Issues Concerning the Notification of Turner Syndrome: A Questionnaire Study of Clinicians

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Abstract. The notification of Turner syndrome (TS) is an important task for clinicians who are treating the condition. To clarify the situation regarding the notification process, we performed a questionnaire survey among 104 clinicians who were involved in the treatment of GH and estrogen for TS in the Kanto and Koshinetsu area of Japan. We distributed a questionnaire concerning both the ideal and actual timings of notifying the name of “Turner”, short stature, hypogonadism, fertility, complications, karyotype, as well as asking “Who is the best person to notify the patient?” We received 34 completed questionnaires, 30 pediatricians, two physicians, and two gynecologists, who were treating a total of 352 cases with TS. Although most clinicians understood the gradual notification process, there were difference between the ideal and actual timings of notifying the name of the condition, short stature, hypogonadism/fertility and karyotype. To the question “Who is the best person to notify the patient?”, replies were, ideally: the doctor followed by parent(s), and, actually: the parent(s). Only five institutes had medical teams to care for TS. The team consisted of clinicians, nurses, and genetic counselors. Concerning the notification process of TS, it is important for us to tell patients that Turner syndrome is a condition, not a disease, at an appropriate time with a gentle attitude. At the same time, we should realize TS support groups could play an important role in contributing to how to take care of the many problems of TS.

Key words: Turner syndrome, notification, questionnaire, parents, clinician

Introduction

Turner syndrome (TS) is the most common chromosomal abnormality in phenotypic females (1). Absence of an X chromosome or the presence of a structurally abnormal X chromosome causes TS. The principal features of TS are short stature,

primary hypogonadism, and some complications. Early diagnosis of TS, appropriate treatment of TS-related disorders at the corresponding age, provision of notification of TS to patients and their parents, and a supporting care system are needed (2). When clinicians inform TS patients about their medical situation, there are many factors to be considered. The notification process of TS is an important task for the clinicians who are treating TS patients. A transcultural study of TS revealed there were some differences about the secrecy and ignorance concerning TS between some countries

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Fig. 1 Department and specialties who replied to the questionnaire.

(3). For this reason, there are rare reports about the notification of TS in Japan (4). To clarify the situation of the notification process, we performed a questionnaire survey among 104 clinicians who were involved in the treatment of GH and estrogen for TS in the Kanto and Koshinetsu areas of Japan.

Methods

We performed a questionnaire survey among 104 clinicians who were involved the treatment of growth hormone (GH) and estrogen for TS in the Kanto and Koshinetsu areas of Japan. The questionnaire was concerned with both (I) the ideal and (II) the actual timings of notifying patients of: (a) the name of their condition, (b) short stature, c) hypogonadism/fertility, (d) karyotype, and (e) complications-I / II / III, as well as asking (f) “Who is the best person to notify patients?”. Complications consist of three categories, I: otitis media and hearing loss, II: obesity, diabetes mellitus, and heart disease, III: osteoporosis and thyroid disease. The ideal and actual timings of the notifications in the questionnaire were categorized onto 4 periods consisting of pre-school, elementary school, junior high school, and high school or later. To the

question concerning “Who is the best person to notify patients?”, there were five possible answers (i) the parent(s), (ii) the doctor followed by the parent(s), (iii) the parent(s) and the doctor together, (iv) the parent(s) followed by the doctor, and (v) the doctor only. There was also a question about the support system caring for TS patients.

Results

We received 34 (32.7%) completed replies (30 pediatricians, two physicians, and two gynecologists. The specialties of the doctors who replied consisted of endocrinology, neonatology, diabetes, nephrology, genetics, and others (Fig. 1), and they were treating a total of 352 females with TS, with age distributions at diagnosis and their present ages of TS as shown in Fig. 2.

About the timing of notifying patients of the name of their condition, the ideal and actual timings were given as before school attendance, 25% and 4%, respectively. Actual notification of the name of their condition was only given to 40% of patients by junior high school days, compared to 80% declared as the ideal timing (Fig. 3-a).

The results of the answers to short stature were similar. The ideal and actual timings of

Fig. 2 Age distribution of Turner syndrome patients.

Fig. 3 The percentage of replies for (I) the ideal timing and (II) the actual timing to notify; (a) the name of the condition, (b) short stature, (c) hypogonadism/fertility, and (d) karyotype.

Fig. 4 The percentage of replies for (I) ideally person and (II) actually person, to the question concerning “Who is the best person to notify patients?”; (i) the parent(s), (ii) the doctors followed by parent(s), (iii) the parent(s) and the doctor together, (iv) the parent(s) followed the doctor caring for patients, and (v) the doctor only.

notification for before school attendance were 45% and 10%, respectively, although 40% of patients were notified of short stature during their elementary school days (Fig. 3-b). In this study, approximately 25% of TS patients were informed that short stature was a feature of their condition during their high school days.

The question about hypogonadism/fertility revealed a striking disparity between the ideal and actual timings of the notification (Fig. 3-c). The rate of the actual timing was 62% in elementary and junior high schools in total, equal to the ideal timing of 64% by elementary school days. Regarding complications, the ideal timing of the notification was earlier than the actual one in all of the categories. Finally, for the notification of chromosomal abnormalities, around three quarters of clinicians responded that the actual timing of the notification was after high school days (Fig. 3-d). Although most clinicians understood the step by step notification process, there were differences between ideal and actual timings of notifying patients of the name of their condition, short stature, hypogonadism/fertility and karyotype.

To the question “Who is the best person to

notify patients?”, most replies were, ideally: mostly the doctor followed by parent(s), and actually: the parent(s) (Fig. 4). However, there was no significant difference between the two timings. Although many clinicians hope to notify patients themselves, followed by parent(s), notification of TS depends on parent(s).

In the free comments section of the questionnaires, problems with the notification process were noted. How do you approach patients or their parent(s), rejection by patients, late diagnosis, and unwished for these notification were the problems noted in the order of frequency. Only five institutes had medical teams to care for TS, and teams were made up of clinicians, nurses, social workers and genetic counselors.

Discussion

Religious, cultural, historical, and social factors influence the acceptance and understanding of diseases and conditions. The notification processes of the name of a disease in children and adolescents have some particular problems, because the patient, their parent(s), and medical staff including medical doctors are all involved. A transcultural study of TS reported that there were some differences about the secrecy and ignorance concerning TS between some countries and the need support societies for TS patients (3). Japanese social and cultural characteristics, which are slightly different from those in the West, might lead to fewer reports concerning the problems confronting for TS patients and their families. Harada *et al.* reported that many mothers and medical doctors had the intention of confronting TS patients but they deliberately held back from doing so (4). They recommended that medical doctors should know the personalities of the patients and their parents, the level of the tolerance in the family, and the support system for both (e.g. TS group, medical staff), and that the explanation of all conditions related to TS would be better at the time of diagnosis.

The present questionnaire survey revealed differences between the ideal and actual timings of notification of some items, especially the name of the condition, short stature, hypogonadism/fertility, and karyotype. In all items, the actual timing of the notification was later than the ideal one. These results represent two aspects. One was the presence of some hesitation, deliberation, and confusion on the part of clinicians responding to this survey, and the other was delayed diagnosis of TS. The former problem might include fear of TS patients and/or parent(s) rejecting the diagnosis or not wishing to be notified. Almost all clinicians notified TS patients and/or their parent(s) at the diagnosis and knew the gradual notification process was most important. In a Japanese study, 33% of mothers were reported to want to hide TS (4), and prevalence of this attitude might lead to later notification. The late notification problem could be resolved with earlier diagnosis, leading to earlier provision of information relating to TS patients at the appropriate time. The problems concerning hypogonadism/fertility and karyotype (chromosomal abnormality) are serious matters for TS patients and clinicians need to work closely with clinical psychotherapists and genetic counselors.

In this study, the age at diagnosis and the current age of TS patients were various. When the age at diagnosis is relatively high, the actual age of notification is limited by the higher age. The highest prevalence of age at diagnosis was elementary school days, probably due to discovery through short stature. This might partially explain the difference of the timing in notification of short stature, but the actual timing of notification of the name of the condition, Turner syndrome, was 25% in elementary school days. Some girls, who are treated with growth hormone, might have no information about their condition. However, the difference between the ideal and actual timings of the notification was thought to be due to more than half the cases being of adult age in this study.

“Who is the best person to notify patients?” was the most important question. This study

showed the majority of replies were, the doctor followed by parent(s), in the ideal timing, and, the parent(s), in the actual timing. Both parent(s) and doctors are responsible for the task of notification to TS patients.

Recently, many reports have emphasized the great need of multidisciplinary care for TS patients to improve quality of life and reduce morbidity (1, 5). From our study, only five institutes in this area had medical teams caring for TS patients and these teams were made up of clinicians, nurses, social workers and genetic counselors. The establishment of TS societies or groups could provide more and better information concerning TS to the clinicians as well as TS patients and their families. Concerning the notification process of Turner syndrome, it is important for us to inform patients and their families that Turner syndrome is condition, not a disease, at an appropriate time with a serious attitude. At the same time, we should realize that TS support groups could play an important role. Such groups could contribute to how to take care of the many problems of TS patients. Attending or supporting proactively such groups could guide clinicians an appropriate timing for notification.

Conclusion

This questionnaire survey revealed there were differences between the ideal and actual timings of the notification process to TS patients. Early diagnosis and provision of information on TS patients could improve their quality of life. Comprehensive team medicine including some paramedical specialists would improve the transition from childhood with TS to adult medical care.

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