

CASE REPORT

Dohsa-hou intervention for reciprocal interpersonal interaction for a girl with Kabuki syndrome and autism spectrum disorder

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Abstract

Although available evidence for psychosocial treatment for patients with Kabuki syndrome is limited, Dohsa-hou, a psychomotor therapy, could be a treatment option for autism spectrum disorder associated with the disorder.

KEYWORDS

autism spectrum disorder, Dohsa-hou, interpersonal relations, Kabuki syndrome, psychological intervention, social interaction

1 | INTRODUCTION

This case report illustrates an intervention using Dohsa-hou, a psychomotor therapy, for a girl with Kabuki syndrome and autism spectrum disorder. Although available evidence is limited, Dohsa-hou could be a treatment option for autism spectrum disorder associated with genetic disorders.

Kabuki syndrome is a rare genetic disease reported by Kuroki et al and Niikawa et al.^{1,2} The frequency of the disease is estimated to be 1 in 34 000 people.³ The disease was defined as a malformation syndrome and reported five major symptoms: (1) characteristic facial features, (2) skeletal malformations, (3) dermatoglyphic abnormalities (various patterns on the surface of the skin), (4) mild to moderate intellectual disability, and (5) growth deficiency after birth. A majority of patients have a pathogenic variant in the *KMT2D* or *KDM6A* genes. However, there are a substantial number of patients who do not show pathogenic mutations in *KMT2D* or *KDM6A*.⁴⁻⁶ A recent international consensus proposed that a definitive diagnosis can be made based on a history of infantile hypotonia, developmental delay, and/or intellectual

disability and typical dysmorphic features and/or pathogenic variant in *KMT2D* or *KDM6A*.⁴

Most cases are characterized by mild to moderate intellectual disability, and a few patients have severe disabilities.^{7,8} There are several case reports of patients with Kabuki syndrome who exhibit features suggestive of autism spectrum disorders (ASD).⁹⁻¹² ASD is primarily characterized by persistent deficits in social interaction and communication with others. Impairment in such abilities often leads to difficulties in development of interpersonal relationships. Therefore, development in communication with other people and experiencing support through interaction with others is an important issue. Generally, social communication was not impaired in most children and adolescents with Kabuki syndrome, although some patients had poor eye contact and repetitive behavior.^{13,14} Such characteristics may differ from the typical behavior profile of ASD patients. In addition, insufficient information is currently available for psychological treatment for this specific population.¹⁵

Although there are several psychological treatments for children with ASD, such as social skills training,¹⁶ evidence for the effectiveness thereof is limited to ASD

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without moderate or severe intellectual disabilities. Nonverbal children with ASD and severe intellectual disabilities may need to be treated differently as suggested in a previous study.¹⁷ Considering the limited evidence of improvement in reciprocal and spontaneous social interaction in nonverbal children with ASD, different treatment approaches may need to be developed.¹⁸ Dohsa-hou is a psychotherapeutic approach developed in Japan,¹⁹⁻²¹ which focuses on body movement and psychomotor experiences in individuals.²²⁻²⁴ There are several case reports presenting Dohsa-hou practice for children with ASD.^{25,26} In addition, Morisaki²⁷ suggested three therapeutic goals in interacting with children with ASD through Dohsa-hou: experiencing relaxation, self-regulating behavior, and sharing intention with others and realizing the presence of others. Joint attention and the sharing of intentions with others are essential aspects of development in social functioning and interaction in play among children. In addition, various studies have focused on the role of joint attention in the development of relationships with others. Joint attention is established through the interaction between the child and others and between the child and objects (such as toys) in a binary relationship, followed by the interaction between the child, objects, and others in a triadic relationship.²⁸ This behavior is essential for the development of shared intentions to notice what others are paying attention to and has been regarded as a milestone of communication development necessary for subsequent social interaction skills.^{29,30}

As suggested in the literature, such an interaction can lead to the development of social relationships and joint attention through the experience of sharing intention with body and movement, even with nonverbal children. In addition to verbal instruction, interaction with physical contact and body movement is the cardinal features of the Dohsa-hou intervention; thus, Dohsa-hou can be a treatment option for nonverbal children with difficulties in social interaction, as occurring in ASD. Currently, there is limited literature regarding psychotherapeutic interventions for children with Kabuki syndrome.¹⁵ However, previous reports related to ASD in other populations may inform treatment considerations in the disorder. Here, we report on a case study of a child with Kabuki syndrome and ASD, in which the changes in the child's behavior were examined through the intervention of Dohsa-hou.

2 | CASE PRESENTATION

2.1 | History

At the beginning of the intervention, Sara was 7 years and 8 months old and in the second grade of a special education

school. When she was 3 years old, Kabuki syndrome was suspected based on the clinical signs and she underwent genetic testing for *KMT2D* and *KDM6A* mutations, but no pathogenic mutations were detected by sequencing. Based on the clinical manifestations and typical dysmorphic features of Kabuki syndrome, she was diagnosed with Kabuki syndrome, based on its cardinal features of the disorder.

Sara had severe intellectual disability and ASD, and her intellectual functioning was assessed using the Enjoji-developmental scale³¹ (Developmental quotient = 22). The subdomains were as follows: motor domain, 36; social, 14; and language, 17. She was nonverbal, and she could not pay attention to the objects only with verbal instructions. She had been receiving physical and occupational therapies once or twice a month at a medical center from the age of 8 months to 6 years and 7 months.

2.2 | Intervention

The Dohsa-hou intervention began in June 2018, and a total of 16 sessions were conducted for 6 months. The content of the Dohsa-hou intervention included interaction tasks; relaxation in the shoulders, back, and trunk; using the hip joint in a kneeling posture; and standing on the ground.^{19,32} The interaction task aimed to improve the interaction between Sara and the therapist by being aware of the therapist's intention. The therapist joined her hands with Sara, then moved the hand and arm upwards, with Sara matching the movement guided by the therapist.

After the intervention, playtime was set, and interactions with the therapist were video-recorded. Playtime was not set at sessions 1, 3, 6, 8, 12, and 14 due to time constraints. From sessions 2 to 5, the child played freely, and in sessions 7-16, she and the therapist played with the therapist's involvement.

Follow-up sessions were conducted 2 months after the end of 16 sessions. The intervention was delivered by a female therapist in training with the supervision of a certified clinical psychologist.

2.3 | Assessment

2.3.1 | Quality of shared intention in Dohsa-hou and play sessions

Shared intention in Dohsa-hou sessions was evaluated by the following six-point scale: indifference (0); looking at the therapist but not trying to confirm the therapist's intentions (1), looking in the eyes when requesting for something (2); children looking at the therapist and the object (alternately) when the therapist points at the object (eg, a body part) (3); making eye contact with the therapist in response to the

action of the therapist (4); and making eye contact with the therapist spontaneously to show the therapist something (5).

Shared intention in play sessions was evaluated by the following six-point scale: playing alone without paying attention to the therapist (0), paying attention to the therapist but playing alone (1); responding to encouragement from the therapist but returning to playing alone (2); playing together with the therapist but exhibiting limited motivation to share intentions (3); responding to encouragement from the therapist, sharing toys and playing, and playing together (4); and the child seems to encourage the therapist to share their intentions (5).

To assess autistic symptoms, joint attention, and social adaptation, we used the Social Communication Questionnaire (SCQ), the Joint Attention Behavior Scale, and the Vineland Adaptive Behavior Scale-II. These measures were administered at preintervention, session 10, session 16, and follow-up (2 months after the intervention).

2.3.2 | Social communication questionnaire

The Japanese version of the Social Communication Questionnaire (SCQ) is a screening tool to clarify the presence or absence of symptoms related to ASD, consisting of 40 items.^{33,34} A higher score indicates higher autistic symptoms. It comprises three symptom domains (reciprocal interpersonal relationships, communication, and limited, repetitive, and stereotyped pattern of behaviors) that correspond to the Autism Diagnostic Interview-Revised (ADI-R), a clinical diagnostic instrument to assess ASD based on parent interviews. These domains were used to evaluate changes in autistic symptoms of the participant. Sara's overall score was 22 at preintervention, which is above the cutoff score for ASD screening.

2.3.3 | Joint attention behavior scale

This scale contains 17 items related to joint attention, which includes gazing, production and understanding of pointing, alternating gaze, understanding of others' emotions, and representations.³⁵ The caregiver responded to the yes/no questions (0/1). The higher the score, the more developed the behavior related to joint attention. She scored 0 at preintervention, suggesting profound impairment in joint attention.

2.3.4 | Vineland adaptive behavior scale-II

The Japanese version of the Vineland Adaptive Behavior Scale-II (Vineland-II) is a semi-structured interview that evaluates the adaptive behavior of individuals. It assesses the

four domains of adaptive behavior: communication, daily living skills, social skills, and motor skills.^{36,37} At preintervention, Sara's Vineland-II scores showed profound impairment in the total score (Total = 21) and all four domains (communication = 22, daily living skills = 20, social skills = 20, and motor skills = 20). The social skills domain (socialization) was repeatedly evaluated (intake, sessions 10 and 16, and follow-up), as this study focused on social relationships and behavior.

2.4 | Ethical considerations

This study was conducted in accordance with the recommendations of the Declaration of Helsinki of the World Medical Association, with written informed consent from the guardian of the participant. The study protocol was approved by the research ethics review board of the Faculty of Education at Oita University. The names and identifying details have been changed to protect the privacy of the participant.

3 | RESULTS

3.1 | The course of Dohsa-hou sessions

During the intake session, Sara made little eye contact and little response when the therapist would call her name. There were many times when Sara seemed to reject the therapist's hand when the latter offered it. At playtime, she did not respond to the therapist's words. She did not seem to respond to the therapist's invitations to play with her by showing her toys; and she even appeared to avoid the therapist's invitations. She did not make any request nor maintained eye contact with the therapist. She just played by herself.

In Sessions 1-5, in the interaction task, Sara sometimes tried to make hand movements with the therapist. She gradually turned her gaze toward the therapist and sometimes touched the therapist, indicating that she was trying to get involved with the therapist. At playtime, she made eye contact with the therapist a few times but turned away from the therapist who tried to engage her, and her response to the therapist's invitations was indifferent or negative. Sara showed little interest in the therapist and played by herself with a toy.

In Sessions 6-9, in the interaction task, when the therapist was assisting her, Sara tried to move her hands following the therapist and stared at the therapist more often. In addition, when the therapist looked at Sara's hand, she looked at her hand more often. She began to look at the therapist's face during the task and smiled several times. She seemed to be enjoying the task with the therapist. At this time, Sara often approached the therapist, held hands with the therapist, and waved her hands up and down as if she wanted to be involved

with the therapist. At playtime in this period, she began to respond to the therapist's approach and smiled, although she sometimes started to play by herself. At that time, she began to reach for the toys and became more involved with the therapist. She began to share her attention with the therapist during play, for example, by looking at the therapist for confirmation.

In the interaction task in later sessions, Sara was able to move her body in accordance with the therapist's movements and intentions. When the therapist used words, such as “slowly” and “move with me” and moved her eyes, she moved following the therapist's movement. In addition, Sara began to show movements to the therapist as if she wanted to perform the interaction task with the therapist. In the task in kneeling posture, she was able to move her back in response to the therapist's instructions and was able to work calmly. At playtime in this period, her interest in the therapist's movements and actions was more pronounced. She sometimes sat in front of the therapist and looked at her expectantly. They often made eye contact, and when the therapist laughed, Sara often smiled as well. She shared her toy and played with the therapist steadily.

During the follow-up session, she was able to maintain eye contact with the therapist and continued to focus on each task. She concentrated on the therapist's gaze and movements and moved her arms in response to the therapist's intentions.

3.2 | Assessment

Changes observed during and after the intervention are presented in Figure 1 and Table 1. Although there were no major

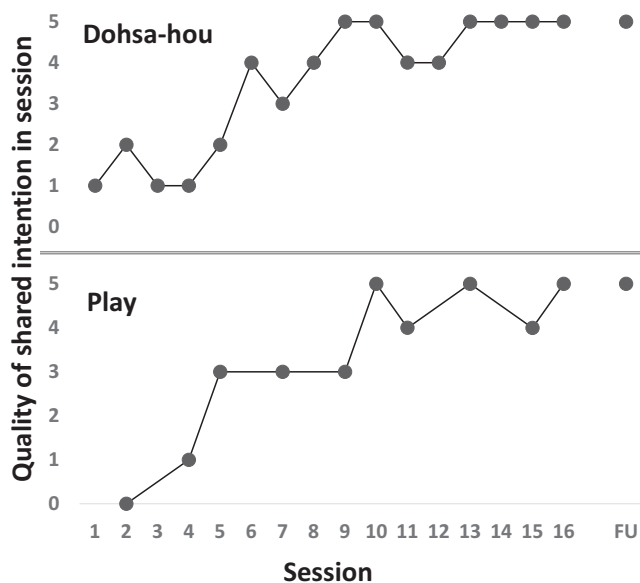


FIGURE 1 Quality of shared intention in Dohsa-hou and play sessions. FU, Follow-up

functional changes in the communication domain of ASD symptoms, there were improvements in reciprocal social interaction as assessed by the SCQ, interpersonal relationships as assessed by the Vineland-II, and play and leisure time by Session 16. No clear improvement was observed in joint attention or the other domains.

At the follow-up, Sara sometimes looked at her mother's eyes and faces and drew their attention to herself more than the preintervention period. Her mother reported that Sara also smiled back when someone smiled at her, tried to join in interpersonal games, such as hand games, and responded favorably when other children approached her.

4 | DISCUSSION

In the present study, we described an intervention using Dohsa-hou for a girl with Kabuki syndrome associated with ASD and examined the changes during and after the intervention. Reciprocal interaction improved in her daily life, as well as in Dohsa-hou and play sessions. Regarding joint attention, changes were observed in the sharing of intentions during the sessions with the therapist, but no clear improvement was observed in daily life.

There are several factors associated with the intervention's effectiveness. In the early stages of the Dohsa-hou intervention, Sara was not able to pay attention to her own body or the intention of the therapist in the interaction. As the session progressed, she became aware of the therapist's intentions and was able to share her intentions in the sessions. This process involved paying attention to her own body movements through physical experience and noticing the therapist's support (ie, intentions) in the process. Through the process, there was a preceding change in the way she interacted with the therapist during the treatment sessions. Following this, the changes in reciprocal relationships with others were observed in daily life. Shared intention and change in interactions through joint action coordination could be therapeutic factors that lead to reciprocal interpersonal relationships in ASD as suggested in recent literature.^{38,39} Therefore, interventions, such as Dohsa-hou and/or psychomotor therapy, can be helpful for this population.¹² While such physical contact enables us to develop reciprocal interactions in these children, children with high tactile sensitivity may have difficulties in this approach. As ASD are associated with certain cases of rare genetic disorders including Kabuki syndrome,⁴⁰⁻⁴² such an approach would be a viable treatment option for these children with the disorders.

In this study, we only evaluated patient functioning; however, parental burden is also important for the management of the families.^{43,44} Parent-mediated interventions are essential treatment options for children with ASD and their parents.^{45,46} Investigating caregiver-delivered

TABLE 1 Assessment of autism spectrum disorders symptoms and socialization skills

Measure	Preintervention (intake)	Session 10	Postintervention (session 16)	Follow-up
SCQ	22	21	16	14
Reciprocal social interaction	13	12	8	6
Communication	8	8	8	9
Restricted, repetitive, and stereotyped patterns of behavior	0	0	0	0
Joint attention behavior scale	0	1	2	2
Vineland-II socialization domain	4	4	5	5
Interpersonal relationships score (index)	11 (1)	14 (1)	16 (2)	16 (2)
Play and leisure time score (index)	5 (1)	8 (1)	11 (1)	11 (1)
Coping skills score (index)	2 (2)	4 (2)	4 (2)	4 (2)

Abbreviations: SCQ, Social Communication Questionnaire; Vineland-II, Vineland Adaptive Behavior Scale-II.

Dohsa-hou interventions for ASD children is also an important consideration in future research which would increase the involvement of caregivers and the possibility of sustained support at home. Because no curative treatment is available for the disease, further research is required to establish good management practice for patients and families. Obviously, interest, compassion, and connection are essential for psychosocial support in these people and in intervention methods.

There are a few issues to be considered in this study. Kabuki syndrome is usually associated with less severe intellectual disability (ie, mild to moderate),⁴⁷ but the participants in this study had both severe intellectual disability and ASD. The outcome may have been affected by the patient's functioning and the frequency of the intervention. As a previous intervention study suggested that more severe autism symptoms were associated with less improvement in spontaneous behavior,¹⁸ baseline severity of the patient may affect the ineffectiveness in joint attention and communication. Additionally, a more extensive program with a high frequency⁴⁸ may provide better results in patients with severe disabilities. The number, frequency, and duration of each session could affect the improvement; however, these parameters in intervention programs for children with ASD vary in studies.⁴⁹ Future interventional and cohort studies would provide insight into the adequate treatment characteristics and persistence of the effects. Although joint attention was established with a specific person (ie, the therapist), it did not generally apply to other relationships. However, the Joint Attention Behavior Scale measures disability and may not be appropriate for detecting therapeutic changes.

In conclusion, we described Dohsa-hou intervention for a girl with Kabuki syndrome, who has ASD and intellectual disability, that resulted in changes in her interaction with others and sharing of intentions. These changes may have

been accompanied by changes in interaction with the therapist during the Dohsa-hou and play sessions. The Dohsa-hou could be a treatment option for ASD associated with genetic disorders.

ACKNOWLEDGMENTS

The authors would like to thank the participant and her family.

CONFLICT OF INTEREST

The authors declare that they have no competing interests.

AUTHOR CONTRIBUTIONS

JK: developed the intervention, performed the intervention, interpreted the data, and wrote the first draft of the manuscript. HF: developed the protocol, supervised the intervention, interpreted the data, and wrote the manuscript. All the authors have read and approved the final manuscript.

ETHICAL APPROVAL

This study was carried out in accordance with the ethical standards set forth in the 1964 Declaration of Helsinki and its later amendments, with written informed consent from the guardian of the participant. The protocol was approved by the Oita University Faculty of Education Research Ethics Committee (30-004). Names and identifying details have been removed to protect privacy of the participants.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request. The data are not publicly available due to privacy or ethical restrictions.

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How to cite this article: Kawano J, Fujino H. Dohsa-hou intervention for reciprocal interpersonal interaction for a girl with Kabuki syndrome and autism spectrum disorder. *Clin Case Rep*. 2021;9:e04296. <https://doi.org/10.1002/ccr3.4296>