

“Difficulties in life” in Turner Syndrome: Comparative Analysis of Narratives of Patients and Their Mothers

Interdisciplinary Journal of Phenomena and Order 2020, Vol. 12
© “Phenomena & Order” Editorial Board 2020
PRINT ISSN:2188-9848
ONLINE ISSN:2188-9856
<http://kashida-yoshio.com/gensho/gensho.html>

 **Phenomena & Order**

Ryotaro KOGUCHI ^a

Abstract

This paper aims to make clear difficulties in life of patients with Turner syndrome (TS). It is said that mental troubles of patients are hard for doctors to understand because while they rarely occur to patients' consciousness, medical staff mainly observe their health status based on laboratory values or patients' complaints. Interview survey of 2 TS cases, oriented to patients and their families, shows two different narratives originated from the same experience of TS. For patients themselves this disease is experienced only as a small part of life. This results from its feature of imperceptibility for nonmedical people surrounding patients. Their colleagues regard them just as short women. On the other hand, patients' families are worried about their sterility and wish for their normalization. Families take life with TS more seriously than patients themselves. This is why patients cannot escape from their reality of TS. It is “a story of deprivation” and “of recovery” by families like these that patients experience as “difficulties in life”.

key words

difficulties in life, Turner syndrome (TS) , loneliness

1 Problem setting, purpose and methodology

Technical achievement of modern medicine enabled patients to live long as “people of remission”. In this paper, this “people of remission” includes those who have a chronic sickness. In Arthur Frank's “remission society”, a relationship between foreground and background of disease and health proceeds gradually interpenetrating(Frank1995=2002 : 27). That is, though people will go back to their conventional life, they'll never completely return to a condition they were normal in. This means nothing else but living their daily lives with agony originated from illness.

Medical staff handle with these pains by facing physical pains or disablements caused by diseases. In most cases, patients' health status is observed mainly based on laboratory values and patients' complaints.

However, patients' agonies are not just those physical. Pains related to diseases are mixed with agony of not being able to live an ordinary life, anxiety for whole life and more (Hoshino 2006:76). That is, “difficulties in life”. Medical staff are difficult to be conscious of that “difficulties in life”

as patients themselves aren't necessarily clearly aware of that “difficulties in life”. However, it is a fact that that “difficulties in life”, hardly told clearly or been conscious of, is indeed related to the existence of patients themselves. To give light on this “difficulties in life”, considering medical knowledge as premise, it is needed to adopt a microsociological narrative method. From this situation, this paper aims to make “difficulties in life” of those clear. As stated below, TS patients are able to live a daily life if they are treated with proper medical practices. Therefore, their “difficulties in life” is less apparent, clearly spoken about, or recognized. Hence to unveil the real fact of implicit “difficulties in life”, cases of TS will give us many suggestions.

Specific cases as objects of observation are A, who is in her twenties and has been affected for 18 years B, also in her twenties and affected for 14 years. From October 2017 to May 2018, I did interviews with A, B and their mothers 4times each, 16times in total. I adopted a semi-structured interview method as an interview technique. The method enables me to collect information in accordance to interviewees, while maintaining to stay into a certain degree of direction. Therefore, in some cases, contents can be unintentionally

^a Gifu University, Gifu, Japan
E-mail : ryotaro.koguchi@gmail.com

structured. In some parts, interviewees accidentally talked so structurally about before and after the outbreak in the present cases as well. This paper utilizes those structural talks and rearrange to extract the common “difficulties in life” of the two.

2 What Turner Syndrome is

TS is an endocrine disease like childhood-onset type 1 diabetes. They both ultimately delegate medical practices to patients themselves and their families. Remission is achieved by patients themselves and their families.

From the beginning, TS is a generic name of diseases originated from a general or partial deficiency of chromosome according to Japan Intractable Diseases Information Center and Information Center for Specific Pediatric Chronic Diseases, Japan. Its main pathophysiology is hypogonadism. It expresses only in born girls and the number of the patients is assumed to be about one person in a thousand.

As main symptoms, (1) short stature, (2) an influence on secondary sexual characteristics, (3) a menstrual disorder are given as main symptoms. Especially, a low body height almost inevitably causes. Moreover, many of them become sterile as hypogonadism is the main pathophysiology.

Many of them see a pediatricist when their short statures get prominent in higher grades in elementary school, which makes it clear that they are affected. Then, a notification only to their mothers is recommended as a standard medicine. Therefore, patients have less opportunity to be told test results by doctors. Substitution of somatotrophic hormone(injections) is conducted for shortness in stature and substitution of female hormone (e.g. patches, oral agents) for hypogonadism, which is continued under self-management. Thus, they can live a daily life with intervention of proper medical practices. In this sense, the term “Turner women” is preferred, emphasizing that it is not an illness or disability, but a kind of conditions.

3 Case of A

A was diagnosed at the age of 11 and now works as a nurse. When diagnosed, a pediatricist who was their family doctor didn't tell her that but told her

mother. A herself got notified of it 10 years after when she was 21 years old by her mother and physician who was their family doctor then. Below, we'll first look at A's mother's story and then A's.

3.1 A's mother's story

A's mother talks about “outbreak”, “impressive events” and “what she now wishes”, as follows.

3.1.1 Outbreak

I took her to a hospital as she was a little short for a 5th grader. When told the results of the checkup, I felt like the end of the world. I first tried to find and talk to some other mothers who have a child suffering from TS. I was shocked that it was difficult for her to give birth to a child, however, the more I'm told about it, the more it made me think my daughter can't have a child, which made me feel difficult to answer like the doctor in the hospital if she asked me “Why, mom, why?”.

3.1.2 Impressive events

As expected, when I notified my daughter that she might not be able to bear children, I would help her and I'd do my best. It was pretty tough to. Why I couldn't tell her for about 9 years was that I thought soon the medicine would develop and she would be fine or heal. Also, I didn't want to bother her by telling unnecessary things since he was studying hard (at school). However, as a matter of course, she and I had a conflict due to the disease.

3.1.3 What she now wishes

I wish there would be a system which TS patients, would be able to be pregnant and give birth to a child in. I wonder if egg providing or egg banks won't be established. I wish to say at least “This can help, that can help.” not only to my daughter but to other Turner women.

3.2 A's story

A talks about “before getting notified”, “when getting notified”, “after getting notified”, “what she now thinks” and “being pregnant”, as follows.

3.2.1 Before getting notified

I didn't think that much about going to a hospital as I often visited the place due to otorrhea or fever, which wasn't annoying to me. ...Though I never thought I was suffering from TS, I felt something was wrong since some of friends started their period. It made me so exhausted to play along with their story about unfamiliar topics (physical

subjects) when talking with my female friends in junior high school.

3.2.2 When getting notified

(With my mother when notified) conflict...to speak strictly, we had a conflict. Well, how shall I express it, it's difficult, it's not that I had TS or not being pregnant, but simply, why I wasn't told for such a long time though I am the person, although I understand my mother's feeling as well. It's hard to tell. However, if she had told me, it must have made me able to examine or ask how I live as much as I want. I won't go the length of thinking I might have a different life, yes. It's just that I wanted to know. Like "She should have told me". Thought I may have entered my late period of rebelliousness, we had conflicts and yes, I feel sorry for my mother.

3.2.3 After getting notified

The first thing in my mind was "I'm glad I'm a nursing student". In a sense that I can make my own living. Though I felt anger to my mother, I was studying hard as a nursing student enough to feel like "It's TS, just as I thought." It was initially rather shocking for me to think "I might have it." when reading text books or training.

I was disgusted with my mother in that she still didn't expect I knew that. ...Now if I think about it, I aim to become a nurse because I wanted to know what is going on my body. Of course, most of the reason is that it's a familiar job.

3.2.4 What she now thinks

When I communicate with a patient, I sometimes wonder if they "want to say like this". I'm also after all a patient so I could act not as a nurse but too much from the perspective of patients. That makes my work stagnant, so I stop it at some extent and keep distance to work.

3.2.5 About being pregnant

I'll think about having a child some time when I get to want them. I'd go to America. I'd rather do so if things turn out than wait until the system changes. My mother thinks I'm most distressed in that it's impossible for me to expect a child. However, I'm thinking of going to United States when I come to desire that. But I never told this to her as I understand her feeling.

4 Case of B

B was diagnosed at the age of 11 and now works as a schoolteacher. Like A, when notified, a

pediatrist who was their family doctor didn't tell her that but told her mother. However, different from A, she was told 2 weeks after by her mother and her family doctor. Below, we'll first look at B's mother's story and then B's story.

4.1 B's mother's story

B's mother talks about "outbreak", "impressive events" and "what she now wishes", as follows.

4.1.1 Outbreak

Though I wasn't really a problem, I took her to a hospital since B's elder sister, by 4 years, said to me "B hasn't started her period yet." and also B was short. Regarding notifying B, I chose to tell her quickly as I thought it impossible to hide it to B because of her sister though I wondered if she could accept it. Shocked by the fact she can't expect a child, I asked the doctor not to tell her that.

4.1.2 Impressive events

Indeed, she'll live a different life from her sister. It's always on my mind that B can't do what her sister normally does. ...As her parent, I think she needs to work hard on many things as much as she has disadvantages with her body and is shorter than others. Her sister helped me a lot by taking care of her. Because, B herself was taking easy, they were getting along (since she was little) more than expected. ... (After notified) B often copied her sister and told me saying "I was able to do such a thing!"

4.1.3 What she now wishes

Did you ask this question to A's mother as well? Just as her, I wish there would be a system those TS patients would be able to have a baby. I'd like to let B, A and at least other Turner women in X prefecture experience bearing a child. Especially, X has been to America for several times with obsession, however, we parents can't have Turner women do such a thing.

4.2 B's story

B talks about "before getting notified", "when getting notified", "after getting notified" and "what she now thinks", as follows.

4.2.1 Before getting notified

After visiting to a hospital, I felt something was wrong as I heard from my sister "Usually, these things will happen" or "I told mother" When

notified, I felt like “Ah, finally”. Until that, I was waiting wondering “when I will be told”.

4.2.2 When getting notified

I have in my memory that I was cheered up a lot that I could live normally in spite of my illness. However, I myself didn't really understand well even told I'm TS. Though my mother told me it would be fine as I'd be able to live an ordinary life if I got treated properly, I remember thinking “I got affected by a pretty serious disease” every time the doctor in the hospital and my mother encouraged me like that. What comes after is that my sister was so noisy, really, but the second daughter also had a hard time.

4.2.3 After getting notified

I wasn't shocked or damaged, nor probably wasn't that important that I can't become pregnant. I was just confused to get told something like “It's perfect if you could do like your sister” when I wanted to think things like “how I should live from now on”. ...For me, suffering TS didn't mean having intellectual disabilities, so I wanted to step forward when in junior high school....Since my homeroom teacher and nurse-teacher were kind, I've hoped to become a schoolteacher since I was a junior high school student.

4.2.4 What she now thinks

I'd like to be a good teacher. Although it's not clear what good teacher is, however, to them, having TS has nothing to do with them. ... (In children's daily life) It'll be nice if I can be their educational material. ... And also, either would be fine for me about acceptance of disease, though the doctor (family doctor) and my mother suppose I was able to accept my illness because notified early. Though I was told (by our family doctor and my mother) it's difficult to be pregnant if I couldn't accept my sickness and though I understand I'm not able to have a child and my mother's feeling, in my case, also because my sister got married, I wonder if they still tell me more that there would be a society I too could live an ordinary life.

5 The result and analysis of the interview

As a result of interviewing, A and B seems to regard their life at workplace as important. In relationship with patients, students and co-workers, who spend time together, they are conscious of how they can utilize their experience as those who

themselves have TS, act and talked about their future prospects. On the other hand, A's mother put emphasis on sterility and fertility. Besides, B's mother referred to that B “isn't able to live an ordinary life” like her sister, comparing the sisters. It turned out that A and B are playing along with those talk in front of their mothers and families in their lives.

Delivery and fertility spoken by A and B's mother is firmly influenced by their experiences of having been able to be pregnant and have a child. For example, B's mother said “Bearing a child and becoming a mother made me grow and changed me in many respects. It's unfair she has been unable to experience that since birth due to her disease and I'd like to let my daughters do”. However, A and B are trying to choose a different life even though “they understand what their mothers feel”. It is related to that they take TS as a disease “they don't think about curing or is able to be cured.” unlike their mothers.

Their experience of being affected by TS is, for them who caused the disease in their childhood, almost part of their social lives. Although their outstanding features are small statures, those matured are just seen as short women. A and B are about 1.5 meters tall. Unless one is a medical staff, it can't be judged, nor do they take it seriously.

As they've continued to be patients, for them, a treatment like supplying growth hormone and female hormone is an embodied everyday life. However, at the same time, they are being unable to escape from the fact they are TS patients by attuning themselves to a story of recovery saying “Although they can't have a child in modern medicine, they might be able to if female hormone develops.”. For they are forced to remember and face themselves suffering from diseases by the story of recovery when having the disease is part of their lives. Told a process to recovery, they are made to recognize they need to aim recovery or a situation of not recovering isn't perfect. Indeed, B's story of “still being told there would be a society she could live an ordinary life” is, to put it the other way, compelling

themselves, due to the surroundings, to face that they aren't normal yet and eventually oblige them to be unable to run away from the reality of themselves having TS. Moreover, the two developed the illness in childhood, so they don't have a comparison axis of when they were healthy. Therefore, when told “the story of recovery”, they don't feel real and rather make them conscience of “myself who suffer from the disease”.

6 Conclusions

Thus, this paper made it clear from the interview data of TS patients themselves and their mothers what their “difficulties in life”. Also, it was conducted successfully to interview about one common experience as two different stories by doing individually. From this, we can concretely identify following 3 points.

- ① Keeping being called “Turner women” by medical staff and their mothers, a term that emphasize not disease or disability but a sort of constitution. That is, they are involved in medical context and unable to escape from it.
- ② Talking about TS as a disease “they don't think of curing or is able to be cured.” as affected in their childhood, however, playing along with their mothers' and families' “story of recovery”. Developed the illness in childhood, they don't know “when they were healthy” as a comparison axis, so it's unable to tell “the story of recovery” for them even though they wish to.
- ③ In the first place, they apparently live a normal life.

Taking these three into account, let me mention that their mother's loneliness shouldn't be overlooked. For instance, like A on this paper, so many patients took long before notified by their family doctor. Also, there aren't many cases earlier notification was done because of their siblings like B, as B's mother told.

Meanwhile, mothers are often lonely who can't tell their daughters the truth. By not being able to do so, they need to tell to their family doctors their daughters' condition for a month as far as they observed once a month when visiting a

hospital together. Not being able to tell the truth to their daughters and having a few people for them to take counsel to, they need to face the fact alone. Also, they can't help feeling anxiety or isolated for that they themselves and other family members live a usual life and, in comparison to people around, their daughters are not able to live an ordinary life. That's why mothers wish for their daughters' delivery appear in their talks. Their solitude is there.

Certainly, it is A and B's “difficulties in life” that they live attuning themselves to their mothers' and families' stories. However, it's also important to listen to the lonely mothers' stories and then back them up. Both TS patients and their mothers are respectively feeling isolated in a different context. The former feel loneliness escaping from a medical context and the story of recovery. They can't adjust themselves to those stuffs nor can they easily share their feelings to someone. In that sense, they feel alone. However, their mothers, who have to face their daughters' disease by themselves are also lonely. Originally, on one hand, agony in illness is individual and subjective, on the other hand, it is deeply connected to a social relationship. (Hoshino 2006: 76). That means, this loneliness is also one of “difficulties in life”.

As made clear in this paper, the patients' “difficulties in life” are established in their relationships with their families and not the same as their families' “difficulties in life”. What became unveiled at the same time is the patients' families are also “people concerned”, who have “difficulties in life” originated from the disease. These facts won't appear from a medical observation (medical examination, medical diagnosis) that faces patients in physical signs. However, it wasn't able to show in this paper what kind of approach(treatment) is possible for these “difficulties in life”. I'd like to consider this as my future theme.

【Note】

- 1) This paper is the modified English version of Koguchi, Ryotaro (2019) "Difficulties in Life of Remission Patients: Sociological Study of Turner Syndrome" in: Tsukuba Journal of Sociology, vol.44, 53-62. Citations hereby refer to "Interdisciplinary Journal of Phenomena and Order" vol.12.

Article

- 2) Both agony and “difficulties in life” are described in this paper. Both express physical and mental pain whereas “difficulties in life” especially indicates pain related to an existence of an individual in living a life.

Reference literature

Frank, Arthur W., 1995, “The Wounded Storyteller: Body, Illness, and Ethics”, *The University of Chicago Press*. (=2002,鈴木智之訳『傷ついた物語の語り手——身体・病い・倫理』ゆみる出版.)

HOSHINO Shin, 2006, *Narrative and Medicine (Narrative To Iryou)*, Kongoushuppan. (=星野晋,2006,「医療者と生活者の物語が会うところ」江口重幸・斎藤清二・野村直樹編『ナラティブと医療』金剛出版.)

KOGUCHI Ryotaro, 2017, ”Difficulty in living from a sociological point of view : a case of juvenile diabetes(type 1)” , The 51st Annual Scientific Meeting of the Japanese Society for Pediatric Endocrinology (Dai 51 Kai Nihon Shouni Naibunpi Gakkai) , Report. (=高口僚太朗, 2017, 「社会学の視点からみた「生きづらさ」とは——小児期発症 1 型糖尿病の 1 例」,第 51 回日本小児内分泌学会,報告要旨.)

KOGUCHI Ryotaro, 2019, ”Difficulties in Life of Remission Patients: Sociological Study of Turner Syndrome”, *Tsukuba Journal of Sociology (Shakaigaku Zyanaru)* , Vol.44, 53-62. (=高口僚太朗,2019,「寛解者の「生きづらさ」——ターナー症候群症例の社会的考察」『社会学ジャーナル』44: 53-62.)

Reference URL

Japan Intractable Diseases Information Center,
Accessed 3 Mar 2020
(<http://www.nanbyou.or.jp/entry/652>)

Information Center for Specific Pediatric
Chronic Diseases, Japan, Accessed 3 Mar 2020
(https://www.shouman.jp/disease/details/05_41_088/)

【邦文要旨】 ターナー症候群の「生きづらさ」 —母と娘の語りの比較分析から—

高口 僚太郎
岐阜大学

キーワード：生きづらさ，ターナー症候群，孤独

【背景】

医療者は、検査値や患者の訴えを中心に、ターナー症候群患者の健康状態を観察することが多い。一方で、患者自身の「生きづらさ」は、患者自身が意識することも少なく、医療者はその「生きづらさ」を捉えることが難しいとされている。

【目的】

ターナー症候群(以下、TS という。)の「生きづらさ」とは何かを明らかにする。

【ターナー症候群(Turner syndrome)とは】

TS は、X 染色体の全体または一部の欠失に起因した疾患の総称である。性腺機能不全を主病態としている。また、出生女兒のみに発現し、患者数は約 1000 人に 1 人と推測されている。原因は、X 染色体の数的・構造的異常と考えられているが、発症にいたるメカニズムについては不明な部分が多い。X 染色体の不均衡による、遺伝子発現量が低下することが原因である。おもな症状は、(1)低身長、(2)卵巣機能不全による二次性徴、(3)月経異常などが挙げられる。とくに低身長は、TS にほぼ必発の症状で、SHOX 欠失、GCY 欠失、染色体不均衡により生じる。さらに、性腺機能不全を主病態とするため、不妊となる場合が多い。多くの当事者は、小学校高学年のころ低身長が顕著となり、小児科を受診することで明らかとなる。このとき、多くの当事者が医師から診断結果をきくことはなく、母親にのみ告知することが標準医療行為と推奨されている。とはいえ、低身長に対しては、成長ホルモンの補充療法(注射剤)を、卵巣機能不全には、健常女性の思春期来発年齢を指標にして、女性ホルモンの補充療法(貼付剤、経口剤)を開始し、その後は自己管理のもと継続することから、適切な医療行為の介入があれば日常生活をおくることができる。その意味で、病気・障害ではなく一種の体質ということを強調

した「ターナー女性」という呼称を推奨している。

【方法と結果】

患者歴 19 年目の 20 代女性(看護師)と家族、患者歴 14 年目の 20 代女性(養護教諭)と家族に、2017 年 10 月から 2018 年 4 月の間にそれぞれ 4 回の非構造化インタビューを行った。その結果、2 症例とも、職場での生活を重要視していた。そこで共に過ごす患者や生徒、同僚との関係性のなかで、いかにターナー女性である自身の経験が有効活用できるかを意識し、実施し、今後の展望を語っていた。一方で、2 症例の母親は不妊に主眼をおき、「女性性の剥奪」の経験として語っていた。そして 2 症例は、母親や家族の前ではそうした「女性性の剥奪」の語りに自身を合わせながら生活していることが明らかとなった。

【考察】

ターナー女性であるという経験は、小児期発症の 2 症例にとって、自身の社会生活のほんの一部である。低身長という顕著な特徴はあるが、成人している彼女たちは小柄な女性とみられる程度で、医療者でなければ一見して判明するものでもなく、彼女たちも深刻に受けとめてはいない。確かに、成長ホルモンと女性ホルモンの補充は行っているが、患者歴を重ねていることからこうした治療方法は身体化された彼女たちの日常である。しかし実際には、母親や妻の発話に完全同意する父親の「現在の医学ではこどもが産めないけど、女性ホルモン剤が発達して産める身体になるんじゃないか」という語りに自身を合わせることで、TS である現実からは逃れられないことが明らかとなった。2 症例への聞き取りを本人と家族個別に実施したことで、ひとつの共通する経験からふたつの異なる語りとして聞きとれた。アーサー・W・フランク『傷ついた物語の語り手』を用いれば、母親は「回復の物語」、本人は「探求の物語」

Article

と分析できる。場面としてではなく、ストーリーとして症例を観察することが重要である。

【結論】

母親や家族の語りに自身を合わせながら生活していることが、2症例の「生きづらさ」である。今後は症例を増やして検討する必要がある。

【編集後記】『現象と秩序』第 12 号をお届けします。本誌ではじめて英語論文が投稿されたことから、投稿規定・執筆要領の改訂をおこないました。今後、英文での投稿をお考えの方々もぜひ本誌に玉稿をお寄せください（編集委員紹介制ですのでまずは委員にご相談を）。

今回は、医療・福祉領域の 4 本の論考が掲載されています。第一論文は、発話も動作も困難な人と、「握る／握らない」というかたちで手を使って意思疎通する際に陥りがちな問題を取り上げたビデオエスノグラフィです。第二論文は、諏訪哲史の『アサッテの人』と三島由紀夫の『金閣寺』という二つの小説のなかで描かれている吃音者像の比較検討を通じて、前者における吃音者像の新しさを論じています。「アサッテ」という言葉に「時間でも空間でもなく、消滅という作用」を読み取る、興味深い論考です。第三論文は、生活看護学という新しい領域を切り開こうとした意欲的作品です。非致死的な病気を抱える患者 3 名へのインタビューに基づき、かれらが医療とのバランスを取りながら、病気をいわば人生の資源とし生活にむしろ彩をもたらすものとして経験している様子を描いています。英語論文の第四論文は、「ターナー女性」2 名とその家族へのインタビューに基づき、「場面としてではなく、ストーリーとして症例を観察する」必要性を見出しています。いずれも医療・福祉をクリティカルにまなざす論考として読みごたえがあります。ご堪能いただければ幸いです。

ところで、本誌編集作業中に新型コロナウイルスの感染が拡大し、世界的に未曾有の事態となりました。グローバルなモビリティの時代である現代、人間・モノ・資本・情報だけではなくウイルスも人間を“乗り物”として移動しているのだと痛感させられます。高齢者などが重症化する傾向にあると言われるなか、「集団免疫」という選択肢にはどうも違和感を覚えます。死者が〇万人で抑えられればいい方だという物言いに対しても同様です。“人為的に”淘汰される人がいることについての想像力は、「正しく恐れる」（寺田寅彦）ことの必要性が叫ばれる現在、あらためて持ち備えて、あるいは鍛えていきたいものです。（H.Y.）

『現象と秩序』編集委員会（2020 年度）

編集委員会委員長：堀田裕子(愛知学泉大学)

編集委員：檜田美雄(神戸市看護大学)、中塚朋子(就実大学)

編集幹事：尾崎友祐(神戸市外国語大学)

編集協力・印刷協力：村中淑子(桃山学院大学)

『現象と秩序』第 12 号 2020 年 3 月 31 日発行

発行所 〒651-2103 神戸市西区学園西町 3-4

神戸市看護大学 檜田研究室内 現象と秩序企画編集室

電話・FAX) 078-794-8074 (檜田研) , e-mail: kashida.yoshio@nifty.ne.jp

PRINT ISSN : 2188-9848

ONLINE ISSN : 2188-9856

<http://kashida-yoshio.com/gensho/gensho.html>