琉球大学学術リポジトリ

染色体構造異常のある子どもをもつ母親の子育てのプロセス

メタデータ	言語: English 出版者: 琉球大学 公開日: 2022-06-17 キーワード (Ja): キーワード (En): 作成者: 沓脱, 小枝子 メールアドレス: 所属:
URL	http://hdl.handle.net/20.500.12000/0002018082

学位論文

Mothers' experiences of parenting a child with chromosomal structural abnormalities: the journey to acceptance

琉球大学大学院保健学研究科保健学専攻

沓脱小枝子

INTRODUCTION

Chromosomal structural abnormalities are due to deletion (partial monosomy) and duplication (partial trisomy) of some parts of chromosomes. The number of diseases is innumerable depending on deletion/duplication and translocated chromosomal regions; however, the incidence of each disease is extremely rare, and for some only a few cases have been reported worldwide. For example, the 1p36 deletion syndrome, which is relatively frequent, has an estimated incidence of 1:5,000- 1:10,000 births (Heilstedt, Ballif, Howard, Kashork, & Shaffer, 2003). In Japan, this syndrome is not well known even among pediatricians, however, there is an estimated incidence of more than 1:39000 births (Sato, 2012). Many of these disorders occur rarely, but can occur even in parents with no obvious abnormalities themselves, but if either of the parents has a balanced translocation of chromosomes, it can also occur in the child by creating a defect in the genetic code in the gamete, leading to a child with chromosomal structural abnormalities.

In recent times there have been remarkable advances in genetic testing techniques, and information on chromosomal structural abnormalities has been increasing (Houdayer et al., 2013). This has improved the diagnosis of unexplained birth defects and increased the labelling of disorders using particular chromosomal deletion/duplication sites. These

advances have been accelerated by the establishment of the International Rare Diseases Research Consortium (IRDiRC) worldwide, followed by the Initiative on Rare and Undiagnosed Diseases (IRUD) in Japan.

Due to advances in medical technology, an increasing number of children in Japan with serious congenital diseases survive. Japan's medical policy has changed to recommend home care, and an increasing number of children with ongoing health problems including chromosomal structural disorders has now live at home (Nakamura, 2020; Nari, 2011). Children receive home nursing care or are referred by medical institutions to a treatment facility to visit depending on their physical and developmental status, and in many cases, the mothers are the primary care provider for their child because working fathers have little time to participate in housework and childcare (Cabinet Office, Government of Japan, 2017). In Japan's increasingly nuclear family-oriented society, many families do not have relatives in the neighborhood to rely on leaving them isolated despite support from home nursing service and treatment facilities.

Even after the diagnosis of chromosomal structural abnormalities, it is difficult for parents to obtain specific information for parenting children with chromosomal structural abnormalities (Kutsunugi, Tsujino, Murakami, Iida, & Endo, 2017). Each chromosomal structural disorder is rare, and even if a diagnosis is given, it is unlikely

that the mother will be able to find a specialist who is familiar with her child's disease. This can easily lead to a situation where the mother raises her child without knowing the best course of action. Houdayer et al. (2013) reported that the diagnosis of cytogenetic traits did not facilitate the parents' understanding of the disorder, that is, parents have difficulty accepting the condition if only a very technical explanation is given such as "1p36 deletion syndrome". Instead, in-depth knowledge about the child's chromosomal abnormalities is vital for parents having children with the condition (Gundersen, 2011), that allows them to apply the knowledge in a practical way in order to make choices about parenting. Waxler et al. (2013) stated that many parents wanted to know about what their child would be able to do and to be given a list of social resources at the time of diagnosis. They also hoped that medical professionals would discuss what is positive about the child (Waxler, Cherniske, Dieter, Herd, & Pober, 2013). Families with children diagnosed with Russel-Silver syndrome identified several stressors including endless worry; the lengthy process to confirm a medical diagnosis; efforts to adjust family roles; and negative responses to society's concerns (Weng et al., 2012). If a child has behavioral problems, it may also affect the well-being of the parent (Griffith, Hastings, Nash, & Hill, 2011; Reilly, Murtagh, & Senior, 2015).

Mothers' experience of raising children with disabilities can also influence their view about what is important in life and change their sense of values and the lifestyle of the family (King et al., 2006; Larson, 1998). Korkow-Moradi et al. (2017) explored the process from "initial reaction" after diagnosis to "new perspective" of mothers of children with Down syndrome and found that support from family members, including partners and siblings of children, and acceptance of the mother's role as an advocate for the child, will facilitate the process of gaining a new perspective. In contrast professionals' bias and stated lack of hope about the prospects for the child's future hinder this process. (Korkow-Moradi, Kim & Springer, 2017). A survey of parents of children with Zellweger spectrum disorders suggested that social, psychological, and physical support from the surrounding community may reduce the parents' burden and influence their perceptions and attitudes (Bose et al., 2019).

Mothers, who are often the primary care providers, have been the focus of research in this area. Previous studies have focused mothers' experiences at the time of diagnosis in the context of specific disorders (Strehle & Middlemiss, 2007; Bruns & Foerster, 2011; Waxler, Cherniske, Dieter, Herd, & Pober, 2013), and stresses during parenting (Weng et al., 2012; Reilly, Murtagh, & Senior, 2015; Adams, et al,2018); however, there is no published research which takes a longitudinal approach to mothers' experience of

looking after a child with a chromosomal structural disorder and how social interaction influences them along with the child growth. Clarifying how mothers raise children with chromosomal structural disorders perceive and respond to their situation and how medical and other professionals influence this situation will help nurses provide future support.

METHODS

Design

A qualitative research design based on a Modified Grounded Theory Approach (M-GTA; Kinoshita, 2003) was used for data collection and analysis.

Participants

The authors recruited mothers who had a child with a chromosomal structural abnormality to investigate their childcare experience in the absence of sufficient information about the disorders. The inclusion criteria were: (i) mothers with a child diagnosed with chromosomal structural abnormalities such as chromosomal partial trisomy and partial monosomy, and (ii) who looked after their child at home. Exclusion

criteria were mothers of children diagnosed with conditions for which there were organized support groups with members from all over Japan.

Data collection

The authors contacted mothers who belong to a family group for children with a chromosomal disorder or who transmitted through their website or blogged the experience of having a child with chromosomal structural abnormalities. Out of 20 mothers, seven consented to participate. Given the difficulty in recruiting a sufficient number from populations associated with rare diseases (O'Connor & Hemmings, 2014), we also used a snowball sampling technique, and nine mothers agreed to participate. In total, 16 mothers from eight different prefectures consented to participate, and each of them was interviewed once. Data were collected through semi-structured interviews based on the following interview guide: (1) demographics, (2) growth history of the affected child, (3) detailed experiences of caring for a child with chromosomal structural abnormalities, and (4) feeling toward support from health professionals. Each interview lasted an average of 55 minutes and ranged from 30 to 122 minutes. Interviews were held in a private room at a community center near the interviewees' home. Interviews were audiotaped and transcribed verbatim with participants'

permission: one participant declined to be audiotape, so handwritten notes were made immediately after the interview. Of the 16 interviews, six were with the mothers accompanied by their husbands; the husbands' comments were not included in the primary analysis. Interviews were conducted between September 2014 and February 2016.

Data analysis

To analyze the data, we used the M-GTA (Kinoshita, 2003), which is a tool for data analysis developed by Kinoshita in Japan based on the Glaser and Strauss's (1967) grounded theory approach (GTA). It is suited to the discovery of emerging patterns in data and has been extensively used in disciplines, such as healthcare, nursing, and education, to develop theory and to offer an explanation for the main concerns of the population (Creswell, 2003). In contrast to GTA, M-GTA does not perform segmentation of words and sentences (Odachi et al. 2017), but considers human's perception and behavior with its factors and conditions reflected in the context and sets "Analytic Focus" and "Analytic Theme" from the obtained data (Takeshita, 2019), to generate a theory within the limited range. An original worksheet is used to visualize the analysis process, and M-GTA is frequently used in Japan. In contrast to Kepreotes

(2014), who used ethnography to analyze the experiences of mothers who had a child with a rare disease, we focused on changes over time in the experience of parenting occurring and M-GTA was therefore chosen as the most appropriate methodology.

Data were analyzed by: (1) carefully repeatedly reading and interpreting the meaning of the data, focusing on that which is relevant to 'Analytical Theme' and 'Analytical Focus to generate concepts using the analysis worksheet. The analysis worksheet included the names of the concepts generated, definitions, the place on the data where they represent the phenomenon as a variation, and theoretical memo. (2) After a concept was generated, it is analysed to assess whether it is similar or opposite to the phenomenon represented by other concepts and generate categories, (3) Data collection was concurrent and analysis continued until theoretical saturation was reached, and the study phenomenon explained by the generated categories and concepts. Through these processes, a diagram showing the mutual relationships of concepts and categories was created.

All the transcription, categories, and concepts were provided and analyzed initially in Japanese. We then translated them into English and discussed and verified the comprehensibility and accuracy of the translation with a native English-speaking author.

Rigor

This study was supervised by two professors specialized in pediatric nursing and genetic nursing to ensure trustworthiness (Lincoln & Guba, 1985). Besides, both the content and process of analysis were presented several times at the M-GTA Study Group in Japan, and advice is given on the interpretation of data from various researchers to minimize bias. Our research team repeatedly discussed concepts, definitions, and categories throughout the analysis to increase trustworthiness.

Ethical considerations

Ethical approval for the study was obtained through the ethical review board of the University of the Ryukyus (669) and Yamaguchi University (63-4). Before the study, we informed each participant verbally and in writing about the study purpose, and assured them of anonymity and confidentiality. We removed identifying characteristics of participants and potential participants were assured that non-participation would not disadvantage them in any way and that they could stop the interview at any time and that if they decided to withdraw consent, they could do so. None of the participants chose to do this.

RESULTS

Participants' characteristics

Participants' demographic details, including the child's disorder, are outlined in Table 1. In order to recruit sufficient numbers from a rare population, we did not set an age limit for their child with the disorder and in 4 cases mothers discussed the care of their now adult children. The mean age of the children was 13.1 years (range: 2-30). Of 16 children, a total of 12 children (75.0%) had congenital heart disease, and particularly children with 22q11.2 deletion syndrome had severe heart disease such as tetralogy of Fallot. Six children (37.5%) had epilepsy results from 1p36 deletion syndrome and 2q duplication syndrome. All of the children experienced illness or disability, but its degree ranged in severity. Two children (12.5%) required specialized medical equipment to support breathing and nutrition; Nine children (56.3%) could walk and were able to attend school or Kindergarten, and two older children (12.5%) went on to have employment. Eight children (50.0%) were diagnosed at between two weeks to two months from birth; one child (6.3%) was diagnosed at three months, and six (37.5%) were diagnosed between the ages of one to four, while one child (6.3%) was diagnosed at eight years old. Of the 16 mothers, 12 (75.0%) had connections to the families of children with the same illness, and four of them had started to have those connections

because of their message on the website. Five mothers reported their experiences on a blog.

Table 1

Pediatricians were the first to inform the families of the diagnosis in every case. Following the diagnosis, four mothers and their husbands underwent genetic testing, and it was clarified that the child's disorder was not hereditary. The other 12 mothers had not undergone genetic testing, because they were advised by their treating doctors that chromosomal abnormalities are often non-hereditary

Categories and concepts

The analytic theme "the process of mothers finding their way to raise their child with a chromosomal structural abnormality only with little information" emerged from the data and the analytic focus was on "mothers raising their child with a chromosomal structural abnormality."

As a result of the analysis, 35 concepts, nine subcategories, and six categories emerged. The six categories were 1: Concern about abnormalities, 2: A healthy child is considered as a standard, 3: Deepening attachment to the child, 4: Acceptance of the

child as s/he is, 5: Changing attitude toward disabilities, and 6: Creating a frontier for other mothers.

After giving birth, mothers were concerned about abnormalities (Category 1), and even after being diagnosed with the abnormality in the chromosomal structure, were raising the child using a healthy child as the standard (Category 2). However, in the course of their child's growth and involvement, they achieved a "deepening of attachment" (Category 3)" and gradually changed to a view of "acceptance of the child as s/he is. (Category 4)" Mothers went back and forth between categories 3 and 4, then changed their attitudes toward disabilities (Category 5). Such changes in perspective prompted mothers to create a frontier for other mothers (Category 6). (Figure 1)

Figure 1

Category 1: Concern about abnormalities

This category consisted of one concept: *Concern about abnormalities*. In the early postnatal period, abnormalities are not seen from external appearances; therefore, few children are diagnosed in this period. However, many mothers were worried about behavioral problems such as feeding difficulties.

My daughter often seemed to strain... I didn't worry much about it and just thought she strained to defecate or something, but she gradually increased the frequency of

doing so. She didn't cry nor suckle, but I just thought that she was a quiet child and wondered what all her behaviour meant... (Participant D)

Category 2: A healthy child is considered as a standard

This category comprised nine subcategories: (i) Impact of diagnosis, (ii) Relief to know the cause, (iii) Determined and prepared, (iv) Preoccupation with the disorder, (v) Extreme confusion, (vi) Feeling alone in the world, (vii) Relying on support, (viii) Feeling a sense of belonging, and (ix) Confidence in parenting.

Impact of diagnosis

When the child was diagnosed with a chromosomal abnormality, mothers received a profound shock. When the child's abnormalities were life-threatening, such as congenital heart disease or respiratory disorder, mothers saw saving the child's life as the priority over chromosomal abnormalities, whether or not the cause was chromosomal.

Chromosome, that is what it is. I thought my daughter will be normal as when the hole in her heart was closed and she started to suckle well. When I heard that, I felt "Oh, what's going on. My daughter will not return to normal, nor get better..." (Participant O)

Relief to know the cause

The diagnosis of chromosomal abnormalities was a shock for the mothers but the medical assurance that the disorder was not caused by heredity or poor parenting gave them relief:

> *The only thing that saved me a little was that it was neither my husband's nor my fault, and we could think of it as something like a programing glitch in the process of having a child. It was a little salvation. (Participant B)*

Determined and prepared

Even though the mother was shocked at the child's disorder, they were able to grasp the situation when the cause was clarified and get out of the stalemate situation they had been in to act for their child:

> *When she was born, I blamed myself that I could not give my baby daughter her health but well, I can't do anything about it. If it is treatable, I'll get her treated, but if she can't be treated, well, I just have to raise her. (Participant I)*

Preoccupation with the disorder

Every chromosomal structural abnormality is often unprecedented, and the majority of mothers had anxiety about an unfathomable future. Due to their constant anxiety, mothers were troubled about minor incidents concerning the child and desperately

searched for information about the disorder. However, when they could not find the information they needed and fell into a state where they had no idea what to do:

First, I searched on the Internet, but I found almost no information in Japanese... Almost nothing - and I was in despair. (Participant B)

Extreme confusion

Mothers were preoccupied with the disorder and extremely confused about parenting. When the hospitalized child was discharged home, many mothers felt anxious about parenting and making decisions by themselves, the baby's fluctuating physical condition and facing ongoing crises. They struggled to breastfeed because they could not parent as they would for a healthy child, and they did not know how to deal with challenging behaviors such as sensitivity and irritability to noise and temperature.

My daughter was sleeping all the time, she didn't cry, she didn't laugh, she only slept. She didn't feed because she was sleeping, and she lost weight... (Participant I)

Feeling alone in the world

It was hard for mothers to find friends or neighbors with a child with a chromosomal structural abnormality, and mothers felt like they are alone in the world. Mothers were looking for sympathetic peers because being alone brought on negative thinking and despondency because they were a minority in society:

I couldn't get on the train... Yeah, it was hard for me. I found it hard to see healthy children around me. (Participant D)

Relying on support

Once they were able to get support for their child's medical treatment or rehabilitation, mothers relied on the support and committed themselves to see the health professionals with a sense of desperate hope:

For the time being, I wanted my daughter to be able to walk, and really did my best to go to physical therapy. (Participant D)

Feeling a sense of belonging

When they met another mother at treatment or rehabilitation service who was raising a child with similar disabilities, mothers realized that they were not the only ones and felt like they were in a safe space. When mothers met a family with a child with the same condition, they could sympathize with each other and needed no explanation about their experience and felt a deep sense of ease:

[About the families she met at the rehabilitation center] I was relieved because I felt they are fellows. I had felt like I've made comrades. (Participant O).

Confidence in parenting

Through professional support and interaction with their peers, the mothers began to understand their child's non-verbal communication and noticed growth. This gave them hope for the child's development, and a feeling of confidence in their own parenting:

For the first year after my son was born, I allowed the feeling to linger, but [my son] was growing well, which gave me the strength to go forward. (Participant K).

The mothers' way of thinking was to compare their child to a healthy one gradually changed to concentrate on their child's reaction and growth.

Category 3: Deepening attachment to the child

This category comprised two concepts: (i) Deepening attachment to the child, and (ii) Acceptance of an uncertain future.

Mothers who learned how to communicate with their child and became hopeful about their child's development and deepened their attachment to the child and began to accept the uncertainty of how the child would grow up, even if they still had insufficient information:

I couldn't accept my daughter (when she was born). But I dote on her now (thanks to) the doctors who have looked after her since she was one year old. The doctors were

so supportive of my daughter's growth by providing care, education, and rehabilitation, and that saved me a lot. (Participant B).

Category 4: Acceptance of the child as/he is

This category comprised four concepts: (i) Accepting the child as s/he is, (ii) Taking the initiative in choosing appropriate support, (iii) Family adjustment to prioritize the affected child, and (iv) Trial-and-error parenting. Mothers stopped considering the healthy development of children as a standard by which to judge their own child and deepened their attachment to the child. Mothers started to parent using a trial-and-error approach, accepting the child as it was. They rebuilt their view about their child through deepening their attachment to the child according to their development:

It is strange, but the load on my shoulders lifted, I used to think that I had to make my daughter do her best, but now I think she should not force herself, she is ok as she is. This is what I came to understand; it made sense to me. (Participant N).

Category 5: Changing attitude toward disabilities

This category comprised one concept: *Rethinking views about disability*. Mothers changed their views on disabilities in general through their child-rearing.

I used to think that people with disabilities were scary, just simply because I did not know what they were like. But through communicate with children with various disorders, and seeing adults (with disabilities) coming to sell bread, I started to wonder if my daughter will also become like this, yeah... I have changed (my awareness about people with disabilities). I think that they are normal, they just have many things that they cannot do. (Participant E)

Category 6: Creating a frontier for other mothers

This category comprised two concepts: (i) Establishing resources for new mothers, and (ii) Raising social awareness. Mothers who changed their view on their standard of parenting and disabilities wished to use their own experiences to help families in the same situation. They shared their experience on the Internet or established family groups to support members and advocate for future mothers and babies with the same disorder.

After I brought all the materials (about my child's disease) to the principal, his facial expression changed quickly, and he asked me to copy the materials, and he said he would have a meeting with all teachers. I felt the door was open because I have been appealing for a long time to be understood in some way. However, my

daughter looked as if she could do more things than she actually could. I suffered a lot for three years. (Participant M)

DISCUSSION

We focused on and clarified the process of mothers finding their unique way to bring up their children with little information. The findings, illustrate the meaning mothers ascribe to having a child with a rare chromosomal structural abnormality, how their understanding changed and their desire to connect with others with a similar experience.

1. Meaning of being diagnosed with rare chromosomal structural abnormalities

The diagnosis of rare chromosomal structural abnormalities in children impacted mothers in two ways – firstly that their child had a disorder but also the distress knowing that it was a lifelong and untreatable disorder. The term "chromosomal abnormalities" made it sound extremely serious. However, when a more precise diagnosis was provided, it became clear to the mothers that the child's symptoms were due to a chromosomal structural abnormality and was not their fault. Each participant had been told by the doctor that the disorder was caused by mutation and was not hereditary. These understandings gave them comfort. This accords with previous researchers who found that most parents were shocked, grieved, and then felt relief and

less guilty when a diagnosis of rare chromosomal abnormalities was made (Hallberg, Óskarsdóttir, & Klingberg, 2010; Houdayer et al., 2013; Waxler et al., 2013). The diagnosis helped mothers and family members understand the cause and avoid imagining other reasons, which may perpetuate their suffering (Lewis, Skirton, & Jones, 2010).

The guilt mothers experience is perhaps exacerbated by cultural factors including the traditional view in Japan that infertility and the birth of a child with disabilities were the woman's fault and this sometimes led to the termination of a marriage (Lebra, 1983). Genetic diseases have been considered to "contaminate the blood" in the family (Kato & Sleeboom-Faulkner, 2009), and many people still have a sense of "fear, pity, and aversion" against genetic diseases. (Hasegawa & Igarashi, 1993).

A diagnosis of rare chromosomal structural abnormalities enables mothers to search for a range of information using the name of the disorder as a keyword and also to access support from relevant agencies and the community. Information retrieval is essential in the process of empowerment and decision making (Alsem et al., 2017). They searched for clues as to how their child will develop for example, whether they would be able to speak or walk. In reality, however, the Internet did not provide sufficient information,

and mothers were extremely confused. Similarly, other researchers found that lack of information is a major stressor for mothers, (Griffith et al., 2011).

2. Change of mothers' view of abnormality

Initially mothers looked after their child with a sense of values and criteria based on "healthy children" when they were informed about their child's chromosomal structural abnormalities; however, they came to accept their child as it is. The results of this study are similar to the five stages proposed by Drotar (1975)—shock, denial, sadness and anger, adaptation, and reorganization, but the results of Drotar's study indicate that parents move back and forth between the stages, whereas the results of this study showed one-way process. This maybe because some mothers in our study were describing their adult children because our focus was on parenting over a longer time span. Mother's views of disabilities have been shaped by custom and the law. The Act on Elimination of Disability Discrimination was enacted in 2013, and inclusive education has been spreading (Mithout, 2016) which has led to a better understanding of people with disabilities and genetic diseases although these attitudes persist.

Some participants stated they had the impression before raising their children that people with disabilities were "scary." The negative image that mothers had against

people with disabilities can become a stigma against themselves when they discover their child has chromosomal structural abnormalities. Notably, mothers might suffer from dual stigma when their child has a “disorder” and chromosomal abnormalities related to “genetic information” (although not necessarily inherited) causing them to withdraw (Angermeyer, Link, & Majcher-Angermeyer, 1987). Some mothers, soon after their child was diagnosed, avoided interaction with others, or encountering healthy children.

The result indicated that "getting support" and "meeting peers" was the key for mothers to get out of difficult situations and to raise their children in their unique way, and this is similar to the existence of " the own" and "the wise." For mothers who consider themselves stigmatized individuals and hesitate to disclose themselves to people around them, it is surmised that their connection with peers and support had the meaning of finding a "place to stay and feel secure" as psychological safety. However, due to its rarity, there is very little chance to encounter peers, and some of the participants tried a number of strategies over a long time before they finally met with family members in the same situation. Also, only a few medical professionals are familiar with a particular disease of chromosomal structural abnormalities, and mothers could not obtain the necessary information and support. Connection with "support" and

"peers" is also essential to families of children with other conditions. However, mothers of children with chromosomal structural abnormalities find it extremely difficult to obtain these, which is the distinguishing characteristic of these families. Because chromosomal structural abnormalities are rare and individual cases are often unprecedented in the community, nurses must understand the needs of the families and children and help establish the necessary support system.

Besides, given that stigma is associated with low self-esteem, it is important to assist mothers in raising their self-esteem and confidence in raising their child (Shea & Tronick, 1988) In their daily nursing care, nurses are required to accept the mother's difficulties and provide support for mothers to be able to pay attention to small growth of their child and see hope for raising the child. Nurses must strive to understand each disease's characteristics and natural history and provide mothers with useful information (Kutsunugi et al, 2017). It is also essential for the nurses to treat the child with affection (Wei, Wei, Brown, Buck, & Mill, 2018) and show the mother how to treat them. Nurses should give positive feedback to mothers in their child-rearing practices by praising them and helping them have confidence and a sense of competence in raising the child (Komoto, Hirose, & Okamitsu, 2013). These support enhance mothers' self-esteem, and

thereby mothers positively perceive their child as well as their identity as a mother of the child with a chromosomal structural disorder.

3. The desire to connect

Through the experience of parenting, mothers learn to engage and communicate with society. Four of the participants created a connection with other families with the same disorder, and five mothers blogged about their status. This indicated their desire to connect with other family members and a strong desire to do something for other mothers in a similar situation. Furthermore, they wish themselves to be recognized and understood in society and disclosed their situation, which had been kept secret until then.

Encounters with peers can be a very beneficial experience for mothers in (1) shared social identity, (2) learning from the experiences of others, (3) personal growth, and (4) supporting others (Shilling et al., 2013). Interacting with other families and their children with the same abnormalities enables parents to create a future vision about their child, and this leads to the view that their child will be able to cope with the disabilities (Kerr & McIntosh, 2000). Mothers' experience of not meeting their peers nor receiving the support they wanted is considered to have motivated mothers to "Create a frontier

for other mothers" and disseminate information on their situation. Supporting others is as important as receiving support, and it proves the knowledge gained as a parent and increases self-worth (Lo, 2010).

Implementations for practice

This study suggests three critical points for nurses in their nursing practice. First, nurses must have genetic knowledge about chromosomal structural abnormalities and compile detailed information related to the specific disorders and daily life of the family and the child to provide mothers with useful information in their child-rearing. Secondly, it is crucial to fully understand that the mothers and the child are minorities in the society and that they may have self-stigma, and therefore to protect their dignity and provide support to raise their self-esteem. Lastly, with the low awareness in the society and the difficulty of adapting the existing support system, the families could not help but become pioneers. When they apply for public support, nurses need to play a role in advocating the situation of children and families so that they can guarantee the support they need. An example of this is "Unique," a website of a family group with children with rare chromosomal abnormalities (Unique), including chromosomal structural abnormalities, was established in the United Kingdom, where information about

chromosome abnormalities caused by each chromosome, and case studies of children with the disorder interaction among families has been enhanced through the website. It is an English language homepage, and it is not easy for general parents of non-English speaking countries to read it and participate in the group at this time, but it is a useful model for other countries such as Japan to accumulate various and useful information in line with the parents' needs.

LIMITATIONS

There is a large variety of chromosomal structural abnormalities, of which this study dealt with only several types of abnormalities, so there may be a bias in capturing the overall picture of chromosomal structural abnormalities. Also, because we recruited participants from those who have posted their status on the Web and their peers by snowball sampling, so all the mothers had some connection with other families, which might have influenced the results. Further research on father's perspectives is warranted as well as families which may not have connected with others in a similar situation.

CONCLUSIONS

This study investigated the process of mothers finding their way to raise their child with chromosomal structural abnormalities without enough information, and a total of 35 concepts, nine subcategories, and six categories were extracted, and their relationship was clarified. The key events that helped mothers get out of difficult situations and start to raise their children were connecting with support and meeting peers; however, these events were not easily occurred due to the rareness of the diseases.

Nurses need to give sufficient consideration to their possible self-stigma and provide support for mothers and children to enhance their self-esteem. Detailed information on specific rare diseases needs to be collected and compiled as useful information for concerned mothers. Nurses sometimes should be the voice and appeal to society on behalf of the families and the child who have difficulty in sending out their message.

Acknowledgments

We would like to express our sincere appreciation to the mothers who participated in the survey. We also would like to express our sincere gratitude to Professor Yasuhito Kinoshita of St. Luke's International University for his guidance and advice regarding

analysis and Ms. Junko Maeda for her advice on English writing. This work was supported by JSPS KAKENHI Grant Number 18K17554.

Disclosure statement

There is no conflict of interest in this research.

Author contributions

S.K. prepared the concept and design, collected data, performed the analysis and interpretation, prepared the manuscript, and supervised the entire process of the study. K.T. and K.M. contributed to the concept, design, data analysis, and interpretation. K.T., K.M, J.K., and T.E.S. contributed to prepare the manuscript. K.I. gave technical advice. T.G, Y.T., T.E.S. and Y.E. critically reviewed the manuscript, and all authors read and approved the final manuscript.

REFERENCES

Adams, D., Clarke, S., Griffith, G., Howlin, P., Moss, J., Petty, J., ... & Oliver, C. (2018). Mental health and well-being in mothers of children with rare genetic syndromes showing chronic challenging behavior: A cross-sectional and longitudinal study.

American Journal on Intellectual and Developmental Disabilities, 123(3), 241-253.

Alsem, M. W., van Meeteren, K. M., Verhoef, M., Schmitz, M. J. W. M., Jongmans, M. J., Meily-Visser, J. M. A., & Ketelaar, M. (2017). Co-creation of a digital tool for the empowerment of parents of children with physical disabilities. *Research Involvement and Engagement, 3*(1), 1-12. https://doi.org/10.1186/s40900-017-0079-6

Angermeyer, M. C., Link, B. G., & Majcher-Angermeyer, A. (1987). Stigma perceived by patients attending modern treatment settings: Some unanticipated effects of community psychiatry reforms. *Journal of Nervous and Mental Disease.175*(1), 4-11. https://doi.org/10.1097/00005053-198701000-00002

Bose, M., Mahadevan, M., Schules, D. R., Coleman, R. K., Gawron, K. M., Gamble, M. B., ... & Rizzo, W. B. (2019). Emotional experience in parents of children with Zellweger spectrum disorders: A qualitative study. *Molecular Genetics and Metabolism Reports, 19*, 100459. https://doi.org/10.1016/j.ymgmr.2019.100459

Bruns, D., & Foerster, K. (2011). 'We've been through it all together': Supports for parents with children with rare trisomy conditions. *Journal of Intellectual Disability Research, 55*(4), 361-369.

Cabinet Office Government of Japan. (2017). Results of the 2016 Basic Survey on Social Life; Time Related to Childcare and Housework for Men. (in Japanese) [Cited 10 Jul 2020.] Available from URL: http://wwwa.cao.go.jp/wlb/government/top/hyouka/k_42/pdf/s1-2.pdf

Creswell, J. W. (2003). *Research Design: Qualitative, quantitative, and mixed methods approaches*(2nd ed.). London: SAGE Publishing Inc.

Drotar, D., Baskiewicz, A., Irvin, N., Kennell, J., & Klaus, M. (1975). The adaptation of parents to the birth of an infant with a congenital malformation: A hypothetical model. *Pediatrics, 56*(5), 710-717.

Glaser, Barney G, & Strauss, Anselm L. (1967). *The discovery of grounded theory; strategies for qualitative research*. NewYork: Aldine Publishing Company.

Griffith, G. M., Hastings, R. P., Nash, S., Petalas, M., Oliver, C., Howlin, P., ... & Tunnicliffe, P. (2011). "You have to sit and explain it all, and explain yourself." Mothers' experiences of support services for their offspring with a rare genetic intellectual disability syndrome. *Journal of Genetic Counseling, 20*(2), 165-177.

Gundersen, T. (2011). 'One wants to know what a chromosome is': The Internet as a coping resource when adjusting to life parenting a child with a rare genetic disorder. *Sociology of Health and Illness, 33*(1), 81-95.

https://doi.org/10.1111/j.1467-9566.2010.01277.x

Hallberg, U., Óskarsdóttir, S., & Klingberg, G. (2010). 22q11 deletion syndrome–the meaning of a diagnosis. A qualitative study on parental perspectives. *Child: Care, Health and Development*, 36(5), 719-725. https://doi.org/10.1111/j.1365-2214.2010.01108.x

Hasegawa, T, & Igarashi, T. (1993). Popular Image of the Term "Genetic Disease." *Journal of Tokyo Women's Medical University, 63*, 94-101. (in Japanese)

Heilstedt, H. A., Ballif, B. C., Howard, L. A., Kashork, C. D., & Shaffer, L. G. (2003). Population data suggest that deletions of 1p36 are a relatively common chromosome abnormality. *Clinical Genetics, 64*(4), 310-316.

Houdayer, F., Gargiulo, M., Frischmann, M., Labalme, A., Decullier, E., Cordier, M. P., ... & Edery, P. (2013). The psychological impact of cryptic chromosomal abnormalities diagnosis announcement. *European Journal of Medical Genetics, 56*(11), 585-590. https://doi.org/10.1016/j.ejmg.2013.09.002

Kato, M, & Sleeboom-Faulkner, M. (2009). Cultures of marriage, reproduction, and genetic testing in Japan. *BioSocieties, 4*(2-3), 115-127.

Kepreotes, E. (2014). *A Critical Ethnography: The Parental Impact of Diagnosed Rare Diseases of Childhood.* (Unpublished doctoral dissertation) The University of

Newcastle, Newcastle, Australia.

Kerr, S. M., & McIntosh, J. B. (2000). Coping when a child has a disability: Exploring the impact of parent-to-parent support. *Child: Care, Health and Development, 26*(4), 309-322. https://doi.org/10.1046/j.1365-2214.2000.00149.x

King, G. A., Zwaigenbaum, L., King, S., Baxter, D., Rosenbaum, P., & Bates, A. (2006). A qualitative investigation of changes in the belief systems of families of children with autism or Down syndrome. *Child: care, health and development, 32*(3), 353-369. https://doi.org/10.1111/j.1365-2214.2006.00571.x

Kinoshita, Y. (2003). *Modified Grounded Theory Approach*. Tokyo: Kobundo. (in Japanese)

Komoto, K., Hirose, T., & Okamitsu, M. (2013). Nursing intervention in infant mental health: Enhancing mother-infant interaction and self-esteem of adolescent mothers. *Journal of Nursing & Care [Internet]*, 2167-1168. https://doi.org/10.4172/2167-1168.S5-006

Korkow-Moradi, H., Kim, H. J., & Springer, N. P. (2017). Common Factors Contributing to the Adjustment Process of Mothers of Children Diagnosed with Down Syndrome: A Qualitative Study. *Journal of Family Psychotherapy, 28*(3), 193-204. https://doi.org/10.1080/08975353.2017.1291238

Kutsunugi, S, Tsujino, K, Murakami, K, Iida, K, & Endo, Y. (2017). Nursing care for children with PWS and their parents during infancy. *Journal of Japanese Society of Genetic Nursing, 15*(2), 57-67. (in Japanese)

Larson, E. (1998). Reframing the meaning of disability to families: The embrace of paradox. *Social Science & Medicine, 47*(7), 865-875. https://doi.org/10.1016/S0277-9536(98)00113-0

Lewis, C., Skirton, H., & Jones, R. (2010). Living without a diagnosis: The parental experience. *Genetic Testing and Molecular Biomarkers, 14*(6), 807-815. https://doi.org/10.1089/gtmb.2010.0061

Lincoln, Y. S., & Guba, E. G. (1985). Establishing trustworthiness. *Naturalistic inquiry*, 289(331), 289-327.

Lo, L. (2010). Perceived benefits experienced in support groups for Chinese families of children with disabilities. *Early Child Development and Care, 180*(3), 405-415. https://doi.org/10.1080/03004430802002625

Mithout, A. L. (2016). Children with disabilities in the Japanese school system: A path toward social integration?. *Contemporary Japan, 28*(2), 165-184.

Nakamura, T. (2020). Current status and future aspects of home care medicine for children with medical complexity. *Organ Biology, 27*(1), 21-30. (in Japanese)

Nari, H. (2011). The life and rights of people with severe motor and intellectual disabilities: With a focus on those who had prolonged admissions. *Pediatric Nursing, 34* (5), 547-552. (in Japanese)

O'Connor, D. J., & Hemmings, R. J. (2014). Coping with small populations of patients in clinical trials. *Expert Opinion on Orphan Drugs, 2*(8), 765-768. https://doi.org/10.1517/21678707.2014.931221

Odachi, R., Tamaki, T., Ito, M., Okita, T., Kitamura, Y., & Sobue, T. (2017). Nurses' experiences of end-of-life care in long-term care hospitals in Japan: Balancing improving the quality of life and sustaining the lives of patients dying at hospitals. *Asian Nursing Research, 11*(3), 207-215. https://doi.org/10.1016/j.anr.2017.08.004

Reilly, C., Murtagh, L., & Senior, J. (2015). The impact on the family of four neurogenetic syndromes: A comparative study of parental views. *Journal of Genetic Counseling, 24*(5), 851-861. https://doi.org/10.1007/s10897-015-9820-1

Lebra, T. S. (1983). Shame and guilt: A psychocultural view of the Japanese self. *Ethos, 11*(3), 192-209.

Sato, Y. (2012). Epidemiology of 1p36 deletion syndrome. In: Yamamoto, T., *1p36 Deletion Syndrome Handbook* (pp.34-36). Tokyo, Japan: Shindan To Chiryo Sha.

(In Japanese)

Shea, E., & Tronick, E. Z. (1988). The maternal self-report inventory: A research and clinical instrument for assessing maternal self-esteem. *Theory and Research in Behavioral Pediatrics, 4*, 101-139.

Shilling, V., Morris, C., Thompson - Coon, J., Ukoumunne, O., Rogers, M., & Logan, S. (2013). Peer support for parents of children with chronic disabling conditions: A systematic review of quantitative and qualitative studies. *Developmental Medicine & Child Neurology, 55*(7), 602-609.

Strehle, E. M., & Middlemiss, P. (2007). Children with 4q-syndrome: the parents' perspective. *Genetic Counseling, 18*(2), 189-200.

Takeshita, H. (2019). Thoughts on and methods used in M-GTA. In: Ishiyama, N., Nakanishi, Y., Koyama, K., and Takeshita, H, *Mechanisms Of Cross-Boundary Learning: Communities of Practice and Job Crafting* (pp.60-89). Newcastle, UK: Cambridge Scholars Publishing.

Unique, Rare Chromosome Disorder Support Group. Understanding Rare Chromosome and Gene Disorders. (2020). [Cited 10 Jul 2020.] Available from URL: https://www.rarechromo.org/

Waxler, J. L., Cherniske, E. M., Dieter, K., Herd, P., & Pober, B. R. (2013). Hearing from

parents: the impact of receiving the diagnosis of Williams syndrome in their child. *American Journal of Medical Genetics. Part A, 161A*(3), 534-541. https://doi.org/10.1002/ajmg.a.35789

Wei, H., Wei, T., Brown, K. J., Buck, S. H., & Mill, M. R. (2018). Parents' perceptions of caring characteristics of physicians and nurses. *International Journal for Human Caring, 22*(1), 49-55. https://doi.org/10.20467/1091-5710.22.1.pg47

Weng, H. J., Niu, D. M., Turale, S., Tsao, L. I., Shih, F. J., Yamamoto - Mitani, N., ... & Shih, F. J. (2012). Family caregiver distress with children having rare genetic disorders: A qualitative study involving Russell–Silver Syndrome in Taiwan. *Journal of Clinical Nursing, 21*(1-2), 160-169. https://doi.org/10.1111/j.1365-2702.2010.03583.x

Table 1 Demographic profiles of mothers and their children

	Number(n=16)	%
Age of mothers		
≦29 years	1	6.3
30-39 years	6	37.5
40-49 years	4	25.0
50-59 years	5	31.3
Age of children		
2-6 years	6	37.5
7-12 years	3	18.8
13-18 years	3	18.8
19-31 years	4	25.0
Gender of children		
Male	4	25.0
Female	12	75.0
Child's age when diagnosed		
2 weeks to 2 months	8	50.0
3 months to 1 year	1	6.3
1-4 years	6	37.5
5-8 years	1	6.3
Type of chromosomal abnormalities		
1p36 deletion syndrome	5	31.3
22q11.2 deletion syndrome	3	18.8
11q deletion syndrome (Jacobsen syndrome)	3	18.8
2q duplication syndorme	2	12.5
1q duplication syndrome	1	6.3
8p duplication syndrome	1	6.3
disease caused by chromosome 16 abnormalities	1	6.3
Symptoms		
Congenital heart disease	12	75.0
Epilepsy	6	37.5
Difficulty in walking	3	18.8
Respiratory disorder (Using ventilator)	1	6.3
Oral feeding difficulties (Tube feeding)	1	6.3
Social situation		
Stay home	5	31.3
Special school	4	25.0
Ordinary school (Special support class)	3	18.8
Kindergarten	2	12.5
Employee as a person with disablities	2	12.5
Siblings		
Having sibling(s)	13	81.3
Only child	3	18.8

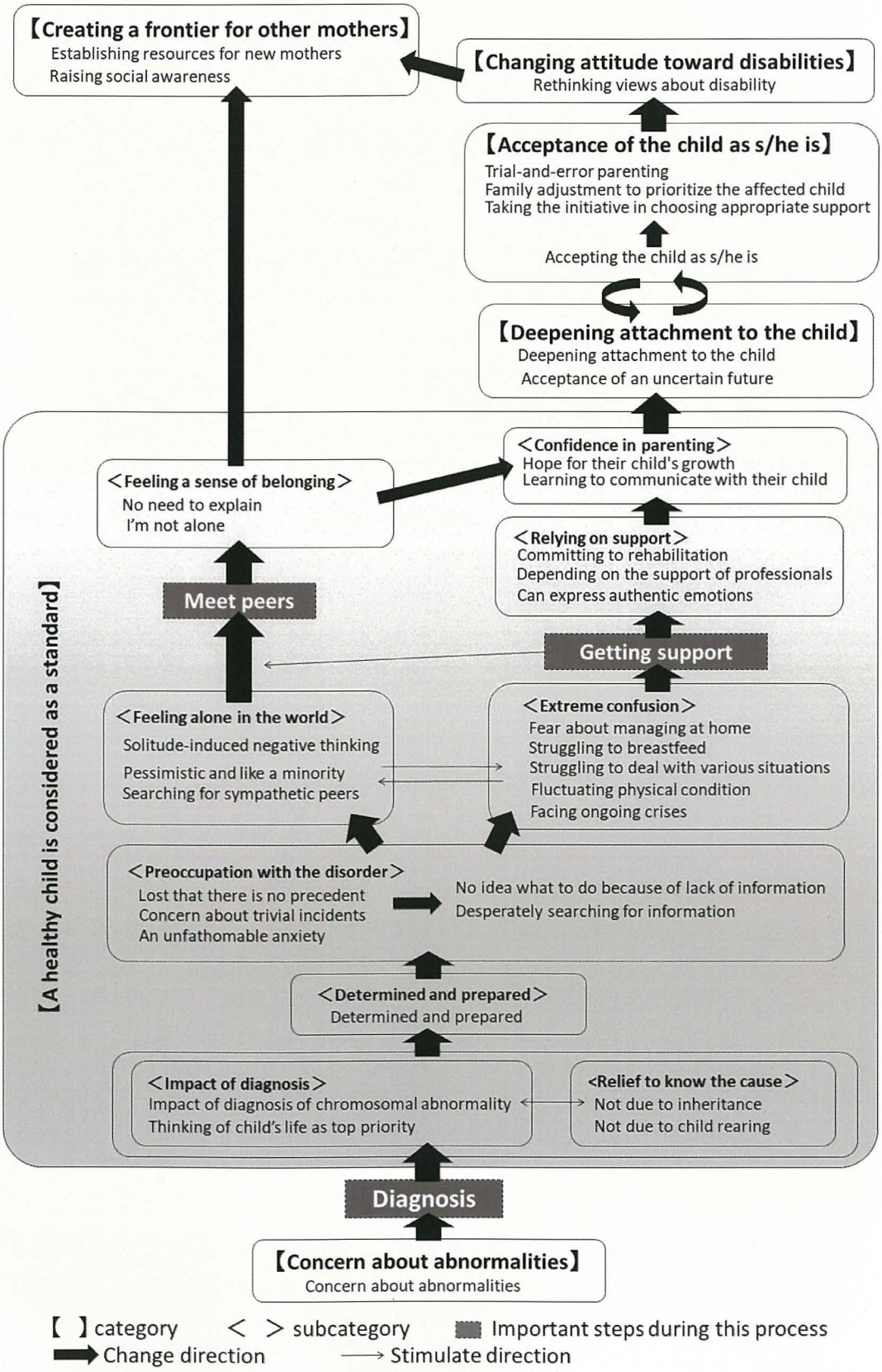

Figure 1. The process of mothers finding their way to raise their child with a chromosomal structural abnormality only with little information