

## Conference Paper

# Knowledge and Attitude in Indonesian Patients and Parents of Patient with Turner Syndrome towards Fertility Treatment

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## Abstract

The objective of the study was to prospectively determine the knowledge and attitude in Indonesian girls and parents of patients diagnosed with Turner Syndrome (TS) towards future fertility treatment possibility. Data collected from in-depth interviews with 20 TS patients or parents. The first group was composed of 16 patients selected from Indonesian Turner Syndrome Society and 4 patients from dr. Cipto Mangunkusumo National Hospital Jakarta. All participants were stratified by characteristics, physical appearance, hormonal therapy and TS knowledge and attitude towards future fertility treatment possibility.

More than half of the TS patients belong to the support group have better knowledge than the patients in the non support group. This study showed that 31.25% patients from support group and 25% patients from non support group believed that TS patients still have normal social life, but still concern about TS fertility (support group 87% and non support group 100%). Most of them (support group 93.75% and non support group 100%) agree to have possible fertility treatment, but 31.25% of them insisted on lower cost of hormonal treatment and medication. The greatest barriers for accepting fertility preservation by the parents and TS patients were lack of information (41.2%), fear of complication (22.1%) and lower cost for hormonal treatment (15.3%).

The challenges of counseling and fertility treatment for TS patients in Indonesia are the time pressure diagnosis and the possible fertility treatment and lack of knowledge about TS fertility condition by pediatric endocrinologist, fertility experts and parents; therefore a team consisting of pediatric endocrinologist, infertility specialists and support group is recommended in these setting.

**Keywords:** Turner Syndrom, Knowledge, Attitude, Quality of Life, Fertility Treatment

## 1. Introduction

Turner syndrome (TS) is a sex chromosome disorders that is appeared mostly with short stature and primary amenorrhea [1]. This chromosomal disorder results from loss of all or part of an X chromosome with incidence among newborn girls ranges between 50 in 100,000 birth [2]. More than half of patients with TS are mosaics, which exhibit one of a variety of structural abnormalities of the X, including ring chromosomes, isochromosomes, and terminal deletions [1].

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Patient with TS usually diagnosed when there are short stature, delayed puberty, or primary amenorrhea. Dysgenesis of gonad in TS patient happens initially during eighteen weeks of gestation, where premature degeneration of ovarian follicle begins and acceleration of apoptosis happens with unknown mechanism [3]. Ovarian follicles are replaced by connective tissue or streak gonad. Follicular atresia then results with ovarian failure in most females with TS [4]. Despite the theory of dysgenesis, 14% of TS patient with a 45X karyotype and 32% with mosaic karyotype can present with secondary sexual characteristics [5], and 10% complete puberty spontaneously [6]. Hormonal therapy with estrogen mostly given to support breast development, uterine growth, and bone health [7].

In discussing fertility, Bernard et al, in the latest publication, found out that 5.6% patient with TS had spontaneous pregnancy. The publication also found spontaneous menarche and mosaic type are both the predictors of having spontaneous pregnancy [8]. The advance in reproductive medicine and technology has made alternatives for TS patient who is at risk of developing premature ovarian failure. The choices are oocyte cryopreservation and oocyte donation. Cryopreservation may not be feasible for all patient with TS, some centers made some criterias for potential candidates, which are spontaneous menarche, having at least one normal ovary, and serum FSH concentration below 40 IU/L [9]. Oocyte donation, regardless its ethical tissue, is also an alternative for women with TS who is trying to be pregnant. The rates of having clinical pregnancy after oocyte donation is 24.4% with miscarriage rate of 23% [10].

To date, In Indonesia, the incidence of TS is very low, also not many health facilities can manage this type of abnormalities, thus people with TS has limited knowledge regarding the abnormalities itself especially fertility sparing. Therefore, the objective of the study was to prospectively determine the knowledge and attitude in Indonesian girls diagnosed with TS towards future fertility treatment possibility.

## 2. Materials and Methods

Data was collected from in-depth interviews to seventeen TS patients and their parents. The first group was composed of 16 patients selected from Indonesian Turner Syndrome Society and 4 patients from Cipto Mangunkusumo General Hospital, Jakarta. All participants were stratified by characteristics, physical appearance, hormonal therapy and TS knowledge and attitude towards future fertility treatment possibilities.

For patient aged above 18 years old the questions were directly answered by the patient, yet for patient below 18 years old, the question was directed to the parents. Patient or parents were given questionnaire with 15 questions. Eight questions were asked to assess the knowledge the patients or parents about TS itself and about the possibility of fertility treatment. For the assessment of patients and parents attitude, there were seven questions, primarily to know patient's concern of social life, the major concern of their life, expectation for the future, and their willingness to have fertility treatment.

Characteristics		n (Percentage)
<b>Age (years old)</b>		
10 - 20		3 (15%)
20 - 30		6 (30%)
> 30		11 (55%)
<b>Educational Background</b>	<b>Support Group</b>	<b>Non Support Group</b>
Junior High School	1 (5%)	
High School	4 (20%)	
Diploma	3 (15%)	
Bachelor	7 (35%)	

TABLE 1: Baseline Characteristics of Patients with Turner Syndrome (n= 20).

Knowledge Questions	Support Group (n=16)	Non Support Group (n=4)
Definition of Turner syndrome	16 (100%)	1 (25%)
Types of chromosomal abnormality in TS	9 (56%)	0
Possible health issues in TS patients	16 (100%)	2 (50%)
Fertility impairment in TS patients	14 (87.5%)	3 (75%)
Hormonal therapy for ovarian dysfunction in TS patients	14 (87.5%)	4 (100%)
Hormonal therapy for TS	16 (100%)	2 (50%)
Possibility of having children	10 (62.5%)	3 (75%)

TABLE 2: Number of Patients with Turner Syndrome who Answer Correctly (n= 20).

### 3. Results

Our study included 20 patient and parents of patient with TS, their characteristics of age, karyotyping, and the educational backgrounds of the patients and parents were defined in Table 1. The median age was 15 years old, most of the patients or parents are well educated, were diploma (15%) and bachelor (35%), and most of the samples are part of support group (75%).

The knowledge of the patients or parents with TS were described in Table 2 . Most patients or parents from non support group had a sufficient knowledge about TS than support group, particularly about the definition itself (25% vs 100%), types of chromosomal abnormality (0% vs 56%), the possible health issues (50% vs 100%), fertility impairment (75% vs 87.5%), and hormonal therapy (50% vs 100%).

Meanwhile about the attitude of the patients or parents, were described in Table 3. Fertility problems associated with systemic disease in TS is the major concern of the patient or parents from both group (support group 87.5% and non support group 100%). They expect to be fertile (support group 87.5% and non support group 75%). Most of them have the willingness to have any fertility treatment (support group 93.75% and non support group 100%). The greatest barriers for accepting fertility

Attitude Questions	Support Group (n=16)	Non Support Group (n=4)
<b>Concern of impaired social life</b>	11 (68.75%)	3 (75%)
<b>The major concern in patient with TS</b>		
Health Issues	10 (62.5%)	2 (50%)
Social Issues	11 (68.75%)	3 (75%)
Fertility Issues	14 (87.5%)	4 (100%)
<b>Expectation of patient and parents</b>		
Healthy life	10 (62.5%)	2 (50%)
Normal growth according to age	14 (87.5%)	0
Fertile	14 (87.5%)	3(75%)
Low cost fertility treatment	5 (31.25%)	0
<b>Willingness to have fertility treatment</b>	15 (93.75%)	4 (100%)

TABLE 3: Attitude of Patients with Turner Syndrome (n= 20).

preservation by the parents and TS patients were lack of information (41.2%), fear of complication (22.1%) and lower cost for hormonal treatment (15.3%).

#### 4. Discussion

Most patients or parents from non support group had a sufficient knowledge about TS than support group, particularly about the definition itself, types of chromosomal abnormality, the possible health issues, fertility impairment, and hormonal therapy. From the educational background, most of the participants are well educated. Most of them are participating in a support group and admitted that they got informations from the fellow peer group and from their pediatric endocrinologist. Yet, the information about future fertility preservation is still scarce, 62.5% participants from support group dan 75% participants from non support group know that there are still chance of fertility, yet they did not know about the alternatives of the treatment. Gonzales et al suggest that TS patient should already got anticipatory guidance regarding induction of puberty in 8 – 10 years old, obtain LH and FSH concentrations to assess for ovarian failure in 9 – 11 years old, begin low-dose estrogen monotherapy, begin the discussion regarding fertility reproductive options, and encourage parents to initiate and discuss with the daughter in 12 – 14 years old [7]. As the ovarian function decrease overtime, even when patient is in prepuberty age, the information about fertility should be delivered to the parents. The parents are also should be capable of making decision for their daughters, considering in that age patient is not competent to make such huge decision. Within this age, patients usually come to their pediatric endocrinologist, thus this informations can be started during their visit. Patient can also be referred to infertility specialist to discuss about the feasibility to have fertility treatment. If the patient already knew about the possibility of being pregnant by doing oocyte preservation in prepubertal age, they will not regret in the future. The other method of

having fertility, which is by oocyte donation, is not feasible, because it is prohibited in Indonesia. Although advanced development in reproduction technology has brought alternatives for TS patient. Fair information about the possibility of having miscarriage in should be informed. The information about certain medical condition that can occur in patient with TS such as aortic dilation that can complicate pregnancy also should be delivered to the patients and parents. Few centers will assess whether any cardiac problem before performed fertility treatment in patient with TS, and even after the patient get pregnant, routine echocardiography check every trimester were obtained to make sure there were no cardiac abnormality interfering the pregnancy.

This study showed that 31.25% patients from support group and 25% patients from non support group believed that TS patients still have normal social life. Wasserman illustrate that some of the issues and challenge in patient with TS are difficulties associated with social attitude and behavior, such as relentless teasing in school, nevertheless, there are still many that married and professionally succesful [11]. Parents and patients should be gathered in a supporting group setting, to share their stories and encouraged each other.

Fertility is still a main concern in TS patient and parents (support group 87% and non support group 100%), even in the presence of another health issues. 87.5% from support group and 75% from non support group still expect to have spontaneous pregnancy in the future. Participants, particularly the parents, are still worried about health problems that can occur, such as cardiac problems and renal problems. Participants that concern about the fertility issues are mostly the patient themselves, which were aged above twenty years old. Most of them have the willingness to have fertility treatment available (support group 93.75% and non support group 100%), yet 31.25% of them insisted on lower cost of hormonal treatment. The greatest barriers for accepting fertility preservation by the parents and TS patients were lack of information (41,2%), fear of complication (22,1%) and lower cost for hormonal treatment (15,3%)

The challenges of counseling and fertility treatment for TS patients in Indonesia are limited window period of diagnosis and the possible fertility treatment and lack of knowledge about TS fertility condition by both pediatric endocrinologist and parents; therefore a team consisting of pediatric endocrinologist, infertility specialists and support group is recommended in these setting.

## 5. Conclusion

Most TS patients who participating in a support group have better knowledge about their conditions, therapies and problems than TS not participating in support group. Almost all TS patients agree to fertility treatment and excited in the possibility in having children trough fertility preservation. Fertility is still a main concern, even in the presence of another health issues.

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