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Reaction to Diagnosis and Parental Concerns in Parents of Children and Young Adults With XYY Syndrome

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ABSTRACT

Background: There is a growing interest in exploring parents' views on the benefits of early diagnosis and awareness of sex chromosome trisomies. However, only a few studies focus specifically on the experience of parents of children with XYY syndrome. The present study aimed to assess, in parents of individuals with XYY, the perceived severity of their children's condition, their level of satisfaction with the disclosure process and their concerns about their children's present and future condition.

Methods: A national online sample of 56 Italian parents of children and young adults diagnosed with XYY syndrome participated in the study. They filled out a specifically developed online survey that assessed their children's areas of concern, their experience with the disclosure process and their worries about their children's condition.

Results: Seventy per cent of the parents received a prenatal diagnosis, whereas 30% received a postnatal diagnosis. High individual variability was found in the parent report of their child's condition. The most frequent areas of concern were attention regulation, emotion control and behaviour control. Individuals with a postnatal diagnosis showed more severe profiles. Parents were generally dissatisfied with the disclosure process, with no differences between prenatal and postnatal disclosure. However, more than 50% of the parents who received a prenatal disclosure reported that their child's condition was less severe than they had expected. In contrast, only 11% of the parents with postnatal disclosure reported this situation. Parents' concerns were negatively related to global satisfaction with the disclosure process and the correspondence between current and expected conditions but positively associated with the child's severity level.

Conclusions: The results suggest that clear and realistic information during the disclosure process to parents is needed in both prenatal and postnatal communication and may alleviate parents' concerns.

1 | Introduction

XYY syndrome, otherwise known as Jacobs syndrome or double Y syndrome, is a sex chromosome trisomy with an estimated prevalence of 1/1000 male births, although many cases probably go undetected due to the mild phenotype characterising these individuals (Bardsley et al. 2013; Berglund, Stochholm, and Gravholt 2020). The detection of this condition has been rising in the last decades due to the increased use of non-invasive prenatal screening (NIPS), which could result in more individuals being diagnosed prenatally (Abramsky and Chapple 1997; Deng, Cheung, and Liu 2021). The physical features of males with XYY syndrome are usually not very evident, varying widely between individuals. They may include

Summary

- High individual variability characterises XXY syndrome and, consequently, the experience of parents having children with this condition.
- Parental concerns are positively related to the perceived severity of their children's condition but negatively associated with global satisfaction with the disclosure process.
- More effort should be given to improve diagnosis disclosure to parents of children with XYY syndrome.

tall stature, macroorchidism, macrocephaly, hypertelorism, clinodactyly and hypotonia (Bardsley et al. 2013). In contrast, the behavioural and neuropsychological profile of children and adults with XYY syndrome is generally more pronounced and may be challenging for them and their families (Jodarski et al. 2023).

There is a marked interindividual variability in the developmental profile of children with XYY syndrome, with children diagnosed after birth usually showing lower competencies than those diagnosed prenatally (Joseph et al. 2018). This is due to an ascertainment bias, as most children diagnosed postnatally are identified due to clinically detectable symptoms. Individuals with XYY syndrome typically show intelligence quotients falling in the normal range, although they could score lower than peers with normal karyotypes in standardised tests or show some degree of developmental delay (Joseph et al. 2018; Leggett et al. 2010). Executive functions, especially cognitive flexibility, are frequently impaired compared to peers with a 46,XY karyotype or peers with Klinefelter syndrome (XXY) (Ross et al. 2008). Language and specific learning impairments are frequently detected in children with XYY syndrome, but there is a paucity of studies dealing specifically with speech and language development in males with an extra Y chromosome (Bishop et al. 2019). However, studies on the whole population of children with sex chromosome trisomies, including XYY syndrome, Klinefelter syndrome and triple X, showed significant delays and impairments in language from the early stages of communicative development (Bishop et al. 2019; Urbanus et al. 2022; Zampini et al. 2021a, 2022).

Children and adults with XYY are at an increased risk for mental health conditions, not only considering postnatally diagnosed children, who are often referred for evaluation because of these symptoms, but also considering individuals identified before birth (Berglund, Stochholm, and Gravholt 2020; Jodarski et al. 2023). Behavioural issues are more frequently detected in these children and adolescents than in peers with a 46,XY karyotype (Operto et al. 2019). Both externalising problems (such as impulsivity, behavioural dysregulation, attention problems and aggressive behaviours) and internalising issues (such as withdrawal, anxiety and depression) are more prevalent in individuals with XYY than in the general population of males (Operto et al. 2019; Lalatta et al. 2012; Ross et al. 2015). It must be noted that behavioural issues may not be evident in the first years of life, and they could emerge only later (Lalatta et al. 2012; Lorini et al. 2022). Concerning neurodevelopmental disorders, individuals with XYY syndrome are more likely than the general population and other sex chromosome trisomies to receive a diagnosis of attention deficit hyperactivity disorder (ADHD) (Bardsley et al. 2013; Tartaglia et al. 2012), autism spectrum disorder (Bardsley et al. 2013; Joseph et al. 2018; Bishop et al. 2011) and specific learning impairments (Joseph et al. 2018).

As for Klinefelter syndrome and other sex chromosome aneuploidies (Ferlin 2020), the prenatal diagnosis of XYY condition is frequently the consequence of an adventitious finding during prenatal karyotyping performed for other reasons (e.g., higher risk of Down syndrome due to maternal age). Therefore, women undergoing prenatal karyotyping are frequently uninformed, or they did not recall being informed, about the possibility of detecting sex chromosome trisomies before undergoing these tests (Lalatta and Tint 2013; Riggan et al. 2021; Zampini et al. 2021b). The discovery of an XYY condition in the foetus involves a high level of anxiety and stress and poses a dilemma for prospective parents who need to understand the impact of an unexpected genetic syndrome on their and their children's quality of life (Lalatta et al. 2012). How the diagnosis is disclosed in the first place could significantly affect the decision to carry the pregnancy to term, parents' interpretation of the information presented later and even future parent–child interaction (Abramsky and Chapple 1997; Zampini et al. 2021b; Bourke et al. 2014; Zampini et al. 2020).

The need for clear guidelines on how to communicate a diagnosis of sex chromosome trisomy is recognised by studies that show that the first disclosure to parents is often made by people not specifically trained to communicate a diagnosis (e.g., midwives or obstetricians) and that the information is often misleading and not up to date (Lalatta et al. 2012; Zampini et al. 2021b; Abramsky et al. 2001; Riggan et al. 2023). Moreover, a high level of parental dissatisfaction is also related to the lack of sensitivity and empathy on the part of those who communicated the diagnosis; in addition, the delay in scheduling an appointment to explain the condition anticipated by telephone or written medical report could contribute to parental dissatisfaction (Zampini et al. 2021b). The need to communicate a diagnosis of XYY syndrome in person in the first place is particularly relevant, considering the risk that parents may be exposed to biased and outdated information on the Internet (Jodarski et al. 2023; Riggan et al. 2021). Clinicians should be prepared to clarify any possible misinformation arising from the limitations of the early literature on XYY syndrome, which is characterised by important biases (e.g., the study of institutionalised individuals and ascertainment bias) (Jodarski et al. 2023). Special attention to parental needs is essential as parents of children diagnosed with XYY syndrome often show high levels of parenting stress and anxiety (Operto et al. 2019; Lalatta et al. 2012), although parents who received the diagnosis prenatally usually show better coping strategies (Jodarski et al. 2023; Operto et al. 2019).

1.1 | Aims of the Present Study

Although there is a growing interest in exploring parental views on the benefits of early diagnosis and awareness of sex chromosome trisomies (Riggan et al. 2023), only a few studies

specifically focus on the experience of parents of children with XYY syndrome (Operto et al. 2019; Lalatta et al. 2012).

The present study aimed to analyse parental views on their children with XYY syndrome. In particular, we were interested in examining three main topics: (1) parents' perceived severity of their child's condition, (2) parents' satisfaction level with the disclosure process and (3) parental concerns about the present and future condition of their son. We were then interested in comparing the experience of parents who received prenatal and postnatal diagnoses concerning these three main topics. We hypothesised that children and young adults diagnosed before birth could show milder profiles because those identified postnatally are frequently referred for evaluation due to clear symptoms (Joseph et al. 2018). However, we did not know whether receiving a prenatal or postnatal diagnosis would affect parental satisfaction with the disclosure process, as no previous studies have focused specifically on this topic in this specific population. Lastly, we were interested in analysing the possible relationships among parental satisfaction with diagnosis disclosure, the severity of the XYY condition and parental concerns. We hypothesised that the perceived severity of symptoms could be positively related to parental concerns and that receiving a realistic description of the XYY condition could positively impact satisfaction levels and reduce parental concerns.

2 | Methods

2.1 | Participants

Participants were the parents of 56 children and young adults diagnosed with XYY syndrome. The questionnaire, specifically developed for the present study, has been filled in by 43 mothers (77%) and 13 fathers (23%). Parents were recruited from national associations for people with sex chromosome trisomies. The participants signed informed consent before being included in the study. Participation was voluntary, and parents received no compensation for their participation. The study was approved by the local commission for minimal-risk studies of the Department of Psychology of the University of Milano-Bicocca (Milan, Italy). We only considered the fully completed questionnaires (80% of the accesses to the online survey), and we excluded incomplete questionnaires. The mean respondents' age was 43 years (SD = 7.56; range = 25–56) at the time of the investigation. All the respondents were Italian. Concerning parental education, four parents (7%) attended primary and middle school (8 years of education), three (5%) had a certificate of professional competence (11 years of education), 19 (34%) had a high school diploma (13 years of education), 23 (41%) had a university degree (16–18 years of education), and seven (13%) had a postgraduate specialisation (21–23 years of education). Seventy-nine per cent of the parents ($n = 44$) were working at the time of the study. Eighty-two per cent ($n = 46$) were living with the other parent of their child with XYY syndrome. The respondents' mean number of children was 1.66 (SD = 0.72; range = 1–3).

2.2 | Procedure

Information on the communication of the children's diagnosis to their parents was collected by means of an anonymous questionnaire developed specifically for the present study. The questionnaire was created using Qualtrics (https://www.qualtrics.com) to be easily filled out by parents living in different Italian areas. The areas investigated in the survey and the questions are based on previous studies on this topic and clinical experience with parents of individuals with sex chromosome aneuploidies. All the questions were yes-no, multiple-choice and rating questions (except for those asking for numbers, e.g., 'How old are you?' and 'How many children do you have?'). The English translation of the survey items can be found in Appendix 1. The survey took approximately 15 min to complete. The questionnaire focussed on three main areas:

1. The severity of symptoms shown by children and young adults diagnosed with XYY syndrome, as perceived by their parents.
2. Parents' satisfaction level for the communication process in which their children's diagnosis was revealed.
3. Parental concerns about the present and future condition of their children.

2.2.1 | Perceived Severity Level

To assess the severity of the symptoms exhibited by children and young adults with XYY syndrome, the parents were asked to indicate if their children showed some problems in 10 areas of development: (1) personal autonomies, (2) motor skills, (3) language, (4) reasoning, (5) attention, (6) behaviour, (7) emotion control, (8) relationships with non-familiar adults, (9) relationships with peers and (10) learning. For each of these areas, the parents were asked to indicate if their children showed any problem and, in the case of an affirmative answer, to rate on a 3-point Likert scale if those problems were (a) *mild* (1 point), (b) *medium* (2 points) or (c) *severe* (3 points). Cronbach's alpha coefficient was 0.85, which indicates that the 10 items have good internal consistency. Then, a severity score (ranging from 0 to 30) was computed by adding the scores totalised in each area (e.g., a child with only mild difficulty in language development and severe difficulty in relationships with peers would score 4 in the severity scale).

2.2.2 | Parents' Satisfaction With the Communication Process

Respondents were asked who disclosed to them their children's diagnosis and when. The parents were then asked to rate their level of satisfaction with the communication process on a scale ranging from 0 (*not at all*) to 10 (*completely*). Five dimensions were measured:

1. Clarity of information ('How clear do you think the information provided was?');

2. Completeness of information ('How exhaustive do you think the information provided was?');
3. Willingness to answer questions ('How willing do you think the person who disclosed the diagnosis was to answer your questions?');
4. Empathy ('How empathetic do you think the person who communicated the diagnosis was?')
5. Global satisfaction ('Overall, how satisfied are you with how your child's diagnosis was communicated to you?').

Cronbach's alpha coefficient for the five items was 0.97, suggesting that the items have excellent internal consistency. Parents were then asked to rate on a scale ranging from 0 (*not at all*) to 10 (*completely*) if the current condition of their children reflected the description given during the diagnosis disclosure. Moreover, they were asked whether, compared to what was prognosticated during the communication of the diagnosis, their child's current condition was (1) much less severe than expected, (2) a little less severe than expected, (3) about as expected, (4) a little more severe than expected or (5) much more severe than expected.

2.2.3 | Parental Concerns

To assess parental concerns about their children's present and future condition, six questions were addressed to the parents: 'Currently, how concerned are you about the following?' (1) that my child now, or in the future, may perceive himself as 'different from others'; (2) that my child now, or in the future, may be considered by others as 'different'; (3) that the diagnosis may influence my child's life choices; (4) that my child may develop problems in the future that are not evident now; (5) that the difficulties my child is experiencing now may increase in the future; (6) that my worries could affect my relationship with my child. For each statement, the parent had to rate, on a 5-point Likert scale, how much they were concerned by the situations described (a) *not at all concerned* (0 points), (b) *not very concerned* (1 point), (c) *quite concerned* (2 points), (d) *very concerned* (3 points) or (e) *totally concerned* (4 points). Cronbach's alpha was 0.91, which indicates that the six items have excellent internal consistency. Then, a concern score (ranging from 0 to 24) was computed by adding the scores totalised in each statement.

2.3 | Data Analysis

Statistical analyses were conducted using IBM SPSS Version 28. Due to the small number of participants, we used non-parametric statistics. First, we computed descriptive statistics for parents' perceived severity of their child's condition, parent satisfaction with the disclosure process and parental concerns about their child's present and future condition. Then, we calculated the correlations (Spearman's rho) of these variables with the age of individuals with XYY and time since diagnosis.

Second, to compare the experience of parents who received prenatal and postnatal diagnoses, we used the Mann–Whitney test to identify possible between-group differences in perceived severity, parent satisfaction and parental concerns. An η^2 of 0.02, 0.13 and 0.26 indicate small, medium and large effect sizes. Lastly, we used Spearman's rho correlation to analyse the relationships among these three factors.

3 | Results

3.1 | Description of Children and Young Adults With XYY Syndrome

The mean age of children and young adults with XYY syndrome was 8 years (SD = 6.08; range = 1 month to 25 years) at the time of the investigation. Five children (9%) were attending nursery school, eight children (14%) kindergarten, 15 children (27%) primary school, nine children (16%) middle school, and four children (7%) high school. Twenty-seven per cent of children and young adults ($n = 15$) were not attending any school or educational service at the time of the study. All children and young adults lived with the parent who filled in the questionnaire, except for two (4%). At the time of the study, 22 children and young adults (39%) were aware of their diagnosis: 11 years (SD = 4.26; range = 5–20) was the mean age at which the diagnosis was communicated to them.

Concerning the perceived severity of the symptoms shown by children and young adults with XYY syndrome, the mean severity score was 10 (SD = 7.33; range = 0–24). The proportion of problems shown at the time of the study by children and young adults in each developmental area is reported in Figure 1. The most frequent areas of concern were attention regulation and emotion and behaviour control; in these areas, about 20% of children and young adults were reported to show severe problems. The perceived severity score appeared to be significantly related to children's age (rho = 0.55; $p < 0.001$) and time since diagnosis (rho = 0.39; $p = 0.003$).

3.2 | Parents' Level of Satisfaction With the Communication Process

The data collected showed that communication of the diagnosis of XYY syndrome was given to the parents by a geneticist in 43 cases (77%), a child neuropsychiatrist in four cases (7%), a gynaecologist in two cases (4%) and other (i.e., an endocrinologist) in one case (2%). Six parents (11%) did not remember the professional role of the person who communicated their child's diagnosis.

Seventy per cent of the parents ($n = 39$) received a prenatal diagnosis, whereas 30% ($n = 17$) received a postnatal diagnosis. For the postnatal condition, the mean age of children at diagnosis disclosure was 6 years (SD = 3.57; range = 4 months to 13 years).

Parents' satisfaction with the communication process was around 5 points (on a scale ranging from 0 to 10) in each investigated dimension. The medium score was 5.41 (SD = 3.21, range = 0–10) for clarity of information, 5.25 (SD = 3.16, range = 0–10) for completeness of information, 6.43 (SD = 2.95, range = 0–10) for

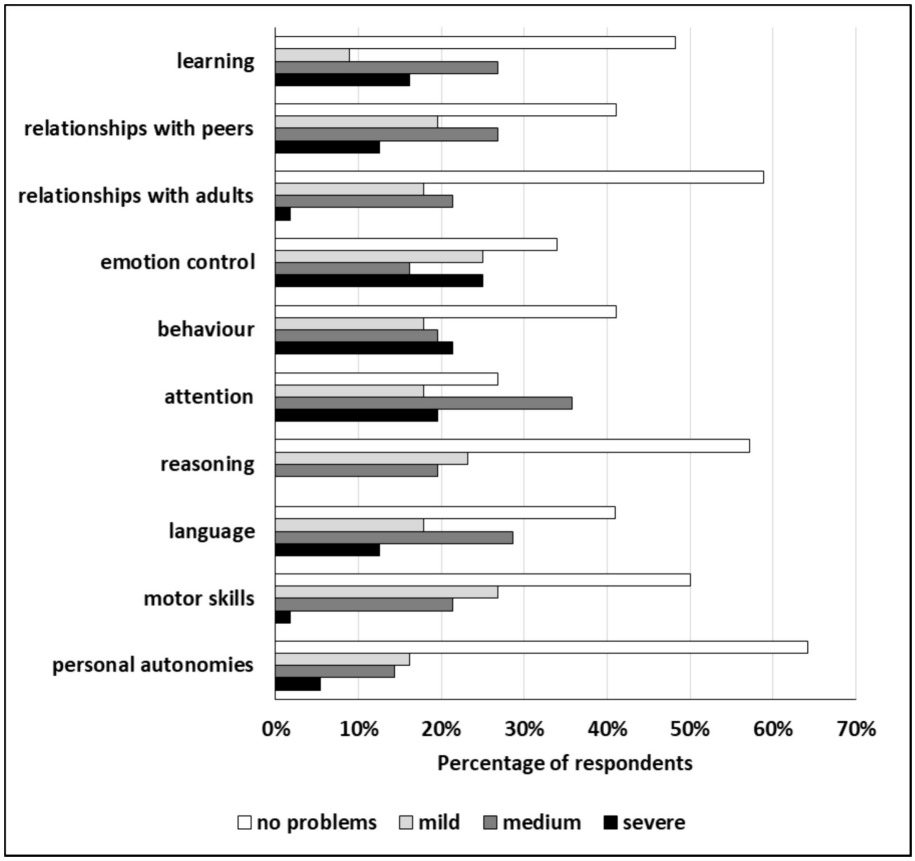

FIGURE 1 | Problems showed by children and young adults in each developmental area, as perceived by their parents.

willingness to answer questions, 5.77 (SD = 3.38, range = 0–10) for empathy and 4.93 (SD = 3.51, range = 0–10) for global satisfaction.

Parents reported that the current condition of their children was not well prognosticated by what was communicated to them during diagnosis disclosure (M = 4.38, SD = 2.95, range = 0–10). Thirty-nine per cent of respondents reported that the current condition of their child was more serious (16% much serious and 23% a little serious) than they expected. In contrast, 39% of respondents reported that the current condition was less serious (23% much less serious and 16% a little less serious). Only 21% of the parents reported that their children's current condition was what they had expected based on what they had been told at the time the diagnosis was communicated. Parent satisfaction level was not significantly related to children's age (rho = −0.09; $p > 0.05$) or time since disclosure (rho = −0.14; $p > 0.05$).

3.3 | Parental Concerns

The respondents showed a mean concern score of 12.66 (SD = 6.21; range = 0–24). The wide range of scores (the maximum possible range for this scale) indicates that for the parents of some children and young adults, the diagnosis of XYY syndrome was not a source of concern, whereas for others, it was a very stressful condition. The concern that scored highest was 'that my child may develop problems in the future that are not evident now' (M = 2.48; SD = 1.19; range = 0–4), whereas the one that scored lowest was 'that my worries could affect my relationship with my child' (M = 1.47; SD = 1.23; range = 0–4). Parental concerns were not significantly related to children's age (rho = 0.03; $p > 0.05$) or time since disclosure (rho = −0.01; $p > 0.05$).

3.4 | Comparison Between Prenatal and Postnatal Diagnosis Disclosure

The mean severity score was 7.79 (SD = 6.69; range = 0–22) for individuals with a prenatal diagnosis ($n = 39$) and 15.06 (SD = 6.27; range = 3–24) for those with a postnatal diagnosis ($n = 17$). Children and young adults with XYY syndrome who received the diagnosis before birth showed significantly less severe symptoms ($U = 134.5$; $p < 0.001$; $\eta^2 = 0.23$) than those diagnosed postnatally. However, we must also consider that children's age was significantly different between groups ($U = 496$; $p = 0.003$; $\eta^2 = 0.16$), with a mean age of 6.52 years (SD = 6.11; range = 0–25) in the prenatal group and 10.88 years (SD = 4.92; range = 3–23) in the postnatal group.

Receiving a diagnosis disclosure before or after childbirth could significantly affect the parental psychological experience. Therefore, we compared parental satisfaction with the communication process between parents who received the disclosure of XYY syndrome in the prenatal period and those who received it postnatally. As reported in Table 1,

TABLE 1 | Parental satisfaction with prenatal and postnatal diagnosis disclosure.

	Prenatal disclosure			Postnatal disclosure					
	M	SD	Range	M	SD	Range	*U*	*p*	η^2
Clarity of information	5.44	3.42	0–10	5.35	2.74	0–10	309	0.686	< 0.001
Completeness of information	5.28	3.36	0–10	5.18	2.72	0–10	314.5	0.760	< 0.001
Willingness to answer questions	6.33	3.15	0–10	6.65	2.50	1–10	324	0.892	< 0.001
Empathy	5.85	3.54	0–10	5.59	3.08	0–10	304.5	0.628	< 0.001
Global satisfaction	4.64	3.67	0–10	5.59	3.10	0–10	285.5	0.407	0.01
Correspondence with the current situation	4.00	3.04	0–10	5.18	2.65	0–10	233.5	0.164	0.04

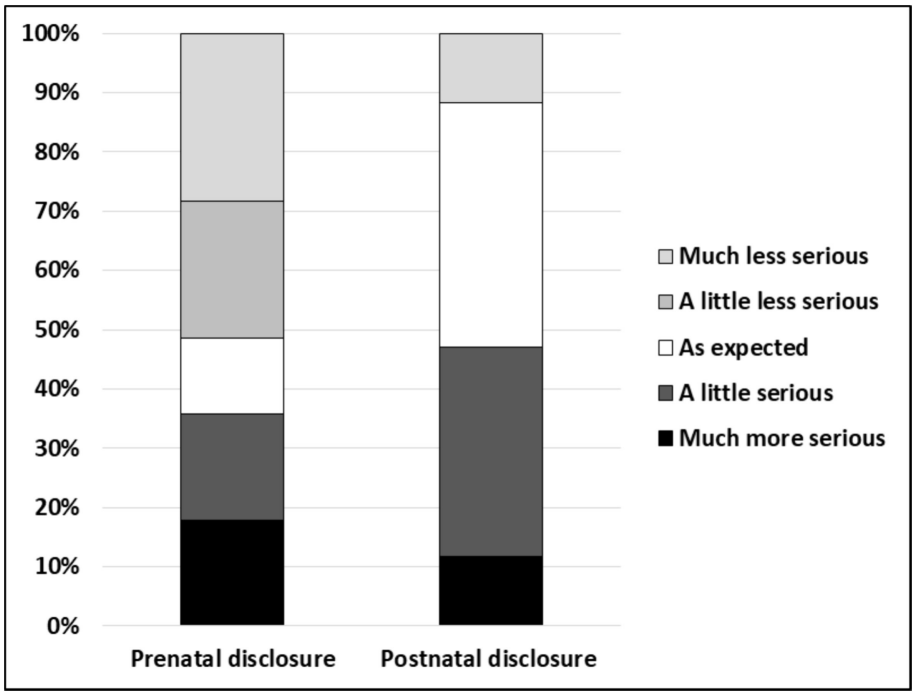

FIGURE 2 | How is the current situation of the child/young adult compared to what was prognosticated during the diagnosis disclosure?

there were no significant differences in the level of parental satisfaction between the groups. In addition, no statistically significant differences emerged in how well the current condition of their child was prognosticated during the diagnosis communication.

Considering the severity of their children's current condition compared to what was prognosticated, as reported in Figure 2, only 13% of the parents who received a prenatal diagnosis reported that the current situation was as they expected. In addition, more than 50% of these parents reported that their children showed less severe symptoms than they expected, whereas 36% reported more serious problems than they expected. Concerning parents who received a postnatal disclosure, 41% reported that the description during the disclosure well represented the current condition of their children. However, 47% of these parents reported that the current problems shown by their children were more severe than they expected based on the communication. This distribution significantly differed between prenatal and postnatal groups (likelihood ratio = 13.81; $p = 0.008$).

No significant differences emerged in parental concerns between parents who received prenatal or postnatal disclosure (prenatal: M = 12.79; SD = 6.73; range = 0–24. Postnatal: M = 12.35; SD = 4.97; range = 6–21. $U = 309.5$; $p = 0.694$; $\eta^2 < 0.001$).

3.5 | Relationships Between Satisfaction With Diagnosis Disclosure, the Severity of the Condition and Parental Concerns

As reported in Table 2, a correlational matrix was calculated to investigate possible relationships among the level of satisfaction with the disclosure process, the perceived severity of children's and young adults' condition, the correspondence between real and expected conditions and parents' concerns. Global satisfaction appeared to be significantly related

TABLE 2 | Relationships (Spearman's rho) among satisfaction with the disclosure process, the perceived severity of the condition, correspondence between the present and predicted condition and parental concerns.

	Severity score	Correspondence with the current situation	Concern score
Global satisfaction	−0.16	0.46**	−0.34*
Severity score		0.08	0.40**
Correspondence with the current situation			−0.34*

*$p < 0.05$, **$p < 0.01$.

(rho = 0.46; $p < 0.01$) to the correspondence between the current condition and what the parents expected based on what they had been told at the time of diagnosis: The parents who received a realistic description of their child's condition were more satisfied. In contrast, no relationships emerged between global satisfaction and the severity of the child/young adult's condition. Parental concerns were negatively correlated to both global satisfaction with the disclosure process (rho = −0.34; $p < 0.05$) and the correspondence between present and expected conditions (rho = −0.34; $p < 0.05$), whereas the concern score was significantly and positively related to the child/young adult severity score (rho = 0.40; $p < 0.01$).

4 | Discussion

The present study aimed to analyse parental views on their children with XYY syndrome, examining three main topics: (1) how severe their child's condition appeared to the parents, (2) parents' satisfaction level with the disclosure process and (3) parental concerns about the present and future condition of their child.

High individual variability was found in the severity of the XYY condition shown by children and young adults. As reported by parents, the severity score ranged between 0 and 24 (within a possible range of 0–30). The most frequent areas of concern were emotion control, behaviour control and attention regulation: About 20% of the individuals were reported as showing severe problems with these skills. These data confirmed previous studies highlighting that the emotional-behavioural area is the most symptomatic one in the population of individuals with XYY, with a greater risk of externalising symptoms (e.g., aggressive, oppositional or defiant behaviours) (Operto et al. 2019). Significant impairments were also found in learning, language and relationships with peers, with more than 10% of the parents reporting severe problems in their children. These impairments have also previously been identified in the literature on individuals with XYY (Joseph et al. 2018) and on the whole population of children with sex chromosome trisomies (Urbanus et al. 2022; Urbanus, van Rijn, and Swaab 2020). Personal autonomies, relationships with adults and reasoning skills were better preserved, as reported by the parents.

Among our participants, 70% received a prenatal diagnosis. As hypothesised, children and young adults diagnosed before birth showed milder profiles than those identified later as the latter usually arrive at the diagnosis due to evident behavioural or psychological symptoms (Joseph et al. 2018). This referral bias is a common problem in studies on individuals with sex chromosome trisomies because these genetic conditions often go undetected if not identified prenatally (Printzlau, Wolstencroft, and Skuse 2017) or after genetic testing to investigate developmental delays or impairments (Bishop et al. 2019).

Concerning parental satisfaction with the disclosure process, our study suggests the need for clear guidelines on how to communicate a diagnosis of XYY. The average scores given by parents were below sufficient in all the dimensions considered (clarity of information, completeness of the information received, empathy and global satisfaction) except for the willingness to answer questions, which reached the level of sufficiency. These results are in line with previous data from the literature (Close, Sadler, and Grey 2016; Jaramillo et al. 2019; Riggan, Close, and Allyse 2020). As reported by Jaramillo et al. (2019), many parents felt that the portrayal of sex chromosome aneuploidy was negative, and the diagnosis was delivered as an unfortunate event. Moreover, a predominantly negative experience at the time of diagnosis was reported by parents of more than 300 children with sex chromosome aneuploidies in a study by Riggan, Close, and Allyse (2020). However, it must be emphasised that most of the parents in that study received the diagnosis from a non-genetic medical provider and this may have influenced the quality of the communication received. In contrast, almost 80% of the participants in the present study received the disclosure from a geneticist.

The need to provide complete and accurate information to parents of children with sex chromosome aneuploidies was highlighted by Close, Sadler, and Grey (2016) through qualitative interviews with mothers and fathers of children with Klinefelter syndrome; these parents reported feeling uninformed and lacking the necessary support to make health and educational care decisions for their children. Considering the Italian context, the satisfaction level of parents of individuals with XYY seems lower than that of parents of individuals with Klinefelter syndrome; as found by Zampini et al. (2021b), around 50% of the parents of children with Klinefelter syndrome (ranging in age from 0 to 13 years) were satisfied or very satisfied with the disclosure process. However, we must note that all the 48 parents considered in Zampini et al.'s study received a prenatal disclosure.

In the present study, no significant differences emerged in the satisfaction level of parents who were told before and after the

child's birth. However, we must consider that the option of terminating the pregnancy may have been presented to parents who received disclosure before childbirth. As our participants were parents of individuals born with XYY syndrome, we do not know whether parents with a lower level of satisfaction would have chosen voluntary termination of pregnancy. The participants' recruitment methods often preclude women who terminated their pregnancy after non-invasive screening or invasive prenatal genetic testing; it is very likely that these parents have a different opinion on the quality of prenatal disclosure than parents who continued their pregnancy (Riggan, Close, and Allyse 2020).

Riggan et al. (2021) found that many parents expressed frustration with the limited quantity and quality of information provided during prenatal counselling for sex chromosome aneuploidies. However, the same authors (Riggan, Close, and Allyse 2020) found no significant differences in the feelings (i.e., depressed mood, anxiety and positivity) of parents receiving the diagnosis disclosure before or after childbirth. The only significant difference was found in relief, as parents who received a prenatal diagnosis did not feel relief when the diagnosis was delivered.

Contrary to what was expected, there were no differences in the accuracy of the description of the child's condition during diagnosis disclosure (i.e., how well the description of the condition given to the parents corresponds to the current child's condition) between parents who received prenatal and postnatal disclosure. For both groups, the description of possible outcomes was quite unrealistic: The experience of parents who were told before childbirth was lower but not significantly different from that of parents who were told after birth. Only 13% of the parents who received a prenatal diagnosis reported that the current situation of their child was as prognosticated, whereas 41% of the parents who received a postnatal disclosure stated that the description received during the disclosure well represented the current condition of their child. It must be noted that nearly 50% of the parents in the prenatal diagnosis group found that their children's condition was less severe than prognosticated. This result supports the importance of giving the parents not only information about the possible negative aspects that are frequently associated with sex chromosome aneuploidies but also information about the possible strengths associated with these genetic profiles (Thompson et al. 2022). In contrast, almost 50% of the parents in the postnatal group found the condition more severe than predicted. This result should be interpreted cautiously because children in the postnatal group are significantly older and more severe symptoms were reported in this group. It is possible that behavioural symptoms gradually emerge with increasing age in individuals with XYY. For instance, as found in a study (Lorini et al. 2022) on young children with sex chromosome trisomies, emotional and behavioural problems might not be evident in the first 2 years of life; however, these issues could gradually emerge with child development (Urbanus et al. 2020), and both internalising and externalising symptoms are more prevalent in preadolescents and adolescents with XYY syndrome than in peers with 46,XY karyotype (Operto et al. 2019). The possible gradual onset of emotional or behavioural issues is a topic to discuss with parents when disclosing an XYY condition. This may help parents not to be unprepared for any emerging symptoms. It should also be noted that parents often tend to attribute typical pre-adolescent/adolescent unwanted behaviour to the genetic diagnosis. Therefore, it is important to help parents distinguish between clinical and non-clinical symptoms.

Parental concerns were not significantly different between parents who received prenatal or postnatal disclosure. This result was unexpected, as mothers of children with XYY syndrome diagnosed postnatally due to unexpected developmental delay or learning impairments have reported higher levels of stress in previous studies (Operto et al. 2019).

Children's age and time since disclosure were not related to parent satisfaction with the diagnosis communication and parental concerns. We must note that parent satisfaction was related to how the prediction was realistic but not to the severity of the child/young adult's condition. In contrast, parental concerns were positively associated with the child/young adult severity score but negatively correlated to global satisfaction with the disclosure process and a realistic description of their child's condition. Therefore, because parents who have received a more realistic prognosis are less concerned, we emphasise the importance of providing clear and complete information to parents by adopting multidisciplinary counselling by geneticists, paediatricians and psychologists, as suggested by Lalatta et al. (2012).

4.1 | Conclusions

High individual variability has been reported in individuals with XYY syndrome, with the emotional-behavioural area emerging as a frequent concern among parents. Parents reported a wide range of experiences regarding satisfaction with the disclosure process. On average, they were dissatisfied with the way their child's XYY syndrome was communicated to them, although they reported a sufficient level of willingness to answer their questions. No significant differences emerged in satisfaction with diagnosis disclosure and parental concerns between parents who received a prenatal or postnatal diagnosis. However, we must consider that our participants who received a prenatal disclosure were those who had decided to go ahead with the pregnancy and gave birth to a child with XYY syndrome. Clear and realistic information during the disclosure process could mitigate parental concerns and should be given to parents in both prenatal and postnatal communication. This information should include the weaknesses and strengths frequently associated with the XYY karyotype and a careful explanation of the fact that having an XXY diagnosis could be a risk factor for certain developmental disorders but should not be interpreted deterministically as a cause-and-effect relationship.

4.2 | Limitations and Future Directions

The results must be interpreted with caution, as this survey has not yet been validated. As we have used unpublished measures specifically designed for this study, our results cannot be compared with those of other populations. Furthermore, as can happen in any survey, it is possible that some participants

misunderstood the meaning of one or more questions that might appear ambiguous to the reader.

A limitation of the study is the wide age range of the children/young adults with XYY considered and the under-representation of the fathers' point of view. Another critical point is the absence of direct assessment of the individuals with XYY; in fact, we evaluated how parents saw their children, but we did not know whether their description corresponded to their actual condition. Future studies will aim to compare parental perceived severity with the condition directly assessed by clinicians. This will allow us to assess whether parental satisfaction with the disclosure process and parental concerns correlate with the actual degree of severity of the behavioural or neuropsychological conditions of these individuals.

Another study limitation is that we did not assess the mental health or well-being of parents. Stress, anxiety or depression could significantly affect not only parents' concerns but also the perceived level of severity and parents' satisfaction. Future studies will consider the effect of psychological state and trait characteristics on parents' perceptions and experiences.

Author Contributions

Laura Zampini: conceptualisation, methodology, formal analysis, writing – original draft. **Paola Zanchi:** conceptualisation, methodology, writing – review and editing. **Gaia Silibello:** conceptualisation, data collection, writing – review and editing. **Domenica Mastromattei:** conceptualisation, data collection, writing – review and editing. **Paola Francesca Ajmone:** conceptualisation, data collection, writing – review and editing. **Francesca Dall'Ara:** conceptualisation, data collection, writing – review and editing. **Federico Monti:** conceptualisation, data collection, writing – review and editing. **Maria Antonella Costantino:** project administration, writing – review and editing. **Paola Giovanna Vizziello:** project administration, conceptualisation, data collection, writing – review and editing.

Ethics Statement

This study was approved by the local commission for minimal risk studies of the Psychology Department of the University of Milano-Bicocca (RM-2020-307). Participants signed a written informed consent form before inclusion in the project. No incentives were provided to the participants.

Conflicts of Interest

The authors declare no conflicts of interest.

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Data Availability Statement

The data that support the findings of this study are available from the corresponding author upon reasonable request.

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Appendix A: **Survey Items (English Translation)**

1	Who is answering the survey? (mother/father)				
2	How old are you?				
3	What is your nationality? (Italian/other)				
4	What is your educational qualification? (primary school/middle school/certificate of professional competence/high school diploma/degree/post-graduate specialisation)				
5	Are you currently working? (yes/no)				
6	How many children do you have?				
7	Are you currently living with the other parent of your son with XYY? (yes/no)				
8	How old is your child with XYY?				
9	Does your child currently attend a school or other educational service? (no/nursery school/kindergarten/primary school/middle school/professional school/high school/university)				
10	Is your child currently living with you? (yes/no)				
11	When your child's diagnosis was disclosed to you? (prenatally/after birth)				
12	Who did disclose your child's diagnosis to you? (geneticist/gynaecologist/child neuropsychiatrist/I do not remember/other)				
13	(if after birth) How old was your child when his diagnosis was disclosed to you?				
14	Is your child aware of his diagnosis? (yes/no)				
15	(if aware) How old was your child when he found out about his diagnosis?				
16	Is your child currently showing problems in the following areas?				
		No	Mild	Medium	Severe
	Personal autonomies (e.g., dressing himself)				
	Motor skills				
	Language				
	Reasoning				
	Attention (e.g., poor concentration)				
	Behaviour (e.g., aggressive behaviour and hyperactivity)				
	Emotion control (e.g., uncontrollable crying or laughing, anger)				
	Relationships with non-familiar adults				
	Relationships with peers (schoolmates and friends)				
	Learning (e.g., reading and writing)				
17	On a scale ranging from 0 (*not at all*) to 10 (*completely*)				
	How clear do you think the information provided was?				
	How exhaustive do you think the information provided was?				
	How willing do you think the person who disclosed the diagnosis was to answer your questions?				
	How empathetic (e.g., respectful of your emotions) do you think the person who communicated the diagnosis was?				
	Overall, how satisfied are you with how your child's diagnosis was communicated to you?				
18	On a scale ranging from 0 (*not at all*) to 10 (*completely*), how much do you think the information you were given when the diagnosis was communicated reflects your child's current condition?				
19	Compared to what was prognosticated during the communication of the diagnosis, the current condition of your child is (1) much less severe than expected, (2) a little less severe than expected, (3) about as expected, (4) a little more severe than expected or (5) much more severe than expected				

20	Currently, how concerned are you about the following?				
	Not at all concerned	Not very concerned	Quite concerned	Very concerned	Totally concerned
That my child now, or in the future, may perceive himself as 'different from others'					
That my child now, or in the future, may be considered by others as 'different'					
That the diagnosis may influence my child's life choices					
That my child may develop problems in the future that are not evident now					
That the difficulties my child is experiencing now may increase in the future					
That my worries could affect my relationship with my child					