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Statistical Compilation

Welfare System Difficulties and Desired Support for Individuals with 22q11.2 Deletion Syndromes and Their Families: Analysis of Web Survey Results by Mixed Method Research

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Abstract

The 22q11.2 deletion syndrome (22q11DS) is a chromosomal disorder designated as an incurable disease that can be complicated by physical, intellectual, and psychiatric disorders. This syndrome is becoming especially well known among Japanese psychiatrists because it may cause schizophrenia-like psychotic symptoms. Most previous studies in the field have been limited to case reports focusing on diagnosis and treatment. Although guidelines do exist from the perspective of lifelong medical care for congenital heart disease, the psychosocial difficulties of individuals and their families

have not been fully understood. The syndrome is highly heterogeneous, and the phenotype within individuals may change with age, including congenital physical disease, intellectual developmental disorder affecting learning during the school years, and onset of mental disorders after puberty. The combination of multimorbidity characteristics and changes to the living environment during life stages creates multilayered difficulties. However, routine support in divided social systems is often inadequate. While focusing on the welfare system, the present study aims to clarify difficulties and identify necessary support required due to the mismatch between disease traits and social systems. The method used was a web-based survey of 125 caregivers. A mixed research method was adopted to conduct quantitative analysis of the selected responses and qualitative analysis of the open-ended responses. The quantitative analysis showed that the diversity of difficulties changed with age: younger individuals tended to have difficulties related to medical care and education, and older individuals tended to have difficulties related to employment, marriage, and housing. The lack of consideration for overlapping disabilities decreased from early childhood to school age but increased again at age 19 and above. In the qualitative analysis, several themes were identified that were not included in the quantitative analysis, such as the psychological aspects of the individual and family and the specific support required. In both quantitative and qualitative analyses, lack of knowledge and understanding of the disease by professionals was a point raised by many respondents. Evidently, a need exists for flexible support that is not bound by existing institutional design and is based on understanding of the disease and the diverse difficulties and needs associated with it.

Keywords: 22q11.2 deletion syndrome, multimorbidity, welfare system, quantitative analysis, qualitative analysis

Introduction

22q11.2 deletion syndrome (22q11DS) is a chromosomal abnormality caused by a deletion in the 11.2 region of the long arm of chromosome 22. In addition to physical disorders, such as congenital heart disease, palatal malformation, and immunodeficiency, patients may

present with intellectual developmental disorder and multiple mental disorders, such as schizophrenia-like psychotic symptoms, anxiety disorder, and neurodevelopmental disorders. There is marked individual phenotypic variation.¹⁹⁾ The prevalence of this syndrome is estimated to be 1 in 2,000

to 6,000 births,²⁰⁾ making it the most common microdeletion syndrome. It is also the second most common chromosomal syndrome causing congenital heart disease after Down syndrome.¹⁹⁾ In 1994, the fluorescence in situ hybridization (FISH) method used to diagnose this syndrome became covered by health insurance, and in 2015, it was designated as an intractable disease by the Japanese government. When physical symptoms are not prominent, the syndrome may not be diagnosed in childhood, but diagnosed later in adulthood when mental symptoms appear. However, considering the prevalence, it is estimated that many cases remain undiagnosed. Approximately one in three individuals with 22q11DS exhibit schizophrenia-like psychiatric symptoms,¹¹⁾ which has increased interest among psychiatrists in Japan. However, most previous studies in the psychiatric field were limited to case reports on diagnosis and treatment.

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In 22q11DS, physical, intellectual, and psychiatric disabilities overlap with varying degrees of severity, ranging from mild to severe, depending on the individual. As a result, the difficulties faced by patients and their families in their daily lives are diverse, and existing standard support measures are often insufficient.³²⁾ While guidelines

exist for medical care related to physical conditions and intractable diseases in 22q11DS,⁵⁾⁸⁾¹⁴⁾²⁸⁾ the psychological and social challenges that individuals and their families encounter in daily life remain poorly understood. In 2017, the Department of Neuropsychiatry, University of Tokyo Hospital, established a mental health outpatient clinic specializing in 22q11.2 deletion syndrome (hereinafter referred to as the “specialized outpatient clinic”), and in 2019, conducted a web-based questionnaire survey targeting caregivers. Furthermore, in 2021, guidance for integrated support was created.³⁵⁾ This study focused on the results of the above-mentioned questionnaire survey related to the welfare system and involved quantitative and qualitative analyses using a mixed research method. The aim was to clarify the difficulties and needs arising from mismatches between disease characteristics and the welfare system.

I. Methods

1. Subjects

The study targeted 125 primary caregivers (mothers or fathers) of individuals with 22q11DS who responded to a web-based questionnaire survey conducted between March 20 and November 8, 2019. The survey was publicized via email and letters with the

cooperation of a family association for 22q11DS (22 HEART CLUB),³⁷⁾ The Japan Association for Children with Heart Disorders,⁴⁰⁾ and other parties involved in medical care for those with 22q11DS. Due to these circumstances, the overall picture of the target population was unclear, and it was not possible to calculate the response rate.

2. Ethical considerations

This study was conducted with the approval of the Ethics Committee of the University of Tokyo Faculty of Medicine [approval number: 2018015NI-(11)]. In accordance with the approval, prior to the start of the questionnaire survey, participants were provided with an explanation of the study, including its purpose, methods, voluntary nature, protection of personal information, and whether compensation would be provided. Participants were asked to read the explanation and respond to the questions, thereby indicating their consent to participate in the study. Participants were informed in advance that they would receive a prepaid card worth 5,000 yen as compensation.

3. Questions and response methods

The questionnaire was created based on previous literature⁵⁾⁷⁾⁸⁾¹²⁾ and the opinions of two caregivers of individuals with 22q11DS. The questionnaire consisted of 10 areas and was divided

into Part A, which was mandatory, and Part B, which was optional. Part A included: 1) basic information about the caregivers, 2) basic information about the persons with 22q11DS, and 3) questions about the caregivers' health and difficulties. Part B included: 4) questions about medical care, 5) the relationship with social welfare services, 6) education, 7) issues arising during the transition from child- to adulthood, 8) siblings, 9) research on 22q11DS, and 10) the impact on caregivers' lives. The web-based questionnaire survey was conducted anonymously, and the questionnaire was posted on the website (22q-pedia).²⁶⁾ For participants who found it difficult to respond online, paper questionnaires were mailed upon request. Of the 125 valid respondents, four responded using paper questionnaires. For details of the web-based questionnaire survey, please refer to our previous study.²⁵⁾³³⁾

In the present study, we used the following items from the above survey: basic information about people with 22q11DS and their caregivers (Part A, 1 and 2), multiple-choice answers about difficulties related to welfare and daily life (Part A, 3), and free-response answers about difficulties and needs using the welfare system (Part B, 5).

4. Research methods and data analysis

First, descriptive statistics were generated using basic information on the respondents (caregivers) and persons with 22q11DS (care recipients) by age group of people with 22q11DS (up to 5 years old, 6 to 11 years old, 12 to 18 years old, and 19 years old or older).

We then conducted quantitative and qualitative analyses and used a mixed research method with convergent design¹⁷⁾ to interpret the results of both analyses in an integrated manner.

In the quantitative analysis, we asked the following question: "During the past year, while supporting a family member with 22q11.2 deletion syndrome, what difficulties have you encountered in welfare and daily life? Please select all that apply. If there are some that do not fit into any of the categories, please write them in the 'Other' section" (125 responses). The selection rates for each item were calculated for each age group of persons with 22q11DS. Additionally, Fisher's exact test was conducted to examine whether there were significant differences in selection rates across the four age groups. The significance level was set at $p = 0.05$. Statistical analysis was performed using IBM SPSS Statistics, version 28.0 (IBM SPSS, Armonk, NY, USA).

As qualitative analysis, we performed theme analysis⁶⁾ of the free-response answers to the following: "Please describe any difficulties you have

encountered in using welfare systems, such as disability pensions, disability certificates, and intractable disease certification due to multiple diseases or disabilities" (33 responses); "What kind of support do you think is necessary for persons with 22q11.2 deletion syndrome and their parents in welfare services?" (55 responses). The content and meaning of the responses were coded, and based on this, they were classified into several categories and named. To ensure reliability, 25% of all responses were randomly selected, and two experts, a psychiatrist specializing in the outpatient clinic and clinical psychologist (Uno and Tanaka), independently coded them. After confirming a consistency rate of 90% or higher, the remaining responses were coded by the lead author, and all categories were reviewed by the researchers. Disagreements were discussed and consensus was reached. Finally, coding, category classification, and naming were finalized through consensus among multiple experienced psychiatrists, psychologists, and mental health counselors involved in the outpatient clinic. Microsoft Excel was used for managing qualitative data.

II. Results

1. Basic information on respondents (caregivers) and persons with 22q11DS (care recipients)

A total of 125 respondents participated, approximately 90% of whom were mothers and the remainder fathers. Individuals with 22q11DS were divided into four age groups: 0–5 years, 6–11 years, 12–18 years, and 19 years or older, with approximately 30 individuals in each age group. The male-to-female ratio of persons with 22q11DS was almost 1:1. Regarding the welfare systems used, 41.6% of respondents received child support allowance, special child support allowance, and welfare allowance for patients with intractable diseases. Regarding acquisition of a disability certificate or handbook, 48.8% had medical rehabilitation handbooks, 29.6% had physical disability certificates, and only 4.8% had mental disability certificates. Forty (40.8%) of the 98 people under the age of 18 who were eligible for the medical care support system for specific pediatric chronic diseases were using the program, and the percentage of people using the medical care support system for intractable diseases increased with age (Table 1).

All persons with 22q11DS had some form of physical disorder, with congenital heart disease (86.4% of the total) and ear, nose, throat, facial, and oral disorders (76.0% of total) being particularly common. The rates of intellectual developmental disorder and

neurodevelopmental disorders (autism spectrum disorder, attention deficit hyperactivity disorder, and learning disorder) increased after the age of 6, when children enter school, and the percentage of schizophrenia increased after the age of 19 (Table 2).

Next, we examined the multimorbidity of physical disorders, intellectual developmental disorder, and mental disorders. In the age group up to 5 years old, physical disorders alone accounted for 60% of cases, but in the age group 6 to 11 years old or older, the majority of cases involved multimorbidity in the physical, intellectual, and psychiatric domains, or multimorbidity in the physical and intellectual domains (Table 3).

2. Quantitative analysis

The selection rates for each item in response to the multiple-choice question: “During the past year, while supporting a family member with 22q11.2 deletion syndrome, what difficulties have you encountered in welfare and daily life? Please select all that apply. If there are some that do not fit into any of the categories, please write them in the ‘Other’ section,” shown by age group (Table 4). The top three items overall were “# 11 Lack of knowledge about 22q11.2 deletion syndrome among supporters such as welfare facility staff or government

officials” (48.0%), “#6 Lack of information about developmental support such as medical rehabilitation” (44.0%), and “#4 Although individuals may have multiple disabilities, the system is divided into physical, intellectual, and psychiatric disabilities, and the assessment does not take into account the difficulties of multimorbidity when applying for support” (34.4%).

Regarding the relationship between the selection rate of each question item and age, the most frequently selected item overall, “#11 Lack of knowledge about 22q11.2 deletion syndrome among supporters such as welfare facility staff or government officials,” was a common difficulty regardless of age group. Questions that showed differences by age group included “#8 Insufficient support for developmental issues such as medical rehabilitation” and “#6 Lack of information about developmental support such as medical rehabilitation,” which were more frequently cited by younger age groups, while employment-related issues such as “#16 Lack of information about employment,” “#18 Lack of employment opportunities,” “#17 Insufficient counselors or offices for employment support,” and “#19 Inability to maintain employment,” “#23 Anxiety about marriage” related to marriage, and “#22 Lack of housing such as group homes” related to housing

were frequently cited among the older age group, reflecting changes in difficulties according to life stage. “#20 Lack of understanding at the workplace” showed a bimodal distribution between the ages of 6 to 11 and 19 or above. Although details were not collected, it is possible that these responses refer to the workplaces of caregivers and persons with disabilities themselves. Although the selection rate for “#4 Although individuals may have multiple disabilities, the system is divided into physical, intellectual, and psychiatric disabilities, and the assessment does not take into account the difficulties of multimorbidity when applying for support” declined temporarily between the ages of 5 and 6 to 11, it increased again between the ages of 12 to 18 and 19 or above.

3. Qualitative analysis

In a thematic analysis of responses to an open-ended question: “Please describe any difficulties you have encountered in using welfare systems such as disability pensions, disability certificates, and intractable disease certification due to multiple diseases or disabilities,” three categories were identified: lack of information provision, difficulties in using the system, and psychological resistance (Table 5). Lack of information included: “difficulty understanding the concept of the

disease” and “difficulty obtaining information about the disease.” Difficulties in using the system included “insufficient, inconvenient, and unclear system,” “vertical divisions preventing comprehensive response,” “severity of multimorbidity not being assessed,” and “lack of knowledge among supporters.” Psychological resistance included “resistance to using the system as a person with a disability.”

Thematic analysis of responses to an open-ended question: “What kind of support do you think is necessary for persons with 22q11.2 deletion syndrome and their parents in welfare services?” revealed four categories: information provision, comprehensive support, supporters with specialized knowledge, and improvement of systems (Table 6). Information provision included: “information about the disease” and “public understanding of the disease,” while comprehensive support included: “comprehensive consultation services.” Supporters with specialized knowledge included: “supporters who think together with the person with the disease and their parents” and “supporters who act as intermediaries between the family and relevant organizations.” Improvement of systems included: “support that considers the characteristics of the person with the disease” “expansion of existing support,” “support that considers multimorbidity,”

“correction of disparities based on the place of residence,” “support after parents become elderly,” “support for parents,” and “support that encourages a positive outlook.”

III. Discussion

In this study, we clarified the difficulties and needs of people with 22q11DS and their families regarding the welfare system by combining quantitative analysis of multiple-choice answers and qualitative analysis of free-response answers in a web-based questionnaire survey.

The results of the quantitative analysis suggested that the younger the age, the higher the percentage of responses related to medical rehabilitation, and the older the age, the higher the percentage of responses related to employment, marriage, and housing, indicating that difficulties change with age. In addition to congenital physical disorders and intellectual developmental disorder, 22q11DS is often accompanied by mental disorders such as schizophrenia after puberty. From the perspective of families, mental health issues are perceived as more difficult to deal with than physical problems, and there are reports that information is needed on early signs of mental health symptoms and how to explain them to the person concerned.²⁾ In addition, the overall IQ

reportedly declines by an average of 7 points between the ages of 8 and 24,³⁹⁾ and the phenotype of the disorder changes with age even within the same individual. For example, in infancy, attention is focused on medical intervention for congenital physical disorders and the development of motor and language skills, while at school age, the focus shifts to academic performance and friendships, and during adolescence and beyond, the focus shifts to independence, behaviors associated with mental disorders, and concerns about recurrence.¹⁹⁾ In this way, central issues shift while being influenced by changes in the living environment in line with life stages. Existing institutional designs, where it is difficult for a single support provider to continue involvement, are insufficient for supporting issues that change with age. In particular, the transition from pediatrics to various medical specialties and psychiatry is a major challenge during the transition from child- to adulthood. Within the vertically divided system, families bear a heavy burden of coordinating with new support providers.¹⁵⁾ Caregivers of people with schizophrenia, a typical mental disorder, reportedly feel a heavier burden of caregiving than caregivers of people with physical disorders such as neurological disorders, despite the relatively smaller physical

and financial burden, due to the additional impact of social intolerance, such as stigma.⁴⁾ In 22q11DS, which is characterized by multimorbidity, it is not difficult to imagine that the burden on caregivers is even heavier than when these conditions occur alone, as it combines physical and psychiatric disorders.

The difficulties not taken into account in multimorbidity decreased temporarily from early childhood to school age, but increased again at age 19 and above. In 22q11DS, physical, intellectual, and mental disabilities overlap with varying degrees of severity from mild to severe, making it difficult to apply a system designed for a single disease. For example, a child with 22q11DS may not be able to keep up physically under the framework for intellectual disability support due to a heart condition, or conversely, may be denied admission to a special needs school for children with physical disabilities due to their intellectual disability. Supporters need to be aware that the standard support provided under the existing framework may not always meet the needs of people with multimorbidity.¹⁶⁾ The Ministry of Education, Culture, Sports, Science and Technology's Special Needs Education Curriculum Guidelines, revised in 2017 and 2019, include measures that take multimorbidity into consideration, such

as changing: “Special Provisions for Persons with Severe Multiple Disabilities” to “Handling of Educational Programs for Persons with Multiple Disabilities.”²³⁾²⁴⁾ However, in reality, much depends on the response of schools and teachers in practice. Although 22q11DS was designated as an intractable disease by the national government in 2015, the severity of heart disease (New York Heart Association functional classification II or higher) is a requirement for medical expense support, and difficulties associated with other complications are not sufficiently taken into consideration. Furthermore, the existence of the “age 18 barrier,” a systemic issue where services for children become inaccessible at the end of March when aged 18, leading to disconnection from previous support networks and providers, is well-known,¹⁶⁾ which may be one of the reasons for the increase in the selection rate among those aged 19 or older in this study.

Both quantitative and qualitative analysis revealed that a lack of understanding and knowledge among supporters was an important point reported by many respondents. This is one of the main themes in qualitative research on 22q11DS, and many parents felt that they had to fight for the care and support their children should receive.³⁸⁾ Additionally, due to the poor

public awareness of 22q11DS, it has been pointed out that explaining the disease itself poses significant challenges.¹⁵⁾ Compared with the difficulties and needs faced by caregivers of people with Down syndrome, a chromosomal syndrome, the difficulties associated with the low-level awareness of the disease are unique to 22q11DS.¹³⁾ Some reports stated that: “the lack of knowledge among specialists was perceived as a personal insult,”¹⁰⁾ and many families who were told by doctors that “the future is uncertain due to individual differences” felt that “not knowing = being rejected.”²¹⁾ Thus, it is necessary for supporters to remember that persons with 22q11DS and their families may have felt disappointed and been forced to distrust supporters around them. Furthermore, it is necessary to actively seek to expand knowledge in areas outside one's field of expertise while learning from conversations with individuals and families.

Thematic analysis as a form of qualitative analysis revealed several topics that were not included in the quantitative analysis questions, such as psychological aspects of individuals and their families (“psychological resistance to using the system” and “support that encourages a positive outlook”) and specific support needs (“comprehensive

consultation services,” “supporters who act as intermediaries between the family and relevant organizations,” “correction of disparities based on place of residence,” “support after parents become elderly,” and “support for parents”). To address these challenges, it is desirable for multiple professionals and organizations to collaborate and provide flexible support that is not bound by existing institutional frameworks. It is clear that a deeper understanding of 22q11DS and the diverse challenges and needs associated with it by each and every support provider is the foundation of effective support.

One limitation of this study was that the questionnaire was developed independently and is not widely used. However, as our department is currently the only outpatient clinic specializing in 22q11DS in Japan, this study can be considered pioneering. The participation of caregivers of people with 22q11DS in the questionnaire development process is also significant from the perspective of co-productive academic research. In future surveys, it will be desirable to revise the questionnaire to better reflect the diverse themes identified in this study's thematic analysis, thereby making it more responsive to the actual experiences of individuals and their families.

Conclusion

This study used a mixed research method combining quantitative and qualitative analysis to clarify the difficulties and needs experienced by persons with 22q11DS and their families in relation to the welfare system. The background to this was suggested to be a combination of factors, including the characteristics of the disease, which can lead to changes in the phenotype with age, changes in the living environment according to life stage, and mismatches with the system. To address the difficulties and needs of individuals with 22q11DS and their families, it is not sufficient to focus solely on medical aspects. Support that is not limited to existing systems is required, based on an understanding of the disorder and diverse difficulties and needs that accompany it.

Conflict of interest

Hidetaka Tamune: Received research funding from the Kanae Medical Promotion Foundation.

The other authors have no conflicts of interest to disclose in relation to this paper.

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Table 1: Basic information on respondents (caregivers) and individuals with 22q11DS (care recipients)

		Up to 5 years old (N=36)		6–11 years old (N=34)		12–18 years old (N=28)		19 years old or older (N=27)		Total (N=125)	
		N (mean)	% (SD)	N (mean)	% (SD)	N (mean)	% (SD)	N (mean)	% (SD)	N (mean)	% (SD)
Respondents (caregivers)	Age	38	3.9	40	5.2	47	4.3	53.4	4.8	43.8	7.5
	Relationship										
	Mother	32	88.9	31	91.2	24	85.7	27	100.0	114	91.2
	Father	4	11.1	3	8.8	4	14.3	0	0.0	11	8.8
Persons with 22q11DS (care recipients)	Age	2.9	1.5	8.5	1.8	15.0	2.1	22.4	4.4	11.3	7.7
	Sex										
	Male	20	55.6	16	47.1	14	50.0	13	48.1	63	50.4
	Female	16	44.4	18	52.9	14	50.0	14	51.9	62	49.6
	Currently enrolled in school	0	0.0	34	100.0	25	89.3	8	29.6	67	53.6
Household income	0-5.99 million yen	17	47.2	14	41.2	10	35.7	8	29.6	49	39.2
	600-11.99 million yen	13	36.1	13	38.2	13	46.4	14	51.9	53	42.4
	12 million yen or more	6	16.7	5	14.7	5	17.9	4	14.8	20	16.0
	Unknown	0	0.0	2	5.9	0	0.0	1	3.7	3	2.4
Welfare systems used	Physical disability certificates	10	27.8	8	23.5	9	26.5	10	37.0	37	24.3
	Medical rehabilitation handbook	8	22.2	18	52.9	19	55.9	16	59.3	61	48.8
	Mental disability certificates	0	0.0	1	2.9	2	5.9	3	11.1	6	4.8
	Medical care support system for intractable diseases	3	8.3	2	5.9	3	10.7	11	40.7	19	15.2
	Medical care support system for specific pediatric chronic diseases	17	47.2	14	41.2	9	32.1	0	0.0	40	32.0
	Disability pension	2	5.6	0	0.0	0	0.0	7	25.9	9	7.2
	Pension other than disability pension (including those of household members)	0	0.0	2	5.9	0	0.0	4	14.8	6	4.8
	Unemployment insurance	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0
	Child support allowance, special child support allowance, welfare allowance for patients with intractable diseases	16	44.4	18	52.9	12	42.9	6	22.2	52	41.6
	Public assistance	1	2.8	0	0.0	1	3.6	0	0.0	2	1.6
	Other	1	2.8	1	2.9	2	7.1	0	0.0	4	3.2

For each figure, the ages of the respondent (caregiver) and person with 22q11DS (care recipient) are shown as the average and standard deviation respectively, and other items are shown as the number of responses (N) and percentage of the total. SD: standard deviation

Table 1: Basic information on respondents (caregivers) and individuals with 22q11DS (care recipients)

Table 2: Complications in persons with 22q11DS

	Up to 5 years old (N=36)	6–11 years old (N=34)	12–18 years old (N=28)	19 years old or older (N=27)	Total (N=125)
	%	%	%	%	%
Physical disorders	100	100	100	100	100
Congenital heart disease	91.7	82.4	85.7	85.2	86.4
Immune system and allergic disorders	22.2	44.1	46.4	59.3	41.6
Endocrine disorders	30.6	38.2	46.4	48.1	40.0
Gastrointestinal disorders	47.2	32.4	46.4	48.1	43.2
Ear, nose, throat, face and oral disorders	63.9	85.3	71.4	85.2	76.0
Orthopedic disorders	38.9	35.3	53.6	51.9	44.0
Other* physical disorders	19.4	26.5	10.7	22.2	20.0
Intellectual developmental disorder	27.8	79.4	75.0	81.5	64.0
Mental disorders	16.7	32.4	32.1	77.8	37.6
Schizophrenia	0.0	0.0	3.6	25.9	6.4
Depression	0.0	0.0	0.0	3.7	0.8
Bipolar disorder	0.0	0.0	0.0	7.4	1.6
Anxiety disorder	0.0	0.0	17.9	33.3	11.2
Panic disorder	0.0	0.0	0.0	7.4	1.6
Obsessive-compulsive disorder	0.0	0.0	3.6	11.1	3.2
Epilepsy	8.3	17.6	14.3	25.9	16.0
Autism spectrum disorder	0.0	8.8	17.9	14.8	9.6
Attention deficit hyperactivity disorder	0.0	11.8	10.7	7.4	7.2
Oppositional defiant disorder	0.0	2.9	0.0	0.0	0.8
Learning disorder	2.8	26.5	14.3	29.6	17.6
Other* mental and neurological disorders	13.9	14.7	3.6	14.8	12.0

* "Other" responses were examined and classified under other applicable items as deemed appropriate.

Table 2: Complications in persons with 22q11DS

Table 3: Multimorbidity of physical disorders, intellectual developmental disorder, and mental disorders in persons with 22q11DS

	Up to 5 years old (N=36) %	6–11 years old (N=34) %	12–18 years old (N=28) %	19 years old or older (N=27) %	Total (N=125) %
PD+IDD+MD	8.3	58.8	53.6	77.8	47.2
PD+IDD	19.4	41.1	42.9	18.5	30.4
PD+MD	8.3	0.0	0.0	3.7	3.2
IDD+MD	0.0	0.0	0.0	0.0	0.0
PD	63.9	0.0	3.6	0.0	19.2
IDD	0.0	0.0	0.0	0.0	0.0
MD	0.0	0.0	0.0	0.0	0.0
None	0.0	0.0	0.0	0.0	0.0

PD: Physical disorders, IDD: Intellectual developmental disorder,
MD: Mental disorders

Table 3: Multimorbidity of physical disorders, intellectual developmental disorder, and mental disorders in persons with 22q11DS

Table 4: Relationship between the selection rate of each question item and age in response to the question, “During the past year, while supporting a family member with 22q11.2 deletion syndrome, what difficulties have you encountered in welfare and daily life?”

	Total (N=125) %	Up to 5 years old (N=36) %	6–11 years old (N=34) %	12–18 years old (N=28) %	19 years old or older (N=27) %	Fisher's exact test (p)	
#11 Lack of knowledge about 22q11.2 deletion syndrome among caregivers such as welfare facility staff or government officials	48.0	47.1	37.1	41.4	68.0	0.066	
#6 Lack of information about developmental support such as medical treatment	44.0	58.8	60.0	24.1	24.0	0.003	
#4 Although individuals may have multiple disabilities, the system is divided into physical, intellectual, and psychiatric disabilities, and the assessment does not take into account the difficulties of multimorbidity when applying for support	34.4	32.4	20.0	27.6	60.0	0.007	
#9 Lack of information about welfare services	32.8	38.2	25.7	24.1	44.0	0.308	
#1 Lack of information about financial support systems	31.2	44.1	25.7	17.2	40.0	0.226	
#7 Insufficient counselors or offices for developmental support	28.8	26.5	42.9	20.7	20.0	0.282	
#8 Insufficient support for developmental issues such as medical rehabilitation	27.2	32.4	42.9	20.7	4.0	0.017	
#12 No daycare center suited to the individual's characteristics or needs	24.8	20.6	20.0	20.7	36.0	0.223	
#3 Insufficient financial support systems	23.2	23.5	28.6	24.1	16.0	0.530	
#16 Lack of information about employment	23.2	2.9	20.0	34.5	40.0	<0.001	
#23 Anxiety about marriage	22.4	11.8	25.7	10.3	40.0	0.014	
#25 The person with 22q11.2 deletion syndrome is unable to obtain life insurance	20.8	23.5	20.0	10.3	32.0	0.121	
#2 Insufficient counselors or offices for financial support	20.0	26.5	22.9	10.3	20.0	0.765	
#10 Insufficient counselors or offices for welfare services	20.0	23.5	17.1	13.8	28.0	0.691	
#15 No support available when parents are exhausted and need rest	20.0	26.5	22.9	10.3	16.0	0.444	
#5 Unable to receive certification for intractable diseases	17.6	14.7	14.3	10.3	32.0	0.132	
#24 Anxiety about pregnancy and childbirth	14.4	8.8	20.0	6.9	20.0	0.551	
#17 Insufficient counselors or offices for employment support	12.8	0.0	14.3	17.2	20.0	0.008	
#22 Lack of housing such as group homes	11.2	0.0	2.9	17.2	28.0	<0.001	
#14 Lack of in-home or home-visit services	10.4	2.9	8.6	10.3	20.0	0.080	
#21 Lack of support for daily life	10.4	2.9	14.3	6.9	20.0	0.200	
#13 Even if a daycare center is available, the individual is unable to attend	9.6	8.8	11.4	3.4	12.0	0.720	
#26 Other	7.2	2.9	5.7	20.7	16.0	0.088	
#18 Lack of employment opportunities	6.4	0.0	0.0	3.4	24.0	<0.001	
#20 Lack of understanding at the workplace	4.8	0.0	5.7	0.0	8.0	0.027	
#19 Inability to maintain employment	2.4	0.0	0.0	0.0	12.0	0.015	

The questions are listed in order of the highest selection rate. Significant p values are indicated in bold.

Table 4: Relationship between the selection rate of each question item and age in response to the question, “During the past year, while supporting a family member with 22q11.2 deletion syndrome, what difficulties have you encountered in welfare and daily life?”

Table 5: Thematic analysis of responses to the question, “Please describe any difficulties you have encountered in using welfare systems such as disability pensions, disability certificates, and intractable disease certification due to multiple diseases or disabilities.”

Category	Examples
Lack of information provision	a. Difficulty understanding the concept of the disease It is unclear which diseases fall under which categories.
	b. Difficulty obtaining information about the disease I was not given any explanation about the procedures to obtain certification or disability certificates and had to research and apply on my own. No information was provided regarding intractable disease certification.
Difficulty in using the system	a. Insufficient, inconvenient, and unclear system He/She obtained a disability certificate for a heart condition, but it doesn't apply unless the chromosomal abnormality is stated. I don't know what to do or when I should do it regarding the certificate.
	b. Vertical divisions preventing comprehensive response He/She receives separate treatment for heart disease, intellectual developmental disorder, and other issues, but he/she cannot receive comprehensive care. Each medical certificate submission costs money, and the system does not consider overlapping disabilities due to its compartmentalized structure.
	c. Severity of multimorbidity not being assessed I want the disability pension system to evaluate multiple diseases and disabilities affecting one individual in an integrated manner. I'm currently applying for a disability pension for intellectual developmental disorder, but it's hard to explain the wide range of difficulties that can't be comprehended by that alone. Since the system evaluates each condition separately and then adds them together, multiple mild disabilities are not recognized. I would like the condition to be assessed as 22q11.2 deletion syndrome. Despite having severe cognitive impairment from schizophrenia, he/she cannot receive certification for intractable diseases because his/her heart condition is mild. He/She has no physical disability, only mild intellectual and developmental disabilities, so I worry whether they can continue renewing the certificate each time. I was surprised to learn that applying for a disability pension under 22q11.2 deletion syndrome alone is difficult and that I need to apply under intellectual or psychiatric disorders.
	d. Lack of knowledge among supporters I couldn't get clear answers about intractable disease designation from the support institutions. When I asked the local government which is better—pediatric medical aid or intractable disease designation—many staff were not well informed, and it took a long time. I constantly feel that municipal and health center staff lack knowledge. Instead of confirming with me, they try to research among themselves, and I am kept waiting for a long time.
Psychological resistance	a. Resistance to using the system as a person with a disability It took courage to accept that holding a disability certificate would identify my child as having a disability. I have not yet disclosed the certificate to my employer for tax deduction purposes due to opposition from my family. My spouse is unwilling to apply.

Table 5: Thematic analysis of responses to the question, “Please describe any difficulties you have encountered in using welfare systems such as disability pensions, disability certificates, and intractable disease certification due to multiple diseases or disabilities.”

Table 6: Thematic analysis of responses to the question, "What kind of support do you think is necessary for persons with 22q11.2 deletion syndrome and their parents regarding welfare services?"

Category	Examples
Information provision	a. Information about the disease Email consultation with specialists in chromosomal abnormalities and genetic counselors. Provision of information from welfare services.
	b. Public understanding of the disease It is still a minor disease and not well known. It is desirable to raise awareness among the general public so that patients and their families can have access to more information and options.
Comprehensive support	a. Comprehensive consultation services A place where we can consult about anything and gain a thorough understanding of the disease. Support that takes into account multiple diseases. Services where you can consult about the kind of support available. Comprehensive consultation services that are available from child- to adulthood, regardless of age. Support that provides comprehensive advice on future career paths based on knowledge of 22q11.2 deletion syndrome.
Supporters with specialized knowledge	a. Supporters who think together with the person with the disease and their parents It is desirable for a third party other than the parents to be involved in decision-making with support as the person grows up, including financial management, so that they can feel secure. Welfare services that respect the person's way of life and staff that think together with the parents.
	b. Supporters who act as intermediaries between the family and relevant organizations When it comes to support for school enrollment, it is desirable to have someone familiar with the hospital and disorder act as an intermediary. Due to the rarity of the disorder, explanations from the family may not be fully understood or may be misunderstood.
Improvement of systems	a. Support that considers the characteristics of the person with the disease After-school day services are helpful, but it can be difficult to find an appropriate place for the individual to stay, as they may become tired from being around children with developmental disorders or severe intellectual developmental disorder. Unlike normal intellectual developmental disorder and neurodevelopmental disorders, one-on-one communication is easier, so there are differences between special needs schools, special needs classes, after-school day services, group homes, and other facilities. He/She may want to be treated normally, but they may also desire people and places to make allowances for the fact that they cannot do things like normal people. Employment support begins after high school graduation, but it would be better to receive employment support upon request if he/she does not attend a special needs school. I hope there is a university that will accept him/her and understand his/her desire to learn. He/She wanted to find regular employment through a quota system for people with disabilities or work at a Type A workplace, but I couldn't find a company that suited him/her because he/she was very nervous around people. He/She currently works at a Type B workplace, but I think they are capable of doing more. I would like employment support tailored to the psychological characteristics of 22q. I would like step-by-step, attentive support for social participation and employment.
	b. Expansion of existing support A place where children can be safely left in emergencies. It would be helpful if school transportation support were available every day. Services that provide long-term nursing care are needed to reduce the burden on parents. One and a half hours per day is insufficient to alleviate the burden. Due to combined disabilities, basic equipment support is needed regardless of parental income.
	c. Support that considers multimorbidity Due to increased susceptibility to infection, the child often has a fever from otitis media and colds, and can only attend nursery school for less than a week a month. Family members and relatives have no choice but to take turns looking after the child. A day service is needed where children with heart disease can be left with peace of mind.
	d. Correction of disparities based on the place of residence In our area, we have to make the most of the support plans ourselves, and we would like to be able to make plans with the help of support staff, as in other municipalities. There are few developmental support facilities nearby, and there are no options. We want to consult with professionals to receive appropriate therapy. There are no social workers in the municipality, so we have to research everything on our own.
	e. Support after parents become elderly We want consideration for income restrictions on welfare benefits when parents are elderly. We want more options for where children can go after their parents pass away. As parents age, we want more options for living arrangements, such as group homes, in addition to hospitalization and respite care.
	f. Support for parents Parent training programs such as anger management for parents.
	g. Support that encourages a positive outlook We want an environment where we can work while growing together with our children in a positive way. Support that helps parents and children accept illness together and look forward to living together in a positive way.

Table 6: Thematic analysis of responses to the question, “What kind of support do you think is necessary for persons with 22q11.2 deletion syndrome and their parents regarding welfare services?”