies even with lower quality scores, meant we could demonstrate the research available within the topic area, and highlight the weaknesses and gaps of knowledge. This could then result in recommendations for real life future research. Yet we recognize the possibility of bias resulting from this decision and the potential reduction in the credibility of the findings. We also observed Patient and Public Involvement in only three of the reviewed studies (e.g., members' feedback on plausibility of study findings) which may influence the credibility (Kylen et al., 2022) of the overall findings of our review.

The studies included in this review were largely focused on the perspectives of parents, primarily those of mothers of children living with genetic conditions. It is not entirely clear if this was related with mothers' stronger sense of responsibility, their higher degree of preparedness to come forward and share their experiences, or unilateral recruitment strategies employed within individual studies. Nevertheless, these studies provide useful insights but carry a risk of bias since mothers' perspectives may not represent or be consistent with fathers' viewpoints and experiences, and more importantly with those of the individuals living with the diagnosis. Future research needs to consider the perspectives of both parents, individuals living with the condition and their siblings, to gain a holistic in-depth insight into individual and family needs and inform family-focused approaches for diagnostic disclosure and family-centred interventions for support during the diagnostic period. Equally important insights may result from studies focusing on the experiences of

healthcare professionals, to identify individual, structural and organisational barriers and facilitators to diagnostic disclosure of genetic conditions.

The studies included within this review were conducted in western countries, with most studies carried out in the United States. It is thus possible that the findings of our review may be ethnocentric or country specific. Furthermore, there were limited descriptions of study samples regarding the ethnicity of participants (and in some cases gender). Studies which provided relevant information indicate a limited representation of non-western cultures, Black and Asian Minority Ethnic groups, and individuals with lower socio-economic status. It is therefore difficult to determine the transferability of the findings in families with different ethnic, cultural or socio-economic backgrounds, whose experiences with services and cultural norms around illness may significantly differ (Furnham and Swami, 2018). For instance, Rosenberg et al., (2017) reported cultural factors influencing power positions around diagnostic disclosure; whereas in western societies the individual living with the condition is usually (though not always) expected to make decisions about disclosure of the diagnosis, community-based cultures seem to favour collusion and place the decision-making process in the hands of the whole family. Such norms could significantly influence disclosure experiences and require further investigation. Future research needs to focus on the experiences of families with different cultural and socio-economic backgrounds, to identify culture-specific needs around diagnostic disclosure and contextual factors influencing the disclosure process.

A final limitation is related with the academic qualification, in the context of which the review took place. As the review was part of a professional doctorate thesis, literature searches, record screening, data extraction, study quality appraisal, and thematic clustering of findings, were carried out by the PI with the guidance of the research team. Although the search strategy was developed with and approved by two librarian experts at Manchester Metropolitan University, the remaining aspects of the review may carry a risk of bias due to potential subjective interpretations by the PI.

5. Conclusion

This review mapped current evidence on experiences of diagnostic disclosure of genetic conditions, from the perspectives of family members and individuals living with the condition. Overall, the findings indicate that openness about the condition and gradual information sharing from an early age facilitate disclosure and adapting to living with the condition. In contrast, fear of stigma and the potential psychosocial impact of diagnosis act as significant barriers that lead to delays and may place family relationships at risk. Sensitive healthcare professionals need to collaborate with families affected by genetic conditions and provide needs-appropriate family-focused information and support, to facilitate a positive experience of disclosure and prevent confusion. Future research needs to examine more closely the disclosure experiences of individuals diagnosed with genetic conditions and families from minority ethnic groups, to enable the development of inclusive condition-specific guidance on disclosure and interventions appropriate to the needs of those affected by diagnosis.

Introduction to Thesis Chapter 2

To follow on from the systematic mapping review conducted, and the findings presented in the previous first chapter, an empirical research project was designed. This empirical thesis project is detailed within chapter 2.

The mapping review focuses on the experience of diagnostic disclosure within genetic conditions in general, yet the findings from this were used to compare the experience found specifically in the genetic condition of Turner Syndrome. As a result of the findings from the review, the current project explored the perspective of individuals living with Turner Syndrome, as well as family members. This is because the perspective of the individual is something that was limited within the review research. It was hoped that the participants who were family members, may have been a more varied display of those that can be involved in the experience of disclosure, for example, fathers, siblings, grandparents. However, as in previous studies, the family member participants were mothers. It was further hoped the participants may have demonstrated the experience of beliefs around disclosure from a variety of cultures and countries, however, this information was not gathered in this study and the participants had to be living in the United Kingdom to take part in the research. Therefore, these are factors which should be considered in future research.

Chapter 2. Diagnostic disclosure in Turner Syndrome: A qualitative exploration of the experiences and needs of individuals living with Turner Syndrome and family members.

1. Introduction

Turner Syndrome (TS) is a genetic condition which affects approximately 1 in every 2,500 live female births (Hutaff-Lee et al., 2019). The condition results from a genetic abnormality characterized by the complete (classic TS) or partial (mosaic TS) absence of an X chromosome (Ackermann and Bamba, 2014). Although individuals living with TS may present with different symptoms, the most common manifestation of the condition includes short stature, a webbed neck and underdeveloped ovaries resulting in infertility (Gravholt et al., 2017). The age of diagnosis varies from the prenatal period to 18 years of age, with significant delays observed when TS is not diagnosed before or up to one year after birth (Swauger et al., 2021). Early diagnosis usually derives from prenatal testing and age-appropriate screening, whereas diagnosis during adolescence is triggered by short stature and pubertal delay. A delayed diagnosis may limit access to early interventions, such as growth hormone, prevent improved adult height and growth response (Baxter et al., 2007), and lead to lower quality of life for those living with more severe phenotypes (Krantz et al., 2019; Swauger et al., 2021).

Most of the research on TS is focused on biological and genetic causes. Despite the importance of such insights, scholars have highlighted the need for a more in-depth understanding of the psychosocial impact of TS (Sandberg et al., 2019), particularly diagnostic disclosure (Sutton et al., 2006). Diagnostic disclosure is broadly defined as the extent to which individuals openly discuss a diagnosis and the associated thoughts and feelings with a range of other people (Munro et al., 2015). Previous studies have emphasized the need to avoid focusing on TS symptoms and painting a pessimistic picture, as conveying disclosure without consideration of the person's needs and sensitivities can negatively impact the self-esteem and self-concept of individuals living with TS (Kagan-Krieger, 2001). Irrespective of age of diagnosis, parents are most commonly involved in diagnostic disclosure (Swauger et al., 2021) and often withhold information about important aspects of the condition, such as infertility (Sutton et al., 2006), due to lack of knowledge and fear of stigma. This can lead to feelings of mistrust and isolation in individuals living with TS (Sutton et al., 2006, Nisbet et al., 2022) and a tendency to conceal the diagnosis until the condition and its lifelong symptoms can be processed, understood and accepted (Nisbet et al., 2022).

Currently, there are evidence-informed guidelines on diagnostic disclosure for different conditions, such as HIV (WHO, 2011), yet none for TS. This is mainly due to the paucity of research exploring diagnostic disclosure of TS and its impact on those affected by the condition. Recent research findings suggest that healthcare professionals should be explicit about the challenging symptoms of TS, discuss with family members the responsibility and process of disclosing sensitive information to the person with TS, and provide continuous advice and support according to the family's needs (Nisbet et al., 2022). As in the case of other lifelong conditions, a more thorough and in-depth understanding of diagnostic disclosure of TS and its impact could enable optimizing the disclosure process and facilitate effective coping and adapting to living with the condition (Gallo et al., 2005; Munro et al., 2015).

Through this UK-based study, we aimed to add to the evidence base and provide TS-specific recommendations on diagnostic disclosure by: (a) exploring the experiences and needs of individuals living with TS and family members around diagnostic disclosure, and (b) identifying the barriers and facilitators involved in the process. (Individual(s) living with TS will be referred to as ILWTS in the rest of the manuscript).

2. Materials and methods

The study received ethics approval by the Ethics Committee of Manchester Metropolitan University, Department of Psychology (Ref. No: 25038).

2.1 Study design

We employed a qualitative design using in-depth semi-structured interviews with ILWTS and family members to gather rich detailed data. Interview data were analysed using inductive thematic analysis (Braun and Clarke, 2022). This flexible data-driven analytical approach was chosen due to its focus on rich meanings and patterns across the dataset, grounded on participants' experiences and perspectives (Braun and Clarke, 2013). We employed a critical realist epistemological position (Willig and Rogers, 2017), which stands between constructivism and positivism. It posits that reality can be influenced by context and research enables the interpretation

of participants' version of reality (Willig and Rogers, 2017) by examining their perceptions, reflections and linguistic expression around the phenomenon of interest (Willig, 2008). In that sense, participants actively contributed to data generation with the meanings they attached to their accounts and experiences. This approach was deemed suitable as diagnostic disclosure of TS is an experience that can take place in different contexts and have different meanings for different individuals.

2.2 Patient and Public Involvement

A patient and public involvement group (PPI) was established prior to the study, comprising an ILWTS and a family member. Both members of the PPI group were identified through the charity Turner Syndrome Support Society UK (TSSSUK), which supports those affected by TS and acted as the study gatekeeper. The group was consulted in regular intervals before, during and after the study to ensure the relevance and appropriateness of the study design, aims and objectives, recruitment methods and materials, data collection instruments, and final study findings. PPI meetings took place before important milestones, such the study design and write up stages. Feedback from PPI members was provided via email.

2.3 Recruitment

Participants were recruited using convenience and snowball sampling. Recruitment took place through the study gatekeeper (TSSSUK) and members of the study Patient and Public Involvement (PPI) group who advertised the study digital poster through their social media, networks, and websites. Prospective participants contacted the PI via email, who then provided the study participant information sheet and consent form and offered to answer any questions participants may have. All participants had at least one week to decide whether they would like to take part. Once participants confirmed their interest and had the opportunity to ask questions about the study, a suitable date and time for a Skype or telephone interview was arranged, according to participants' preferences.

2.4 Participants

Participants were females with a confirmed diagnosis of TS or family members who cared for a person living with TS. All participants had to be aged 16 or above, living in the UK and able to communicate in English. Caregivers who took part also had to have experience of disclosing the diagnosis to ILWTS (or be preparing to disclose the diagnosis to ILWTS) and/or receiving the diagnosis from a healthcare professional. Family members and ILWTS did not have to be from the same families. Only one family member and one ILWTS from the same family took part in the study.

Consistent with Braun and Clarke's views (2016, 2019), there was no predetermined sample size, as sample size calculations and data saturation are not valid concepts/processes in studies employing reflexive thematic analysis. Instead, the research team regularly monitored the quality and richness of data during data collection and analysis which were concurrent. In line with Braun and Clarke's guidelines, data collection only ceased once it was jointly decided that rich data telling an insightful and coherent story had been gathered. At the point when this decision was made, 16 ILWTS and 8 family members had been recruited to the study.

2.5 Data collection

Due to the coronavirus pandemic, all interviews were conducted over the telephone or Skype from November 2020 to February 2021. Each interview lasted between 40 and 60 minutes. All participants provided informed consent at the beginning of each interview, which was audio-recorded, and were offered a list of relevant sources of support. All interviews were carried out by the principal investigator (PI) using a semi-structured interview schedule which was developed by the research team. Due to the sensitive nature of the topic, we decided to interview ILWTS and family members separately (see appendices A and B for the respective interview schedules) to facilitate open honest discussions and prevent potential concerns about the impact of disclosing criticisms around diagnostic disclosure on family relationships (Voltelen et al., 2018). Additional information was gathered on participants' socio-demographic background (age, education level, time since diagnosis, if disclosure to ILWTS had taken place, age of ILWTS at disclosure of diagnosis, and relationship of

family member with ILWTS) at the start of the interview. At the end of each interview, participants selected a pseudonym of their choice which the PI used to pseudonymize the interview transcripts.

Participants' verbal consent, sociodemographic information and interview data were recorded in separate audio-files and stored in separate password-protected folders in the secure servers of Manchester Metropolitan University, to protect participants' anonymity and confidentiality. After each interview, all data was transferred to the aforementioned servers and were erased from the recording device.

2.6 Analysis

Audio-recorded interviews were transcribed verbatim by the PI (EC). During transcription, all transcripts were pseudonymized by the PI and any other data that could render participants identifiable (e.g., names of individuals, names of services, places) were anonymized by removing the relevant details. Qualitative interviews were analysed by the PI using manual coding and following the six steps of thematic analysis identified by Braun and Clark (2006; 2013). All members of the research team (EC, VS, MT, VL) read repeatedly three interview transcripts and engaged in independent coding. Individual codes and notes were then discussed and compared during team meetings to ensure consistency and a detailed approach which encapsulates essential aspects of participants' accounts. The PI then analysed the remaining transcripts and discussed sets of these with the supervisory team during fortnightly team meetings. By consensus, the codes were clustered into subthemes and major themes based on conceptual similarity. The PI rechecked the full dataset repeatedly to ensure that the results of analysis and thematic map accurately depict participants' voices and accounts. After numerous iterations and discussions within the team, the subthemes were clustered into 3 major themes and the thematic map was finalised.

2.7 Quality assurance

Specific quality criteria are recommended for trustworthiness in qualitative research (Shenton, 2004). To increase credibility, fortnightly research team meetings took place, and the emerging final themes and thematic map were presented to the study PPI group to receive feedback on the relevance, coherence and plausibility of the findings. All interviews were carried out by the principal investigator who also lives with TS. We believe that this 'insider' position facilitated building trust and rapport with participants and provided a safe space for participants to report and reflect on their experiences (Ross, 2017). However, to check for potential bias resulting from this position and enhance confirmability, the PI used a reflexive journal throughout the study (Robson, 2011) and engaged in regular discussions with the supervisory team during data collection and analysis to discuss tentative results and consider different interpretations. All decisions were made by consensus among the members of the research team, all of whom have expertise in qualitative research and analysis.

3. Results

Overall, 16 ILWTS, and 8 family members took part in the study. Tables 1 and 2 present the socio-demographic data of participants.

Table 1. ILWTS socio-demographic data (age of diagnosis is the same as the age of disclosure unless stated otherwise).

Participant identifier	Age range	Level of education	Age at diagnosis
1	30-40	University Degree	Diagnosed 3 days old, 4 years old at beginning of disclosure
2	40-50	University Degree	18 Years old
3	40-50	University Degree	Diagnosed at 4 years old, Disclosure at 11 years old
4	20-30	University Degree	13 Years old
5	20-30	University Degree	12/13 Years old
6	16-20	Age 18	9 Years old

7	40-50	University Degree	16 Years old
8	30-40	Vocational Qualification	14 Years old
9	20-30	University Degree	6/7 Years old
10	30-40	University Degree	15 Years old
11	40-50	University Degree	11 Years old
12	16-20	Vocational Qualification	12 Years old
13	50-60	Age 18	12 Years old
14	50-60	University Degree	Diagnosed at birth, Disclosure at 6 years old
15	30-40	University Degree	25 Years old
16	20-30	Other	14 Years old

Table 2. Family member participants' socio-demographic data.

Participant identifier	Age range	Level of education	Relationship with ILWTS	Disclosed diagnosis	Time since disclosure
1	30-40	University Degree	Mother	Yes	7 years
2	50-60	University Degree	Mother	Yes	10 months
3	40-50	University Degree	Mother	Yes	2 years
4	40-50	University Degree	Mother	Yes	5 years
5	50-60	Age 18	Mother	Yes	10 years
6	30-40	University Degree	Mother	No	n/a
7	60-70	Vocational Qualification	Mother	Yes	14 years
8	50-60	University Degree	Mother	Yes	16 years

3.1 Qualitative findings

Thematic analysis resulted in three major themes: ‘Guardianship of disclosure’, ‘Coping with infertility’ and ‘Awareness of Turner Syndrome and its impact’ (see Figure 1 and table 3).

Figure 1. Presentation of thematic map resulting from analysis.

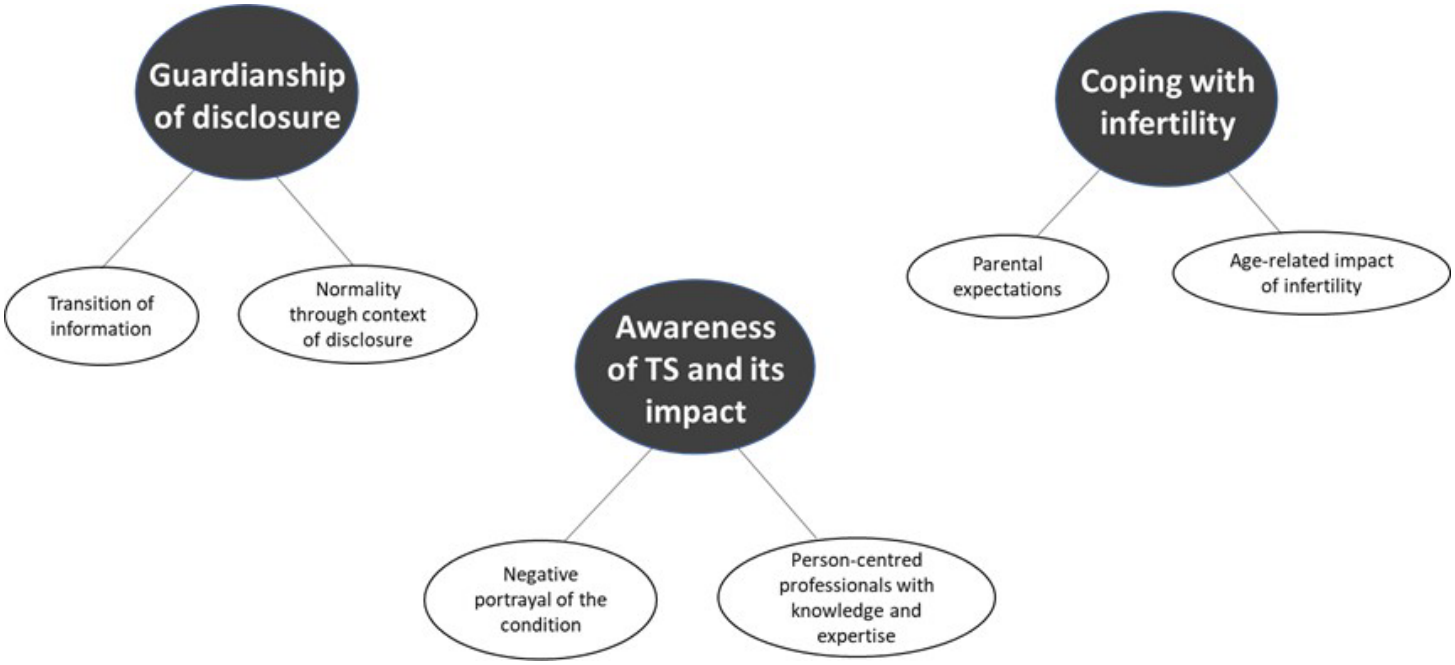


Table 3. Presentation of major themes, descriptors and sub-themes resulting from analysis.

Major Theme	Theme Descriptor	Subthemes
Guardianship of disclosure	This theme illustrates the processes involved in the disclosure of information about the condition from family members to ILWTS. The theme focuses on the careful management of information flow by the family member and the gradual transition of management to the individual, to enable ILWTS to maintain a sense of normality.	• Transition of information • Normality within the context of disclosure
Coping with infertility	This theme describes the influence of infertility in the disclosure process and the different impact it may have on ILWTS and family members, based on evolving expectations about the ILWTS forming a family.	• Parental expectations • Age-related impact of infertility
Awareness of Turner Syndrome and its impact	This theme encompasses the need to raise awareness in the public and promote knowledge and understanding of the condition and the impact of disclosure among healthcare professionals, to prevent stigma resulting from disclosure and enable ILWTS and family members to receive strengths-based advice and support.	• Negative portrayal of the condition • Person-centered professionals with knowledge and expertise

3.1.1 Guardianship of disclosure

This theme encapsulates two subthemes related with controlling the flow and disclosure of information about the condition, particularly management of information and the timing of transition of management from the family member to the individual, to minimize the impact of disclosure and promote a sense of normality in ILWTS.

Transition of information

This subtheme focuses on family members typically controlling and managing the disclosure of information about the condition. Some family members stressed the need to protect ILWTS from any worry or burden around living with TS and control or manage who has access to information about the condition: *"I've always said, if she gets to eighteen and she wants to stand on the rooftops and shout it, that's fine by me ... but until that point it is, you know I'm the guard, I'm like the guardian of that information ... she shouldn't be growing up with that label."* (Mother, Participant 24). This quote indicates the parent's perceived responsibility to carry the burden of the diagnosis and protect ILWTS from potential stigma until they reach adulthood, so that they can live a normal childhood and make their own informed decisions around disclosure at an age of controlled vulnerability. However, this view on responsibility for the management of information about the condition was challenged by ILWTS who expressed a desire to be involved in the disclosure process earlier on: *"If it's you know an older girl who's been diagnosed sort of maybe 12, 13 something like that ... ask them, involve them in how, in the decision on how to tell people and whether they want to tell people or want people to know, because ultimately it's their, it's their lives and their condition."* (ILWTS, Participant 1). In this extract, the ILWTS elaborates on their need to develop a sense of agency over the condition and its disclosure from early adolescence; it appears that such an approach could contribute towards the person's sense of ongoing growth and an empowering sense of independence. Whereas the parent reported a perceived need for continued protection of ILWTS, the person living with the condition highlighted their desire to be involved in major decision-making processes as part of their socioemotional development. However, views around the right age for this process to take place varied across participants, depending on individual experiences of disclosure. For example, another ILWTS who experienced disclosure at the age of 13 felt that they were overwhelmed: "*I didn't really understand a lot of what was being said… a lot of stuff was just like medical jargon to me…*" (ILWTS, Participant 8). This quote illustrates the need to consider individual age-related needs and the way the relevant information should be conveyed when making a decision to disclose information about the condition to ILWTS. Heavy jargon and an excessive amount of information could undermine the disclosure process and elicit confusion in the individual who may not be able to process and accept the diagnosis.

Participants reported the usefulness of individual cues from ILWTS indicating the ‘right time’ to engage in initial discussions about the condition. Questions during early adolescence around sex education and slower development appeared to provide useful triggers. An ILWTS reported: *“I wasn’t growing, and I was always at a different level than the other people in my class I think I always figured out that.. there was something different.”* (ILWTS, Participant 11). Another ILWTS said: *“I learnt more about it when I was probably about nine or ten, when they started doing sort of sex education and things like that and puberty, teaching us all about that at school … And then my parents explained to me that I was slightly different.”* (ILWTS, Participant 1). These responses showcase how ILWTS noticing differences in development and learning progress often provide cues for parents to start the gradual disclosure of information around the condition, according to the needs of ILWTS and their preparedness and ability to process the relevant information. The element of gradual exposure to information about the diagnosis appeared to be pivotal in the process. An ILWTS reported:

> *If I was to speak to them [family members], I’d say get it straight in your head first, so, do all your research, speak to the medical professionals, speak to [Charity], so that you understand how the condition is going to affect your child, kind of in the future. So that you can then gradually have a conversation with them and just discuss it bit by bit, you know, don’t try and tackle all aspects of the condition in one conversation because it’ll just be overwhelming. My main piece of advice is adapt to how the person reacts.* (ILWTS, Participant 14).

This quote indicates the significance of avoiding an overwhelming experience of disclosure and adjusting to the needs of ILWTS and their capacity to assimilate and respond positively to information about the condition. Careful observation of the person’s reactions during the gradual disclosure of information can inform decisions about the right amount to be disclosed at each stage.

Normality within the context of disclosure

This subtheme is focused on the efforts and choices made by family members to control the context of disclosure, in order to normalize the process for ILWTS and help them to assimilate information that could be perceived as distressing or even devastating. Typically, parents experienced significant pressure and fear regarding the

reaction of ILWTS when learning about their diagnosis. This made some parents try to prepare as much as they could to control the disclosure process. A family member reported:

> *It would be a second consultation in June and I knew I that it was gonna be discussed there, so I knew I had to, that was my deadline, and I knew I had to find a way to talk about it … I kind of scripted it, scripted it to myself and I tried to think of what are the key things that need to be disclosed at this point.* (Mother, Participant 4)

This quote indicates some parents' perceived need to arrange a formal context for disclosure in the form of a 'big sit down' event, for which they had to prepare all details in advance. The process appeared to be very distressing due to a lock of control over the outcome. In contrast, other parents described the effectiveness of a completely different approach which enabled them to disclose the diagnosis during a usual daily activity and promote a 'normal' experience:

> *We were sitting at the table to have something to eat ... I think I must have taken a deep breath before saying it but then I'd said it, it was out and actually, you know it was, it was a relief ... I'd not had to do some big sit down and make some big thing about it cos she asked the question at the right time.* (Mother, Participant 24).

In this extract, the family member was also experiencing significant pressure in anticipation of a frightful impact of disclosure. Instead of a 'big sit down' event organised by the parent at the 'right time', the family member 'followed the lead' of the ILWTS during an ordinary discussion over dinner which facilitated the process and minimised the impact of disclosure on both the parent and the ILWTS.

In cases where family members chose to disclose important information about the condition at an earlier age, the need to normalize the disclosure process was met through creative age-appropriate strategies. A family member described their experience:

> *I also found it a lot easier talking about some of the stuff cos my kids at the time liked watching this thing called Fuller House on TV and one of the characters, she couldn't have babies so I kept saying you know mummy's told you before that you know you can't have children when you grow up, or you won't be able*

to have them naturally or maybe you can adopt them, there's nothing wrong with adopting." (Mother, Participant 5).

And another family member reported:

> *There were games and things like that and then you could plot your growth on this chart… there was also a couple of little files about why people need to take growth hormone and it talked about Turner Syndrome and she said to me: do I have Turner Syndrome? And I just said yeah because I wasn't gonna lie and that was when she was four.* (Mother, Participant 23).

These responses illustrate how parents normalized the disclosure process by relating aspects of TS to characters of a TV program and embedding the disclosure process in playful activities. Through such creative approaches, the parents placed the condition within a familiar non-threatening context, so that children can process the relevant information and relate to the condition with minimum impact.

Of interest, family members were not the only ones who at times felt the need to prepare for the disclosure process. ILWTS also provided evidence of a perceived need to be ready to say specific things or use a script when disclosing their diagnosis to others. When asked how they would describe Turner Syndrome, ILWTS used phrases such as *"This is the speech I give whenever I tell people about it"* (ILWTS, Participant 17) or "*I always describe it to my friends as like…. That's how I normally explain it"* (ILWTS, Participant 10) or "*I usually would describe it as….*" (ILWTS, Participant 15). These responses indicate a need to share the diagnosis in a specific way which ILWTS believe is functional and does not interfere with their relationships with peers. These 'typical' explanations or 'set speeches' appeared to frame the diagnosis in a specific way which facilitated maintaining a sense of control over and promoted a sense of normality during disclosure to others.

3.1.2 Coping with infertility

This theme describes the influence of infertility on the disclosure process and the different impact it may have on ILWTS and family members, based on evolving expectations about the ILWTS forming a family.

Parental expectations

Most family members held specific expectations about their child's future family life and reported the point of disclosure as the shocking realization that these could not be met due to the common symptom of infertility. A family member said: *"You've spent 15 years with a certain view of things… Because of the amount of time, the number of years you've been holding consciously or not an idea in your head, including an idea of what might happen."* (Participant 4, Mother). This realisation appeared to have a devastating impact on family members who stressed it as a barrier to disclosure. A family member said:

> *The absolute heart-breaking issue which still remains, erm, was to learn about her infertility, and picturing her not having that, erm, and then how do you tell her that, I never ever thought that as a mother, I would be in the position to tell my child about her fertility, that just never crossed my mind, when I became a parent.* (Mother, Participant 3).

These quotes highlight the conflict experienced by family members due to shattered expectations and hopes for their children's lives and future. Parents assumed that the ILWTS shared the same expectations and would struggle to come to terms with infertility, therefore initially avoiding revealing information about the diagnosis. However, ILWTS reported different views about this aspect of the condition. An ILWTS said:

> *Sometimes when I read stuff, and it's mothers of very very young children, they get very upset that you know they're not going to have their own family. And I'm reading that and I'm thinking, your child is two years old please do not be putting that pressure on them because you don't know how they're going to want to live their life.* (ILWTS, Participant 12)

This quote implies that not all ILWTS may share the same vision and hopes about their future family lives and may not even wish to have children. The erroneous assumption that this would always be the case can elicit significant distress in parents which, in turn, may delay the disclosure process or be transferred to ILWTS and elicit a negative impact during disclosure.

Age-related impact of infertility

Participants reported the different impact infertility can have in different stages of their lives. An ILTWS described her feelings when learning about infertility at a young age:

> *It didn't really impact me too much when I found out about the growth hormone, obviously as a child I was more like, urgh, why do I have to have these stupid injections, but then it was more when I was told about the infertility side of things. I was still really young, so it wasn't as much of an issue at that point, but it sort of immediately made me feel different to my peers.* (ILWTS, Participant 1)

This response illustrates the major concerns of ILWTS around infertility during early adolescence, which is a common age of disclosure. Socio-emotional needs appear to be more prominent during this developmental stage, with particular emphasis placed on 'fitting in' and not feeling different from peers, to avoid potential stigma and feelings of impairment resulting from infertility. This challenge, along with concerns during adulthood about forming one's own family, could be addressed through the timely provision of information on alternative solutions. An ILWTS said:

> *I think one aspect of information that just isn't widely available out there is about infertility and the options that are there for you … I find it very hard to sort of understand what the options are and what each option actually entails, so like how to go about actually doing it, how long it takes, what the risks are, erm, what chance is it of actually being successful, things like that … That information just isn't, isn't widely available.* (ILWTS, Participant 8)

This quote highlights the need to inform ILWTS about alternative options, such as in vitro fertilisation (IVF) and adoption, for building a family. At earlier stages, such as childhood or early adolescence, relevant information provided by family members in an age-appropriate manner could counteract potential feelings of impairment, while at later stages, such as late adolescence and adulthood, professional healthcare services could provide the relevant advice and expertise to help ILWTS make informed decisions about their future.

3.1.3 Awareness of Turner Syndrome and its impact

The final major theme encompasses the need to raise awareness in the public and promote knowledge and understanding of the condition and the impact of diagnostic disclosure among healthcare professionals, to prevent negative stereotypes and stigma resulting from TS disclosure and enable ILWTS and family members to receive strengths-based condition-specific advice and support.

Negative portrayal of the condition

This subtheme focuses on the negative perceptions of TS unintentionally promoted through online resources and informational leaflets. Participants emphasised the impact of the negative portrayal of the condition in informational resources used at the point of or soon after disclosure:

> *I don't like [it] when people just label the symptoms down, because that's not treating [you] like a human, do you know what I mean? So yes, say some of the symptoms potentially because obviously it's raising awareness to what we deal with, but don't describe us like monsters cos we're not monsters.* (ILWTS, Participant 17)

This response illustrates the negative impact of focusing exclusively on symptoms when providing resources for ILWTS and family members to help them understand the condition. This approach was perceived as disempowering and dehumanising, as it focused on the symptoms and impact of the condition, rather than the person. A family member added:

> *They gave me a booklet and told me not to look in the back of it, it was just such an outdated booklet because it made me feel like there wasn't gonna be any quality of life ... We came away and, and, and we were devastated, you know cos like I said cos we had this booklet, and, and it just didn't paint a very good picture of, of what life was gonna be like. Obviously, we know now that that was worst case scenario, but they never told us, they just said there's a booklet and that was it really.* (Mother, Participant 23)

This quote reiterates the importance of a sensitive strengths-based approach when disclosing information about the condition to ILWTS and family members, particularly

as experiences of TS can be unique and vary across individuals. Focusing on how to live well with the condition could prevent the underlying sense of hopelessness depicted in worst case outcomes and extreme presentation of symptoms. An ILWTS provided further insight into the need for professionals and parents to place emphasis on the strengths of the person when sharing information about the condition, to facilitate acceptance and empowerment and promote a positive outlook: *"I would say put it in a positive way, when you tell them, just kind of show them that they can live life, with it and enjoy life as much as if they didn't have it."* (ILWTS, Participant 11).

Participants also expressed the fear of potential discrimination and stigma resulting from disclosure of the condition. A family member described the family's hesitation to share information about the condition with others due to the impact this could have on the social interactions of the ILWTS:

> *Our concern with sharing was, if you went and googled TS what comes up is your nipples are widely spaced, you don't get your period and you can't have children. That could be one interpretation of TS, so our concern with being open was if a nasty child got hold of that information like one of her peers and then googled it what, what they would do with that information.* (Mother, Participant 3)

This quote highlights how the typical negative portrayal of TS in public resources, focusing exclusively on symptoms, may indirectly result in discriminatory behaviour against ILWTS due to societal or cultural norms on appearance and lack of awareness and education in the public.

Person-centered professionals with knowledge and expertise

This subtheme represents the challenges experienced by ILWTS and family members during the diagnostic process, due to limited awareness and understanding of the condition and the impact of diagnostic disclosure by healthcare professionals. An ILWTS reported how the general practitioner repeatedly dismissed the family's concerns around physical development before agreeing on a referral: "*My mum noticed that everyone else was growing and I wasn't, so after like numerous time of taking me to the GP and they were like oh there's nothing wrong she'll grow, she managed to persuade them in the end."* (ILWTS, Participant 10). Once referral led to a diagnosis, participants experienced difficulties with understanding the condition

during disclosure from medical professionals, due to the provision of limited vague information which elicited confusion. An ILWTS said:

> "*They explained the basics, but they didn't really say, what it is or why it is, it was down to luck that I had a really good biology teacher who understood cell structure and took an interest in my condition and took the time to take me aside and explain it to me. Otherwise, I don't think today I would've had as good an understanding of what it is that I've got … And I just don't think that it should be down to luck you know, it's something that should be provided at every Turner's clinic."* (ILWTS, Participant 16)

This quote illustrates that during diagnostic disclosure, ILWTS and family members need to consult knowledgeable healthcare professionals who can take the time to provide clear information about TS, to enable those affected by the condition to understand, accept and live confidently with TS. However, participants also underlined the need for person-centred interactions with attentive healthcare professionals who are mindful of individual needs and understand the varying impact that diagnostic disclosure may have on different individuals. An ILWTS described: *"The consultant should pay attention perhaps to what people want a little bit because some people will not want that information will they? … If you're telling something big that they've got TS, then you should also make sure you've got the time to do that."* (ILWTS, Participant 2). In contrast with the previous quote, this response highlights that professionals should carefully consider the amount and nature of information revealed during diagnostic disclosure, as the degree of preparedness to accept and take in information that may be distressing or perceived as life-changing can vary across different individuals. A family member reiterated the importance of supportive and sensitive professionals who understand the impact of disclosing the diagnosis: *"I should not in any way have been in that appointment on my own ... They may be giving that information every day to people, but they were giving us life-changing information for our child that we were not expecting, and I think to not take that seriously is really unfair."* (Mother, Participant 3). In this response, participants highlighted the need for professionals to consider the needs of all those affected by the condition and prepare them accordingly to reduce the impact of disclosure. The family member expressed the need for the spouse's presence and support during the medical appointment, so that she could share the shock and distress resulting from diagnosis.

4. Discussion

This study aimed to explore the experiences and needs of ILWTS and family members around diagnostic disclosure and identify the barriers and facilitators involved in the process. Through individual in-depth interviews with 16 ILWTS and 8 family members, we identified three major themes representing fundamental aspects of the disclosure process and associated needs: ‘guardianship of disclosure’, ‘coping with infertility’ and ‘awareness of Turner Syndrome and its impact’.

Our study findings indicate that typically there is a transition in the ownership and management of information about the condition. Initially the family member is the guardian of information until a certain point at which this responsibility passes to the ILWTS. However, our findings demonstrate that there are mixed views as to when this transition should occur. Consistent with Sutton et al. (2006), many parents wished to protect ILWTS from potential stigma or feelings of impairment by concealing the diagnosis, delaying disclosure and controlling who has access to information about the diagnosis. ILWTS expressed different views about disclosure. Individuals who had a positive early experience of disclosure showed a preference for taking ownership of the disclosure process and deciding who they share their diagnosis with. This seemed to be prominent during adolescence as part of their ongoing growth and sense of independence. On the other hand, ILWTS with negative experiences of disclosure seemed to prefer a more controlled process of gradual release of information both to themselves and others, to ensure minimal impact on self-concept and social relationships.

Many family members reported uncertainty regarding the right time to initiate the disclosure process and start sharing information about the diagnosis with ILWTS. Previous studies have suggested an age-appropriate disclosure with gradual release of information, to facilitate a positive experience (Nisbet et al., 2022; Sutton et al., 2006). Our study expands on these findings. The age of disclosure in our sample varied between 4 and 16 years of age, except for one participant who learned about TS at the age of 18 and another one at the age of 25. Although in some cases an age-appropriate disclosure took place in the context of creative playful activities at an early age, quite often parents felt distressed and perplexed as to when and how disclosure should be initiated, resulting in significant delays in fear of a devastating outcome. Most of ILWTS reported that parents’ confusion could be alleviated by ‘following the

lead' of their children and noticing signs and opportunities that may indicate the right time to start disclosing information about the condition. Questions around sex education and slower development were commonly reported as triggers during adolescence. Similar to the approach taken for early disclosure within a playful environment, these discussions did not require a formal 'big sit down' event but rather a discussion within usual daily activities to normalise the experience of disclosure and minimise its impact on ILWTS. This finding seems to be consistent with previous studies which showed that placing the disclosure of HIV within a familiar non-threatening context facilitates processing and relating to the diagnosis and minimises feelings of impairment (Zanon et al., 2017). To our surprise, family members in our study did not report consulting healthcare professionals about the disclosure process nor did they consider a joint disclosure to ILWTS as suggested for other genetic conditions (Gallo et al., 2005). Most parents felt the need to take on the full burden of disclosure, indicating a strong sense of responsibility and a perceived lack of appropriate support.

An important aspect of disclosure was related with the gradual release of information across different developmental stages. Nisbet et al., (2022) reported age-appropriate progressive disclosure from an early age as a key factor in positive experiences of diagnostic disclosure, arguing it can minimise distress and facilitate adapting to living with TS. Infertility was most commonly one of the TS symptoms disclosed at more mature developmental stages (Nisbet et al., 2022; King et al., 2016). Our study provides different insights into this aspect of disclosure. Our participants reported that disclosure at an early age through creative playful activities facilitated acceptance and normalisation of TS symptoms, including infertility and short stature. The process was reinforced as ILWTS grew into adolescence with more subtle information about the condition and its symptoms. When early disclosure did not take place during childhood, careful consideration of the needs of ILWTS and their preparedness to process and accept the relevant information was required by both parents and professionals. This could be achieved by paying close attention to the reactions of ILWTS during gradual disclosure, to determine the right amount of information to be disclosed at each stage.

Another important aspect affecting the disclosure process was related to the impact of infertility, which is a common symptom of TS (Gravholt et al., 2017). Consistent with previous studies on TS and other genetic conditions (Close et al.,

2016; Hallberg et al., 2010; Middleton et al., 2018; Sutton et al., 2006), many parents experienced significant distress and feelings of sorrow due to shattered expectations and hopes for their children's lives and capacity to form a family. The underlying feelings of grief, loss (Wormer, 2019) and potential guilt for passing on a genetic condition (Middleton et al., 2018; Gallo et al., 2005) often led to delaying disclosure until the diagnosis was not possible to conceal from ILWTS. This was due to parents assuming that learning about TS-related infertility would have a devastating impact on their children. However, this was not always the case, indicating potential intergenerational differences in views around motherhood (Maher and Saugeres, 2007; Meyers, 2001). Similar to Nisbet et al. (2022), we found that the concerns of ILWTS about infertility were primarily focused on potential stigma rather than the capacity to have children. As adolescence was a common age for disclosure, ILWTS primarily worried about potential feelings of pity or rejection by their peers (Kaushansky et al., 2017). Our findings seem to be consistent with previous studies on HIV or autism, which showed that fear of stigma and discrimination is one of the main barriers to disclosure of diagnosis to others during adolescence (Humphrey and Lewis, 2008; Michaud et al., 2009). Of interest, ILWTS described how they often prepared a 'speech' or description of the condition before disclosing their diagnosis to others. Although it was not explicitly stated, it is possible that this was part of an attempt to normalise the condition in the context of social relationships and facilitate acceptance by peers or prevent stigma and breakdown of friendships. In line with Sutton et al. (2005), ILWTS in our study appeared to be comfortable with considering alternative options, such as IVF and adoption, and highlighted the benefits of having easy access to clear relevant information, particularly during adulthood. Including these alternative options for infertility in TS descriptions may have been perceived as a buffer to potential stigma, depictions of impairment and breakdown of social relationships.

An important theme in our findings was associated with awareness of TS and its impact among healthcare professionals and the public. Both parents and ILWTS emphasised the need for TS to be positively portrayed in informational and publicly available resources through a strengths-based approach. Overfocusing on symptoms was perceived as disempowering, stigmatising and dehumanizing, and instilled a sense of hopelessness and permanent impairment. Although no actual experiences of stigma were reported, it appeared that participants experienced self-stigma in the

form of apprehension or anticipation of exposure to stigma (Bos et al., 2013) due to the negative portrayal of the condition in public resources or during diagnostic disclosure by healthcare professionals (Sutton et al., 2006). Indeed, previous studies have reported that many parents encouraged their children to avoid disclosing their diagnosis to anyone beyond the immediate family, to avoid stigma (Ergin et al., 2018; Nisbet et al, 2022). Parents experienced a conflict between normalising the condition within the family and protecting ILWTS against discriminative behaviours resulting from widespread stigmatising norms around illness and the symptoms of TS. Our participants emphasised that public information focusing on the strengths of the person and how to live well with the condition could facilitate empowerment and social acceptance, promote a positive outlook, and reduce potential stigma.

Participants also stressed the importance of engaging in person-centred interactions with sensitive professionals who understand the impact of disclosure and are considerate of both ILWTS and family members' needs. Similar to previous studies (Sutton et al., 2006), family members often felt unheard by professionals. This was either due to professionals dismissing their concerns about their children's development or because of limited time during medical appointments. Similar concerns have been previously reported by family caregivers (Hallberg et al., 2010; Roach et al., 2008) indicating a potential lack of knowledge among general practitioners and systemic flaws within the design and delivery of primary and specialist healthcare services. Parents also wished to have advance knowledge of the potential impact of diagnosis, so that they can be accompanied by their spouse or another family member with whom they could share the shock and burden of diagnosis. In line with previous studies, ILWTS underlined the importance of clear up-to-date information about the condition (Nisbet et al., 2022; Turner and Hozjan, 2019) and avoiding heavy jargon during medical appointments to prevent confusion. Previous studies have stressed the negative impact of outdated information about genetic conditions (Waxler et al., 2013) or forcing service recipients to find information about the condition themselves (Close et al., 2016). Contrary to Nisbet et al. (2022), our ILWTS participants did not report positive experiences of diagnostic disclosure by clinicians. Our findings are more in line with Sutton et al. (2006), as our participants highlighted the uniqueness of individual TS experiences and the need for healthcare professionals to consider age-related needs, individual circumstances, the amount and nature of information revealed, and the way diagnosis is conveyed, to prevent

overwhelming experiences of disclosure. Complex medical terminology and excessive details about TS during the initial delivery of the diagnosis were key factors eliciting confusion and a negative outlook.

Finally, our findings provide some additional insights into the way ILWTS experience or view TS as part of their identity and personhood. Nisbet et al. (2022) reported that part of normalising the condition occasionally included ILWTS and family members distancing themselves from the symptoms to minimise the impact of TS on self-concept and identity. Although we also received similar responses from some family members, our findings indicate that ILWTS wish to embrace and take ownership of their condition as part of their identity. Having a clear understanding of TS and how to cope with symptoms appeared to facilitate gaining confidence and control over the impact of the condition and the way ILWTS choose to disclose their diagnosis to others. Although some ILWTS reported their wish to not be consumed by the diagnosis, the underlying message in the majority of responses was that ILWTS were proud of their newly formed identity as people living with TS and wished to amend public misconceptions around the condition and help others live well with TS.

4.1 Strengths and limitations

Our study has certain strengths and limitations. It included a PPI group comprising an ILWTS and a family member who actively contributed to the study to ensure the appropriateness of the study design and materials (e.g. feedback on recruitment materials and data collection methods and instruments), enhance the study quality and impact (meetings for feedback on progress and upcoming stages of the study), and increase the credibility of the study findings (feedback on plausibility and relevance of findings) (Tomlinson et al., 2019). We also held fortnightly research team meetings to ensure the rigorous implementation of the study, monitor the safety and well-being of participants and the PI, and ensure the credibility of the study findings. During the first stages of analysis, all members of the team were involved in coding and active discussions to ensure consistency. The evolving and final themes and thematic map were regularly discussed within the research team. Additional feedback was sought and received from the study PPI group to enhance confirmability of the findings.

We purposively chose to recruit both ILWTS and family members to gain an in-depth understanding of diagnostic disclosure of TS, synthesise new knowledge from the perspectives of those involved in disclosure, and ensure that both voices are represented in the findings. Due to the sensitive nature of the topic, we interviewed ILWTS and family members separately to ensure open honest discussions and avoid concerns about the impact of potential criticisms on family relationships.

Due to the COVID-19 pandemic, all interviews took place over the phone or Skype. It is thus possible that we may have missed some contextual data resulting from non-verbal cues and body language. Although we tried to recruit participants through a wide range of third-sector organisations, only TSSSUK accepted to act as a gatekeeper for the study. Despite our efforts to recruit a diverse sample of participants, ILWTS and family members from ethnic minority communities did not take part in the study, which may limit the transferability of the findings. The age of participants ranged from 16-20 to 50-60. Although this may have allowed capturing a wide range of experiences, we cannot eliminate the possibility of recall bias in some of the collected responses. Nevertheless, our study included a considerably large sample of participants with a wide age range of diagnostic disclosure, which increases our confidence regarding the credibility of the findings.

4.2 Reflexivity

The PI has a background in psychology and is a Professional Doctorate student in Psychological Therapies. She has previous experience in qualitative research and conducted all the interviews. Prior to data collection, the PI had no previous relationship with the study participants.

As the PI lives with TS, the research team engaged in detailed discussions before the beginning of the study as to whether the PI should disclose her condition to participants. Upon consulting the relevant literature and considering the needs and preferences of the PI, all members of the team were in favour of advance disclosure on the grounds of openness and honesty with participants. The PPI group was also consulted and agreed with this approach. It was therefore decided to include the relevant information in the study participant information sheet. Although we anticipated that this would facilitate building rapport with participants and creating a safe space

for the disclosure of sensitive experiences, the PI used a reflexive journal throughout data collection and analysis to eliminate the possibility of bias resulting from personal experiences or pre-suppositions and increase the trustworthiness of the findings (Dodgson, 2019). We also held fortnightly research team meetings to discuss ttentative results and consider different interpretations before finalizing the thematic map by consensus. Furthermore, all members of the team were involved in coding and active discussions during the first stages of analysis, to ensure triangulation, consistency and agreement (Shenton, 2004). Additional feedback on the relevance and coherence of the findings was sought and received from the study PPI group.

During data collection, the PI recorded further observations in the reflexive journal to consider whether participants' awareness of her condition facilitated or hindered the disclosure and truthfulness of personal experiences reported during the interviews. One family member and one ILWTS shared potential concerns; the family member initially felt worried of eliciting potential distress, due to providing negative accounts which may resonate with the PI's own experiences of TS disclosure; the ILWTS enquired about the PI's feelings when listening to negative experiences of other people living with the condition. In both cases, the PI reassured participants that: (a) her primary interest was in exploring other people's experiences in depth, without any judgement or preference, (b) she maintained a curious perspective that would enable her to better understand other people's experiences and help those who may learn about their TS diagnosis in the future, and (c) if at any time participants felt uncomfortable during the interviews, they could take a break and consider whether they would like to continue, without any consequences if they chose to withdraw from the study. Both participants took part in the study without any further issues observed or reported.

Due to the COVID-19 safety guidelines and measures, all interviews were conducted via telephone or Skype. The PI had limited experience of remote interviewing, which instilled a slight sense of nervousness during the first two interviews and may have affected the collection of the relevant data. Although this did not appear to be noticed by participants, the PI used the reflexive journal after each interview to reflect on the relevant issues. These were discussed on a regular basis within the research team to consider solutions, check progress and ensure rigour in the data collection process. The first two interviews were slightly shorter than the

remaining 22 interviews, but still produced rich useful data which were deemed appropriate to include in analysis.

4.3 Implications

Our study has the potential to inform recommendations on diagnostic disclosure of TS. As the number of studies on TS diagnostic disclosure grows, third-sector organisations can use the relevant findings and collaborate with ILWTS and family members to develop user-friendly recommendations for disclosure by parents. For instance, our study demonstrated that parents should consider age-appropriate gradual disclosure from an early age, to facilitate accepting and normalising TS. Our findings also indicate that parents need to be mindful of transferring their own feelings about infertility, as ILWTS do not always share the same concerns and are comfortable with considering alternative options, such as adoption and IVF. Parents may delay disclosure due to feelings of guilt (Middleton et al., 2018; Gallo et al., 2005), anticipated anger from their children, or assumptions about the devastating impact of infertility. However, it is keeping secrets about the health of ILWTS that elicits anger and feelings of mistrust or betrayal (Sutton et al., 2006) and place family relationships at risk. These findings can inform resources for parents' and professionals' education, particularly as parents may often turn to healthcare experts for advice and support around diagnostic disclosure.

Our study also showed that fear of stigma constitutes a significant barrier to disclosure by parents to ILWTS and by ILWTS to others. This fear seems to be related with negative stereotypes around illness and limited awareness of TS in the public. TS-specific public campaigns should aim to raise awareness by employing a strengths-based approach that focuses on normalizing the condition rather than overemphasizing symptoms.

Our findings highlight the importance of professionals' awareness regarding TS and the impact of diagnostic disclosure. Education for general practitioners could facilitate timely referrals and address parents' emerging concerns triggered by TS symptoms. A timely diagnosis could facilitate early access to post-diagnostic treatment, such as growth hormone and Hormone Replacement Therapy (HRT) and improve symptoms such as short stature (Gravholt et al, 2017). Specialist training on

person-centered diagnostic disclosure should also be offered to healthcare professionals to minimize the impact of diagnosis. Awareness of the emotional impact of disclosure could enable a family-oriented approach to reduce the shock of diagnosis on all those affected, while emphasis on strengths and living well with condition could promote acceptance, a sense of agency and reduced fear of stigma. The amount and nature of disclosed information should also be considered on a case-by-case basis. Information should be shared in a sensitive, person-centred and age-appropriate manner, according to the person's age, individual and family circumstances, and preparedness to process and accept the diagnosis. In some cases, detailed information would be more beneficial to facilitate understanding, whereas in other cases a more staggered approach could prevent overwhelming experiences of disclosure. Healthcare professionals should also be supportive and sensitive to the person's needs throughout diagnostic disclosure, explain the diagnosis in lay terms, dedicate sufficient time to address the concerns of the whole family, and be ready to signpost to relevant services for post-diagnostic treatment and psychosocial support. Social support groups for families affected by TS could be particularly useful for the latter and enable parents to prepare for diagnostic disclosure by learning from others with previous lived experience.

The unique experience of TS indicates the need to advocate for the development of condition-specific recommendations on disclosure. These could be included in the National Institute for Health and Care Excellence (NICE) guidelines. Similar recommendations already exist for other chronic conditions with life-changing implications, such as Motor Neurone Disease (NICE, 2016), Cystic Fibrosis (NICE, 2017a) and Parkinson's Disease (NICE, 2017b). Our findings provide in-depth insights into the barriers and facilitators to diagnostic disclosure and may constitute a useful basis for future recommendations on disclosure of TS.

Despite the increasing knowledge resulting from recent studies in diagnostic disclosure of TS, important gaps in knowledge remain and need to be addressed. Future research could focus on experiences of diagnostic disclosure in ILWTS and family members from ethnic minority communities, particularly as norms and stereotypes around illness may be different in these groups (Furnham and Swami, 2018). Relevant differences may also be observed in young adults who recently became parents and may hold different views or expectations about the impact of TS and symptoms such as infertility. Further studies with ILWTS and family members with

low socio-economic background could enable identifying additional factors influencing disclosure and the support required for those affected by TS. Exploring the perspectives of healthcare professionals would also enable insights into current gaps in knowledge and practice and potential organisational or systemic barriers to effective diagnostic disclosure. Finally, studies examining self-stigma in ILWTS and other family members could inform the development of family-oriented interventions, which could then be tested and become available through statutory services if effective.

5. Conclusion

To the best of our knowledge, this is one of the very few studies exploring diagnostic disclosure of TS. Our findings indicate that age-appropriate disclosure may facilitate normalising the condition and minimise potential self-stigma. Gradual disclosure initiated at an early age can be particularly useful to this end. Our study also highlights the need for person-centered strengths-based approaches in TS service provision and public campaigns, to minimize social stigma, meet the unique needs of those affected by TS and enable them to live well with the condition.

Conflicts of interest

No conflicts of interest to report.

Funding

This study has not received any funding.

Introduction to Thesis Chapter 3

Following the results and findings of the empirical article, chapter 3 will focus on the critical reflection of the research process. In particular, reflection upon the unique position of the principal researcher having personal experience of living with the condition being studied. This will involve critical reflection upon the lessons learnt by the researcher throughout the process, to provide recommendations for future researchers. The lessons learnt included: positionality, self-disclosure, researcher personal knowledge and assumptions, the emotional impact of exploring one's own condition, and the influence of involving a PPI group. Previous research has found that advantages of this perspective involved the researcher having prior insight and understanding of the condition. This means that there is an increased ability to build a rapport with the participants due to a sense of mutual understanding. Nevertheless, disadvantages of this perspective include personal prior assumptions. For example, the researcher may believe that they hold an 'insider' position within the group/community being studied, when the participant may view them as more of an 'outsider'. This may influence how the participant interacts with the researcher, for example, disinhibition, or social desirability leading the participant to mispresent their experience to avoid judgment or embarrassment.

Gibb's Reflective Cycle was used to help express the lessons learnt and reflections made within the research process. The six stages of the cycle include:

1) Description of what happened.
2) Feelings of what you were thinking and feeling.
3) Evaluation of what was positive and/or negative about the experience.
4) Analysis of the sense you can make around the situation.
5) Conclusion of what else could have been done.
6) Action plan to reflect upon what you would do differently next time.

This cycle was used to provide a structure and points to refer to in a very personal and strongly reflective piece of writing. The selection of Gibb's reflective framework allowed for a more in-depth reflection, for example, compared to the model of Rolfe et al., 2001. This is because the model of Rolfe offers only three questions, 'What?, So What?, Now what?', compared to Gibb's which explores the feelings of the

researcher at the time. Furthermore, Gibb's advocates that the framework be used to generate transferable lessons for others. (Williams, Woolliams, & Spiro, 2020).

Chapter 3. Conducting research into one's own condition: Lessons learnt from reflections on a study exploring diagnostic disclosure of Turner Syndrome

1. Positionality

The positionality of a researcher reflects how they position themselves in relation to participants and how participants position themselves in relation to their perception of the researcher (Chavez, 2008). Scholars suggest a bilateral distinction as an 'insider' or 'outsider' (Ross, 2017). An 'insider' researcher is considered as an individual who identifies with or is a member of the groups or individuals whose experiences are being studied (Ross, 2017). An alternative description refers to the study of one's own social group (Green, 2014). In contrast, an 'outsider' researcher is viewed as an individual who does not have prior personal knowledge of the community or the members whose experiences are being studied (Green, 2014). Nevertheless, there has recently been a movement towards differentiating the levels of an 'insider's' position, namely distinguishing "total insiders" from "partial insiders" (Ross, 2017). A 'total insider's position involves researchers sharing multiple social identities (e.g., race, ethnicity) or profound experiences (e.g., wars, family membership) with the study participants, whereas partial insiders only share a single (or few) identity (Chavez, 2008). Upon reflection on this typology, I considered myself as a "total insider" for this study since I shared specific social identities with ILWTS (i.e., gender, living with TS), as well as profound experiences of diagnostic disclosure, adherence to required treatments such as growth hormone, and the socio-emotional impact of living with TS and coping with symptoms such as short stature.

I anticipated that my position as an 'insider' would facilitate building rapport with participants (Ross, 2017) and enable me to better understand and depict their experiences (Rooke and Rooke, 2015). However, it soon became evident that my position also carried a risk for potential unconscious bias. During our early research meetings on study design and data collection instruments, I realized that I had pre-existing assumptions regarding the challenges involved in TS disclosure due to the negative connotations attached to my own experiences of disclosure and those of my parents. They had gradually shared with me information about TS with very little support from statutory services. As a result, I had erroneously assumed that this would be the case for most ILWTS and parents (in contrast, see Nisbet et al., 2022 for some examples of positive experiences of disclosure) and expected to receive accounts of significant challenges experienced during disclosure. This realization highlighted the need to monitor and reflect on the potential influence of my own experiences and

emotions on my role as a researcher throughout the study, particularly during data collection and analysis. I considered this approach pivotal for the delivery of a rigorous study and the generation of new credible knowledge that would represent participants' voices. As a research team, we decided that I should use a reflexive journal throughout the study and establish a study Patient and Public Involvement (PPI) group, to ensure that: (a) my own experiences and pre-suppositions about TS diagnostic disclosure would not influence the design and delivery of the study, and (b) my previous experiences would not overshadow participants' voice in the study findings.

Furthermore, I soon realized that I could also be considered as a "partial insider" by family members who took part in the study, since they provided the perspectives of a parent which I had not experienced personally. It is possible that this position may have led some parents to view me as an 'other daughter' rather than a researcher. A relevant account was provided by a family member "*I'm saying things about my daughter that you're... you know that, that's you as well isn't it"* (Participant 9, Family member). This demonstrates that parents' awareness of my condition may have led them to believe that I would be at risk of feeling hurt or distressed when hearing about their experiences. It also highlights their empathy and their fear that I could make potential associations with my own experiences of disclosure and reconsider, or even doubt, the approach of my parents and their experiences when disclosing to me information about TS.

2. Disclosure of diagnosis

Studying a condition of which one has lived experience raises concerns as to whether the researcher's diagnosis should be disclosed to participants. One could argue that disclosing the diagnosis could make participants feel more comfortable and willing to disclose in-depth information about their experiences, as they feel a sense of connection and rapport with the researcher and anticipate a non-judgmental behaviour (Hoffman and Barker, 2017). Alternatively, participants may fear comparison of experiences or potential feelings of inferiority resulting from poor personal experiences and may withhold information or misrepresent their experiences due to social desirability (Hoffman and Barker, 2017). Of interest, previous studies conducted by researchers sharing the same condition with participants revealed

conflicting beliefs. In one study into type two diabetes (Hoffman and Barker, 2017), the researcher decided not to disclose their own diagnosis of diabetes 1 which they received at six years of age. The researcher felt that disclosing their diagnosis would result in struggling to manage the desire to provide advice to participants, if they expressed difficulties similar to theirs. Conversely, in a study on inflammatory bowel disease (Murphy et al., 2022) the researcher, who had only received the diagnosis a few years prior to the study, decided to disclose the diagnosis as they believed and found it helped them to develop rapport with participants.

The personal dilemma around self-disclosure further includes aspects related with ethical research, which require careful consideration of personal beliefs, expectations and motivations, as well as the need for transparency with participants unless there are risks related with the credibility of findings, participants' partaking or the researcher's wellbeing. Dickson-Swift et al., (2007) argue that researchers should carefully consider the level of disclosure they feel comfortable with and mentally prepare for the process. They highlight that self-disclosure can be used to promote a sense of equality and trusting rapport with participants but may also place the researcher in a vulnerable position and influence data collection. For instance, participants' awareness of the researchers' diagnosis may lead to potential transference and expected demonstration of associations with their own experiences. Dickson-Swift et al., (2007) recommend that ethics committees should conduct a risk assessment to ensure the researcher's awareness of and capability to cope with such risks when exploring sensitive topics, including the availability of contingency plans and support as necessary. In our study, we decided to disclose my diagnosis after an in-depth discussion with the supervisory team before the ethics application process. I was diagnosed with TS before birth and I have lived with and known about TS from an early age. I, therefore, felt comfortable with participants' awareness of my condition and further wished to be open and honest. We decided to include information about my condition in the study poster, to ensure open and honest communication from early on and help prospective participants to establish a sense of relevance and comfort with discussing sensitive experiences. Upon reflection, I feel that I could have been more prepared mentally (Dickson-Swift et al., 2007) to address potential participants' concerns about my condition, as I pre-assumed that self-disclosure would only have a positive impact and was caught off guard in two cases. These included a family

member and an ILWTS who raised potential concerns about discussing their experiences with a researcher living with TS. The family member stated:

> *I'll tell you what's hard actually Emma, talking to you about it (laughter) that's, do, do you find that hard? Is it sort of, is it sort of I'm saying things about my daughter that you're, you know that, that's you as well isn't it, that's making me sort of think that, it could be hard for you to listen to that it's, it's, it's not, or is it?* (Participant 9, Family member).

When the participant highlighted this concern, I initially panicked and was unsure how to respond. I took a moment to form a response and I then attempted to reassure them that my role was solely to gather information and listen to their experiences as a researcher. I also checked that they were comfortable to continue with the interview:

> *Interviewer: No, I mean I am, I do have Turner Syndrome myself but I'm also, I'm not taking anything personally that's said, I'm just interested to find out about people's experiences. Are you, are you comfortable to carry on?*
>
> *Participant: Yeah, I'm absolutely comfortable to carry on, I just wouldn't want to upset you (laugh).*
>
> *Interviewer: Thank you for considering that, for me it's more just erm, as you said, a learning experience.*

Although I felt that I handled the situation relatively well and did not detect any influence of my response on the data collection process (the participant appeared to be happy to continue with the interview without any concerns and discussed their experiences in detail), I later discussed this incident with the supervisory team. After an in-depth discussion, we formulated a response that I could use if this were to occur again, to ensure that data collection would not be affected, and I would feel comfortable with addressing similar concerns. The response focused on my curiosity and genuine interest to explore other people's experiences of disclosure without any preference or judgement, so as to help families receiving a TS diagnosis in the future. I also informed participants that if at any time they did not feel comfortable, they could pause or stop the interview and withdraw.

The above incident, albeit unexpected, enabled me to feel more confident with addressing similar concerns by subsequent participants. I was satisfied that

participants felt comfortable with sharing their concerns and experiences openly with me as a researcher living with TS, and my prepared response helped me to ease the concerns of an ILWTS:

> *Participant: The only other thing that I just wanted to, kind of, make sure of is that like, you're okay with it because obviously I'm sitting here, talking about it positively but, you might not think, the same, and you know how, how, are you okay with it?*
>
> *Interviewer: Thank you for considering that, I appreciate it. I am an individual with Turner Syndrome but for this I'm also taking a role as a researcher and learning about people's experiences, I'm not kind of taking anything personally if that makes sense. I'm just here to listen and learn about people's experiences.*

I felt that the responses I provided to participants were genuine and based on a bracketing approach, to set aside any prior assumptions of mine and capture participants' voices. In line with previous literature, the transparency regarding my background and interest was useful to this end (SAGE, 2021). I also used a reflexive journal immediately after each interview and during analysis, to document my observations and discuss these with the supervisory team. As I was aware that I would not be able to suppress my personal experience and emotions, self-reflexivity enabled me to monitor, manage and feel more comfortable with my dual role as a researcher and an individual living with Turner Syndrome (Pillow, 2003; SAGE, 2021).

Of note, participants appeared to be more concerned about my personal well-being when hearing about their experiences. Both the family member and ILWTS seemed to wish to protect me from negative and positive connotations of their experiences, acknowledging a vulnerability resulting from TS disclosure and projecting that vulnerability on me, who in their eyes appeared to me more of an 'insider' ILWTS rather than a researcher. I did not further explore this to avoid interfering with the data collection process, but future research could provide useful insights and shed light on how participants relate with researchers who live with the same condition and the relevant implications for qualitative data collection processes. Exploration of similar concerns could also be conducted pre- or post-interview, through brief open-ended questionnaires or interviews focusing on participants' views, feelings and attitudes regarding how they feel about discussing their experiences with someone living with

the same condition and the underlying reasons. Benefits of other self-disclosure processes have been previously reported by Rosenberg and Tilley (2021), who explored how transgender individuals felt about discussing their experiences with transgender researchers. Their follow-up interviews with six participants revealed a feeling of mutual understanding and sense of trust regarding the researchers' intentions and motivation for conducting the study, which facilitated developing and maintaining rapport.

3. Personal knowledge, understanding and assumptions

Previous research has demonstrated some advantages of conducting research into one's own condition. One example of such an advantage is the researcher's prior knowledge and insight into the condition (Hofmann, and Barker, 2017). This may help participants to discuss their experiences openly with someone with a shared understanding (Dwyer and Buckle, 2009). In our study, some ILWTS participants highlighted infertility very early in the interviews when responding to the opening question (i.e., 'How would you describe TS?'). An ILWTS said: *"If I was describing it to someone else, I would say it's a genetic condition. It's why I'm short… and then if it was a closer person then I might tell them about the infertility that goes with it. And for anybody else they don't need to know any more than that…"* (Participant 2, ILWTS). Individuals may prefer to avoid raising sensitive and potentially distressing issues, such as infertility, with someone who does not have prior understanding. In our study, many participants appeared to be comfortable with sharing or discussing sensitive aspects of their experiences from the beginning of the interviews, potentially due to speaking with a researcher who understands what it is like to live with TS. However, not all participants provided similar responses, as some were more hesitant than others. Another ILWTS provided the following response to the same question:

> *A genetic condition which only effects females, erm, and it's, it's to do with the development of the, erm, chromosomes and cells at conception, a female with, erm, Turner Syndrome will then either be, either missing an X chromosome or part of erm an X chromosome, erm, and then that can then erm, lead onto erm, other kind of symptoms and side effects that go along with the condition such*

as, erm infertility, erm, you know lack of growth hormone. (Participant 14, ILWTS).

Similar to the quote above, some participants responded to this question by providing a generic description of physical symptoms and the genetic explanation of TS, indicating a potentially erroneous pre-assumption of the 'total insider' position, which may require more time and rapport to be established with participants. I did not explore participants' responses to the opening question further, as I would have responded in a similar manner, and this was not the focus of the study. Yet, with further exploration this could have provided a unique insight into why participants chose to describe TS in that way to a researcher living with the condition, and how that description may relate with evolving participant-interviewer dynamics during the interview. Such insights could provide useful contextual information and elucidate the perception of the researcher by participants.

My preference to focus on TS in my study could also indicate an underlying personal need to better understand the impact of the condition and my experiences with disclosure. On reflection, this personal investment meant that although I genuinely cared and wanted to make a difference in the lives of those affected by TS, I also wished to normalize and alleviate any negative connotations of TS. Before the data collection process, we became aware of the potential link with my own previous experiences and engaged in fortnightly supervision meetings on progress and tentative results, independent coding, and regular consultations with the study PPI group, to ensure that the findings would be representative of participants' voices and not overshadowed by my previous experiences or motivations. The supervision meetings also allowed a safe space where I could discuss my personal reflections and realizations with the rest of the team, to ensure a rigorous approach to data collection and analysis. Furthermore, the supporting feedback received from the PPI group enabled an enhanced sense of confidence regarding the appropriateness of the interview schedules and the relevance of emergent and final findings. The use of a reflexive journal was also pivotal in minimizing the possibility of potential bias, particularly as I held the 'insider' position (Greene, 2014) It provided a space where I could reflect on the research processes, my feelings and thoughts during data collection and analysis, and how these may have influenced participants' engagement. The journal also enabled me to recognize early on some contradictions between

participants' accounts and my pre-existing understanding of TS and its impact, as evident in one of the journal entries:

> *In the past, when I have asked people about their experience of living with TS they have typically said positive things and that it hasn't really impacted their lives. I found it surprising to hear such a negative account. The negative experience has clearly had a lasting impact for the individual which shows the influence of early events. The participant seemed to laugh a few times throughout the interview, perhaps a defense mechanism to compensate for talking about a negative subject? Overall appeared to be open and talkative with me.* (Journal entry, 7/01/21)

This entry highlights the need for reflexive practice, so that researchers rapidly become aware of their own pre-existing understanding of the phenomenon of interest. Such approaches can ensure that data collection and analysis are not influenced by pre-suppositions and a 'total insider's' position that is erroneously assumed to guarantee a clear understanding without further exploration of participants' experiences through a curious perspective.

4. Emotional impact of exploring one's own condition

Hofmann and Barker (2017) highlighted the emotional impact of studying one's own condition as a researcher, particularly the distress experienced when hearing about the negative consequences of living with the condition. Participants' accounts of the impact of infertility triggered some post-interview reflections which were registered in the reflexive journal:

> *Infertility seems to have a significant impact for participants, both family members and ILWTS. It feels strange that it does not seem to have had as big an impact on me as others. Perhaps it's because that is all I've ever known, and I have come to accept my options of either adoption or IVF. Though it is hurtful sometimes seeing other people my age having children, and members of my family having children, and knowing it is not possible for me. It does make me wonder what my family think, as they may feel differently than the acceptance they show to me.* (Journal Entry, 25/01/21)

The supervision team were aware of the potential emotional impact of sensitive issues, such as infertility, and regularly enquired about my well-being to provide a 'safety net' of support as and when needed. The anticipated impact was enhanced by the social restriction measures resulting from COVID-19, which occasionally instigated a sense of loneliness and feeling overwhelmed. Although no significant incidents took place, the aforementioned feelings were primarily addressed through pastoral support from the research team and academic peer support networks. Indeed, previous studies have shown that working alone can have a negative impact on wellbeing (Paolucci et al., 2021) which may be counteracted through mentorship and peer support aiming to promote a sense of belonging and facilitate maintaining motivation (Watts, 2008). The support I received from my research team and peers made me feel valued and respected and served as a reminder that my well-being throughout the research process is equally important as that of participants.

5. Patient and Public Involvement

PPI is defined as an active partnership between researchers and members of the public or service users. It can promote new insights and reduce misconceptions about important topics, such as the needs of those living with a condition (NIHR, 2014). We established a PPI group prior to the beginning of the study, to receive feedback and guidance on important aspects, such as: (a) identifying the phenomenon of interest (i.e., diagnostic disclosure of TS), (b) appropriateness of the study design and methods, recruitment materials and data collection instruments, and (c) the plausibility and relevance of the findings (Tomlinson et al., 2019). The PPI also helped with managing aspects related with positionality by: (a) alleviating concerns about the 'insider' position of the researcher, (b) providing regular feedback to eliminate potential bias resulting from erroneous pre-assumptions about the study sample and the methods that should be employed, and (c) ensuring the voice of the target populations were heard and represented in the study design and findings. In our study, the PPI group consisted of an ILWTS and a family member, both of whom were consulted biannually and/or before important study milestones. This ensured that in each stage of the study there was a representative of both target populations.

6. Discussion and recommendations for future researchers

In this paper, I aimed to discuss my role as a researcher with lived experience of TS and how this may have influenced the research processes of the study. I placed a particular focus on my positionality, prior knowledge, and the emotional impact of studying the experiences of families affected by TS.

Our study has provided insights which may be useful for future postgraduate researchers aiming to conduct a qualitative study on a condition of which they have lived experience. Future postgraduate researchers may benefit from being mindful of potential pre-suppositions resulting from their own experiences of the condition and the phenomenon of interest. Such biases can influence the overall approach and questions asked during an interview, the process of analysis, and the credibility of the final findings. It is important to regularly monitor and reflect on their own experiences and pre-assumptions and consider whether these may be overshadowing participants' voices (Ritchie et al., 2013). This may be particularly useful in relation to positionality, as some researchers may erroneously assume that a 'total insider's' position will ensure a better understanding of participants' experiences. In our study, we observed inconsistencies between the researcher's previous knowledge of the condition and participants' accounts, which showcases that the 'total insider' identity may be misconstrued and requires careful follow-up exploration of participants' responses to ensure rigorous data collection and representative findings. The use of a reflexive journal, multiple coders, and validation from a study-specific PPI group can also be useful to this end.

Regarding the decision to disclose their condition or not, future researchers should carefully consider a range of factors influencing the data collection process and their own wellbeing. Disclosing the condition may facilitate developing rapport but may also elicit comparison of experiences (Hoffman and Barker, 2017; Rosenberg and Tilley, 2021) and have a negative impact on data collection and the breadth or depth of information shared by participants. Although participants appreciate honesty and openness, researchers should be prepared for unexpected questions or negative reactions to disclosure (Dickson-Swift et al., 2007), to ensure that participants feel at ease and the researcher-participant dynamics are not disturbed by disclosure. In the

current study, participants were aware of my diagnosis as they had access to the study poster and information sheet. I assumed that any concerns would be expressed when participants were offered the opportunity to ask questions about the study; however, two participants expressed their concerns during the actual interviews, which caught me off-guard and may imply that other participants may have had similar concerns which they chose not to share. Nevertheless, preparing answers for specific questions facilitated building confidence to address relevant concerns expressed by participants. Future researchers could consider explicitly stating and discussing the diagnosis (e.g. during initial contact, the consent process, and/or at the beginning of the interview), as suggested by Chavez (2008), to gain a more in-depth understanding of participants' views and preferences on discussing their experiences with a researcher living with the same diagnosis and offer alternative options for participation if needed (e.g. online qualitative surveys or interviews with another researcher). Researchers should consider the level of disclosure and the emotional impact this may have on their wellbeing throughout the study delivery. Risk assessments before the beginning of data collection may be useful to this end (Dickson-Swift et al., 2007). It is also possible that researchers may experience significant distress when hearing about negative experiences of the condition they live with, whether these resonate with their own experiences or not (Hofmann and Barker, 2017). The supervisory team should be in regular contact with the researcher, particularly during data collection, to monitor their wellbeing and provide or signpost to support appropriate to the needs of the researcher. Pastoral support from the supervision team and peer support from academic networks can further promote a sense of belonging and prevent overwhelming experiences (Watts, 2008). This support network was pivotal for me and helped me to manage the emotional impact of exploring other people's experiences of TS.

Previous knowledge of the condition and the 'insider' position may be an advantage for developing rapport and exploring specific topics in depth (Dwyer and Buckle, 2009; Hofmann and Barker, 2017) but also carries the risk of blindfolding researchers if they assume that this on its own will ensure rapport and a safe space for the disclosure of sensitive experiences. In some of our interviews, we observed the 'total insider' position being negotiated and established throughout our discussions with participants, rather than being 'offered' in advance. We also observed shifting or fluctuating perceptions of the researcher by participants. Furthermore, researchers

need to be mindful of the potential psychological mechanisms involved in their perception as ‘total insiders’ by participants. Although researchers are not expected and should not engage in therapeutic work, they need to carefully observe participants’ reactions, assumptions and expectations during interviews, to use validation of participants’ responses in a meaningful way and prevent maladaptive power dynamics.

The description of TS varied across participants in our study sample. It is possible that this may relate to their degree of trustworthiness initially attributed to the researcher. It is important to follow-up on these responses to acquire in-depth insight into evolving participant-interviewer dynamics and how these may influence the disclosure of sensitive experiences. As mentioned above, positionality did not appear to be static and researchers should be aware of the volatile nature of these dynamics, to ensure safe and in-depth explorations of sensitive topics.

7. Conclusion

In this article, I outlined the main lessons learnt from conducting research into a condition of which I have lived experience. The reflections included in this paper highlight that future researchers should carefully consider self-disclosure and the amount of information they would like to share with prospective participants in advance of the study. Depending on the decision they make, relevant preparations will be needed to address potential concerns expressed by participants. Additional caution is warranted regarding assumptions resulting from a perceived ‘total insider’ position, as a researcher’s perception of their own position may differ from that of participants. It is also important that future researchers conducting studies on the condition they live with employ different tools (e.g., reflexive journal, PPI involvement) to ensure rigor and enhance the credibility of the findings. Finally, a support network comprising pastoral and peer support needs to be put in place before the beginning of the study, to facilitate managing the emotional impact of conducting research into one’s own condition.

General Conclusion

This thesis aimed to outline current evidence on the experience of diagnostic disclosure of genetic conditions and explore in depth the diagnostic disclosure experiences of ILWTS and their parents. During the study, we identified specific needs of those affected by TS, as well as barriers and facilitators to the disclosure process. These add to current knowledge and can inform condition-specific needs-based recommendations on diagnostic disclosure of TS.

The findings of our original study present some similarities with the previous knowledge outlined in the mapping review, but also provide novel insights and showcase the condition-specific context of TS diagnostic disclosure. In line with the findings of the mapping review, our original study showed that parents usually need time to process and adapt to the diagnosis, while considering the right time and way to disclose information about the condition to ILWTS. A gradual disclosure from an early age seemed to facilitate positive experiences of disclosure and adapting to living with the condition. However, our participants provided details and examples of how the responsibility of managing information about the condition and disclosure can be gradually passed from parents to their children in a timely and person-centred manner that facilitates normalising the condition. Contrary to many parents' views, ILWTS wished to take ownership of the disclosure process from early on, as this seemed to promote a sense of agency and independence.

Our study also revealed specific needs related with coping with TS-specific symptoms, such as infertility. Of interest, parents' expectations about their children's wish and capacity to form a family did not appear to be consistent with the views of ILWTS. Whereas many parents feared that their children would be devastated to learn about infertility and experienced a sense of guilt for passing on the condition, ILWTS appeared to be open to alternative options, such as IVF or adoption, and were primarily concerned with peer rejection and stigma during adolescence. Fear of stigma was also highlighted by parents as a barrier to disclosure to their children and others. It was associated with the negative portrayal of the condition in informational resources and with negative experiences of disclosure from professionals who primarily focused on symptoms, both of which appeared to promote a sense of disability. Such negative experiences may partially explain why parents did not consult healthcare professionals

to discuss the best time or way to disclose information about the condition to ILWTS, as opposed to findings from studies which explored diagnostic disclosure of other genetic conditions. Our study also showed that those affected by TS valued strengths-based person-centred approaches employed by sensitive, knowledgeable and supportive professionals, who considered individual and family needs on a case-by-case basis and took the time to provide needs-appropriate information. The study findings indicate that healthcare professionals need to actively collaborate with families affected by TS and employ a person-centred family-focused approach, to facilitate empowering experiences of disclosure and living well with the diagnosis.

The last article of this thesis provided insights into the main lessons learnt from conducting research into one's own condition. The author's reflections highlight the need to carefully consider self-disclosure in advance of the study, as appropriate strategies need to be in place to address potential concerns of participants. Consideration of pre-suppositions regarding a 'total insider' position is also warranted, as such assumptions may not always match participants' initial perception of the researcher. Furthermore, future researchers who aim to carry out studies on a condition of which they have lived experience need to actively use reflexivity and PPI input, to ensure rigor and representativeness of findings. Finally, an academic network of pastoral and peer support needs to be proactively put in place to ensure the emotional wellbeing of the researcher.

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Appendices

Appendix A.

Ethics approval letter

10/11/2020
Project Title: Turner Syndrome and Disclosure

EthOS Reference Number: 25038

Ethical Opinion

Dear Emma Clarke,

The above application was reviewed by the Health, Psychology and Social Care Research Ethics and Governance Committee and, on the 10/11/2020, was given a favourable ethical opinion. The approval is in place until 09/01/2022.

Conditions of favourable ethical opinion

Application Documents

Document Type	File Name	Date	Version
Consent Form	Consent form	15/09/2020	1.0
Information Sheet	Debrief Sheet	15/09/2020	1.0
Information Sheet	Withdrawal form	15/09/2020	1.0
Information Sheet	Incident Log	15/09/2020	1.0
Information Sheet	Family Member Interview Schedule	21/09/2020	1.0
Information Sheet	Interview Schedule for Individuals Living with Turner Syndrome	21/09/2020	1.0
Information Sheet	Family Member Socio-demographic Questionnaire	21/09/2020	1.0
Information Sheet	Individual Living with Turner Syndrome Socio-demographic Questionnaire	21/09/2020	1.0
Project Protocol	Thesis Protocol FINAL	26/10/2020	1.1
Recruitment Media	Poster (2)	26/10/2020	1.1
Information Sheet	Participant Information Sheet (1)	26/10/2020	1.1
Information Sheet	Poster (2)	26/10/2020	1.1

The Health, Psychology and Social Care Research Ethics and Governance Committee favourable ethical opinion is granted with the following conditions.

Adherence to Manchester Metropolitan University's Policies and procedures

This ethical approval is conditional on adherence to Manchester Metropolitan University's Policies, Procedures, guidance and Standard Operating procedures. These can be found on the Manchester Metropolitan University Research Ethics and Governance webpages.

Amendments

If you wish to make a change to this approved application, you will be required to submit an amendment. Please visit the Manchester Metropolitan University Research Ethics and Governance webpages or contact your faculty research officer for advice around how to do this.

We wish you every success with your project.

HPSC Research Ethics and Governance Committee

HPSC Research Ethics and Governance Committee

For help with this application, please first contact your Faculty Research Officer. Their details can be found here

Appendix B.

Interview guide for family members

1. Opening Question: Thanks again for agreeing to take part. I'd like us to begin with asking you, how would you describe Turner Syndrome?
2. Could you tell me a bit about the events that led up to the diagnosis of the individual you care for?
    - Could you tell me a bit more about how you were told about the diagnosis?
    - What was your understanding of the condition at the time?
        - Did this change after you received the diagnosis?
        - If yes, how?
    - Were you provided with any resources at the time to help you understand the diagnosis or find support?
        - (If yes,) Could you tell me a bit more about this?
    - Looking back, is there anything you felt was positive about that experience?
        - Why is that?
    - Is there anything that you wish you was different about that experience?
        - Could you please elaborate on that?
3. Did the way you learned about the diagnosis influence how or when you decided to share the diagnosis with the individual you care for?
    - Why was that?
    - Could you tell me a bit more about that?
4. I would now like us to talk about your experience of sharing the diagnosis with the individual you care for. Is that okay?
    - Could you tell me about the events that led up to your decision to share the diagnosis?
    - What was your understanding of the condition at the time?
    - Did you have a plan on how you wanted to share the information?
        - (If yes,) What was that?
        - (If yes,) Why did you develop this plan?

- Could you tell me about how you shared the diagnosis?
    - What information about the diagnosis did you share?
    - Did you use your plan? (If relevant)
    - How did the plan work out? (If relevant)
- Did you use any resources to help you with the process of disclosure?
    - (If not,) Why was that?
    - (If yes,) Could you tell me a bit more about that?

5. I was just wondering, was there anything which may have concerned or prevented you from talking about the diagnosis with the individual you care for?

- Why was that?
- Could you tell me a bit more about that?

6. Are there any other resources or forms of support that you think could have helped you with the process of disclosure?

- Why is that?
- Would you like to elaborate on that?

7. What would be your advice to families who are currently planning to disclose the diagnosis to their child/grandchild?

- Why is that?
- Could you tell me a bit more about that?

Appendix C. Interview guide for ILWTS

1. Opening Question: Thanks again for agreeing to take part. I'd like us to begin with asking you, how would you describe Turner Syndrome?
2. Could you tell me about the events which lead up to you learning about the diagnosis?
3. I'd now like us to talk about the time when you were told about the diagnosis. Is that okay?
4. So, I was just wondering, how did you learn about the diagnosis? Could you tell me a bit more about who told you?
    - Were you provided with any information or resources at the time?
    - What was your understanding of Turner Syndrome at the time?

4. If it's okay, I'd be interested to know, how did you feel when you were told about the diagnosis?

- Why is that?
- How did you react when you learned about the diagnosis?
    - Why do you think that was?
- Did you take any action after learning about the diagnosis?
    - (If yes) What was that?
    - (If yes) Why did you do that?
- Looking back, is there anything which you felt was positive about that experience?
- Is there anything you wish would have been done differently for the disclosure of the diagnosis?

5. I was just wondering, were there any resources or sources of support that you used after learning about the diagnosis?

- (If not,) Why was that?
- (If yes,) Could you tell me a bit more about that?
- Looking back, are there any other resources or forms of support you would have liked to have available?
    - Why is that?
    - Could you tell me a bit more about that?

6. What would be your advice to family members who are currently planning to share the diagnosis with their child or grandchild?

- Why is that?
- Would you like to elaborate on that?

7. What would be your advice to individuals who have just learnt about their diagnosis?

- Why is that?
- Could you tell me a bit more about that?