Prosthetics and Orthotics International

A physiotherapeutic approach to a baby with right disgenesis of thumb and left agenesis of upper extremity: A case report

Habibe Serap Inal[1] and Osman Reis[2]

Abstract

A boy with bilateral congenital anomalies of the upper extremities with transverse absence of the left arm (agenesis) and absence of right thumb (disgenesis), fixed elbow in extension due to humeroradial synostosis thought that the humerus was intact. His wrist could move passively with 50° flexion, 0° extension, and 70° radial deviation. The other 4 fingers were intact, 4–5 metatarsal bones were in synostosis, and the fifth finger was clinodactyly. He was born by normal delivery. The physiotherapist started sensorimotor training to achieve functional movements of the right upper extremity. The exercises for the trunk muscles were emphasized to prevent the future possible thoracolumbar curve due to the absence of loading of the left arm on trunk muscles. Serial splinting was performed to position the wrist and fingers for functional purposes and abduction and internal rotation of the index finger to perform the opposition position until the pollicization surgery. At 1 and a half, his trunk was straight, and the wrist was partially corrected (15° extension; 35° radial deviation). He was able to play with toys and eat food. The mother rated his performance as 8/10 and 10/10, respectively. Both mother and father were satisfied with his performance in activities of daily living according to his age compared with his peers (9/10; 10/10, respectively). Thus, the physiotherapist's sensorimotor training and the positioning splints may be considered as feasible interventions in this case.

Keywords

bilateral congenital amputation, thumb amputation, infant, exercise, hand splints, activities of daily living, participation

Date received: 23 January 2024; accepted 21 November 2024.

Background and purpose

Congenital anomalies of the upper extremities are estimated to be 27.7 per 10,000 births.[1]

Bilateral congenital upper extremity anomalies are among the most severe congenital deformities that affect the daily life and level of independence of children.[2,3] International Classification of Function, the biopsychosocial approach that describes human function at 3 levels as function, activity, and participation is a good tool for clinical assessment of upper extremity, especially the hand function.[4] For understanding the function of the extremity, the strength, sensibility, range of motion, and flexibility, and for assessing the activity, the capacity of doing the activity (Can do!) and the actual performance in the activity (Does do!) are assessed. The last level is participation that assesses the amount of participation to the daily living activities and social and recreative activities. However, this last level is not in the issue since the case presented here is an infant.[5]

Thus, under the guidelines of International Classification of Function, a multidisciplinary rehabilitation team approach is essential[2] not only for improving strength, range of motion (ROM), and trunk asymmetry but also for promoting participation of the child as a whole person in the mainstream of the society. Therefore, starting from the first day of diagnosis in intrauterine life, the physiotherapist's responsibility is to increase muscle strength and normal range of motion to achieve functional movements, a good posture specially in his trunk against the thoracolumbar curves.[6]

The other important aspect of the rehabilitation of children with bilateral congenital anomalies is the resilience of the parents. They are often shocked and feel guilty when they learn of their baby's condition. Parents are expected to adapt to their child's different condition, to help their child accept treatment interventions, and to support him/her in coping with possible emotional distress related to functional limitations[7] or the experience of being visibly different.[8]

Therefore, good follow-up by the physiotherapist from the first day of diagnosis is necessary, including counseling, exercise, orthotic, and prosthetic treatment. However, to our knowledge, the literature on physiotherapy and rehabilitation of children with bilateral congenital anomalies and family counseling is limited.

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Associate Editor: Christopher Wong

DOI: 10.1097/PXR.0000000000000430

Thus, we aimed to present our experience and discuss the results of early exercise and splinting interventions for children having bilateral congenital upper extremity deformities in relation to the achieved outcomes of this case. Our goal was those physiotherapeutic approaches including sensorimotor exercises and right-hand positioning splinting for a baby with right disgenesis of thumb and left agenesis of arm may be effective to achieve a functional right hand.

Case description

A one-and-a-half-year-old boy with bilateral congenital anomalies of the upper extremities with transverse absence of the left arm (agenesis) and absence of right thumb (disgenesis) who was born by normal delivery (Figure 1). The other 4 fingers were intact, except the fifth finger that was clinodactyly and 4–5 metatarsal bones were in synostosis. Owing to humeroradial synostosis observed in the X-ray, his elbow joint was fixed straight in extension; however, his humerus was intact. His wrist was positioned in flexion and radial deviation that could move passively with 50° flexion, 0° extension, and 70° radial deviation. The absence of right thumb (disgenesis) classified at level V according to Blauth classification[9]; 2–5 fingers developed and freely movable, synostosis of 4–5 metacarpals seems to prevent the functional movements of the baby as hook type holding and releasing the objects with his 4 fingers.

Parents were in their late 20s when he was born, and he had a brother 3 years older than him with no medical history. Parents are teachers, and there is no family history of congenital malformations. The study conforms to the Declaration of Helsinki.[10]

The case has been under physiotherapy since birth, including family counseling, home exercises, and splinting that was

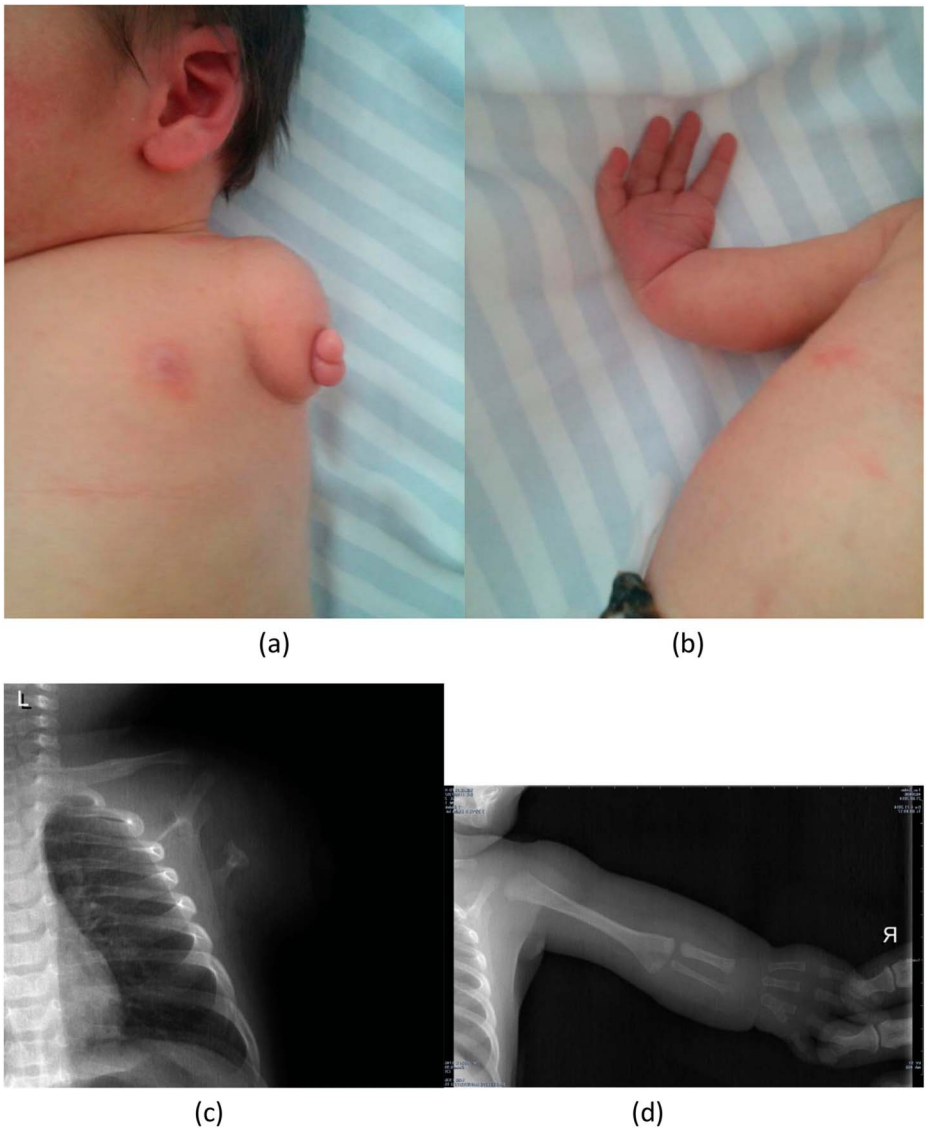

Figure 1. Transvers absence of left arm (Agenesis) (a); disgenesis of the right thumb (Level V according to the Blauth Classification) (b); X-ray of the left side (c); and X-ray of the right arm before splinting (d).

Table 1. Physiotherapist approach in sensorimotor training of the severe bilateral congenital abnormalities for body awareness and good posture.

Age	Aim	Exercises	Timing	Repetitions
First 3 mo	Feeling and recognizing the hypoplastic and amputated parts	• Tiny touches and tactile stimulations (little pinching, tapping, touching by fingers, and with rubber or furry toys to right arm and hand, and shoulders • Active range of motion exercises for right shoulder; active/active assistive range of motion exercises for wrist, hand, and fingers • Not to hold always on 1 side, change the holding positions	From the first day in every diaper change and after feeding and on any occasion	5–6 times in each
	Not to ignore or deny the amputated arm and the thumb	• Passive/active assistive trunk extension exercises • Changing the position of the bed relative to the door alternatively so that he can look at the door from both sides	During the day in every 2 wk	Minimum 10 times many times as the baby grows
	Prevent the possible right-side scoliosis and feeling the body symmetrically	• Symmetrical active assistive exercises for arm and legs (hips, knees, ankles, shoulder, wrist, and fingers flexion-extension; hips and shoulder abduction-adduction and rotations) • Active/active-assistive protraction and retraction of the left shoulder while exercising the right shoulder for symmetrical feeling	Three times in a day while he is awake	5–6 times each
4–6 mo	Body awareness by feeling and recognizing the hypoplastic and amputated parts	• Interventions in the first stage continued	Same as in the first stage	5–6 times each
	To achieve good muscle structure and prevent the possible right-side scoliosis, and additional deformities and keeping the segments functional	• Symmetrical active assistive exercises for trunk and left arm • Symmetrical active assistive trunk extension exercises by tickling the right side of the trunk for right-side lateral flexion in prone position with symmetrical head extension • Shoulder active ROM exercises, active and active assistive wrist and finger extension/flexion, and ulnar deviation ROM exercises	Three times in a day while he is awake	5–6 times each

(*continued on next page*)

Table 1. Physiotherapist approach in sensorimotor training of the severe bilateral congenital abnormalities for body awareness and good posture. (Continued)

Age	Aim	Exercises	Timing	Repetitions
7 mo and above	Feeling and recognizing the hypoplastic and amputated parts in different positions	• Sitting, right side supported by pillow/soft toy, holding/ releasing toys, playing with mother, brother and siblings	3–4 times in a day	For 15–20 min each
		• Lying down supine trunk slightly raised with triangle pillow to support arm left shoulder for the right shoulder approximation sense	Twice in a day	For 10 min each
		• Symmetrical sitting	Always	As required as the baby grows

ROM, range of motion.

prescribed by the physiotherapist (HSI) and manufactured by the orthotist (OR). The family counseling started during the 6th month of pregnancy when they got to know the situation after the first ultrasound examination. The parents were suggested to consult a physiotherapist by their family physician.

Physiotherapist approach in rehabilitation of the case

The physiotherapeutic approach was set as (1) family counseling during pregnancy, which continued all stages until the present; (2) first 3 months, the initial stage for sensorimotor training and integration of the affected parts with the rest of the body; (3) 4 to 6 months for active and active assistive exercises combined with play for a symmetrical body shape and body image; and (4) 7 months and above for activity involvement and participation in activities of daily living (Table 1).

Family counseling during the pregnancy consisted mainly of answering their questions and comforting them. Although the parents were shocked, they were also resilient to this new condition. Both had questions like how to hold him or feed him. The mother was worried if she would do any harm while holding him or dressing him. They were told that these were all expected worries, possible questions. The answers were hidden in her instincts as a mother.

The father was curious about the future of the baby. He did some research on internet before the meeting. Although the physiotherapist gave some explanations and examples about the activity level of upper extremity amputees and their quality of life, he was not satisfied. However, the information on the possible success in sports as a recreational or competitive athlete made him suddenly relieved and led him to set a future goal for his son. However, from the physiotherapist point of view, the most

Figure 2. Symmetrical trunk extension exercises by rising the head.

Table 2. Time of onset of the activities in a typically developing child compared with the presented case and his skill levels, According to parents grading from 1 to 10.

Activities	Time of onset in a typically developing child (mo)	Time of onset in the case (mo)	Skills of the case in the activities (0–10)[a]
Hands fist, reflex grip	1	2–3	7/10
Taps toys lightly with hand	2	4	7/10
Fist opens	3	5	6/10
Brings hands to midline		5	5/10
Grasps/holds objects		7	9/10
Reaches with both hands/arms	4	5 (unilaterally)	10/10
Brings hand/fingers to mouth		5	7/10
Gripping/squeezing begins		8	9/10
Reaches for objects purposefully	5	7	10/10
Brings objects/toys to mouth		6	8/10
Lifts spoon or glass by handle		7	9/10
Begins to catch your feet		12	8/10
Reaches and grasps objects	6	8	8/10
Passes toys from 1 hand to another		—	—
Makes sound by hitting the glass on the table		8	10/10
Crosses body while lying down	7	8	8/10
Uses whole hand to scratch		—	—
Begins to hold with 2 fingers		10	7/10
Cubes crash into each other	8	10 (unilaterally)	10/10
Uses 3 fingers when holding		12	9/10
Holds using thumb and index finger	9	—	—
Gross releasing starts		10	10/10
Points with index finger		10	—
Pokes with fingers	10	10	10/10
Precise hold with thumb and index finger		—	—
Nests objects		10	8/10
Release the cube intentionally	11	11	8/10
Removes toy nails from its socket		—	—
Puts objects into a large bowl	12	14	9/10
Release the objects precisely		15	8/10

[a]The range of grading as 0: not possible to perform and 10: able to perform in a normal way.

important aspect at this stage is to be realistic and prepare the parents to meet their child in the most comfortable way. Counseling continued twice weekly in person for 4 weeks, 15 minutes each, and then by phone as needed for 5–10 minutes.

The goal of the *exercise treatment* was to prepare the child to actively participate in activities required according to the stages of normal motor development and to prepare the parents on how to cope with different stages of development to achieve progression in his functional activities.[11]

The exercises given starting from the first week in the context of sensorimotor training consisted (1) vestibular stimulation for integrating the right upper limb, hand and finger movements—as affected parts—with the head, trunk, and lower limb movements; (2) tactile stimulation for improving body awareness by means of little pinching, tapping by finger tips, touching with rubber or furry toys to the skin of amputee sides during the exercises; (3) auditory stimulation given by the mother as usual baby language or by means of little sound toys during exercises to appraise and express joyfulness in relation to the created movements; (4) complementary play exercises as every mother play with their babies, however, in this case supported with vestibular and tactile stimulations; and (5) gross motor

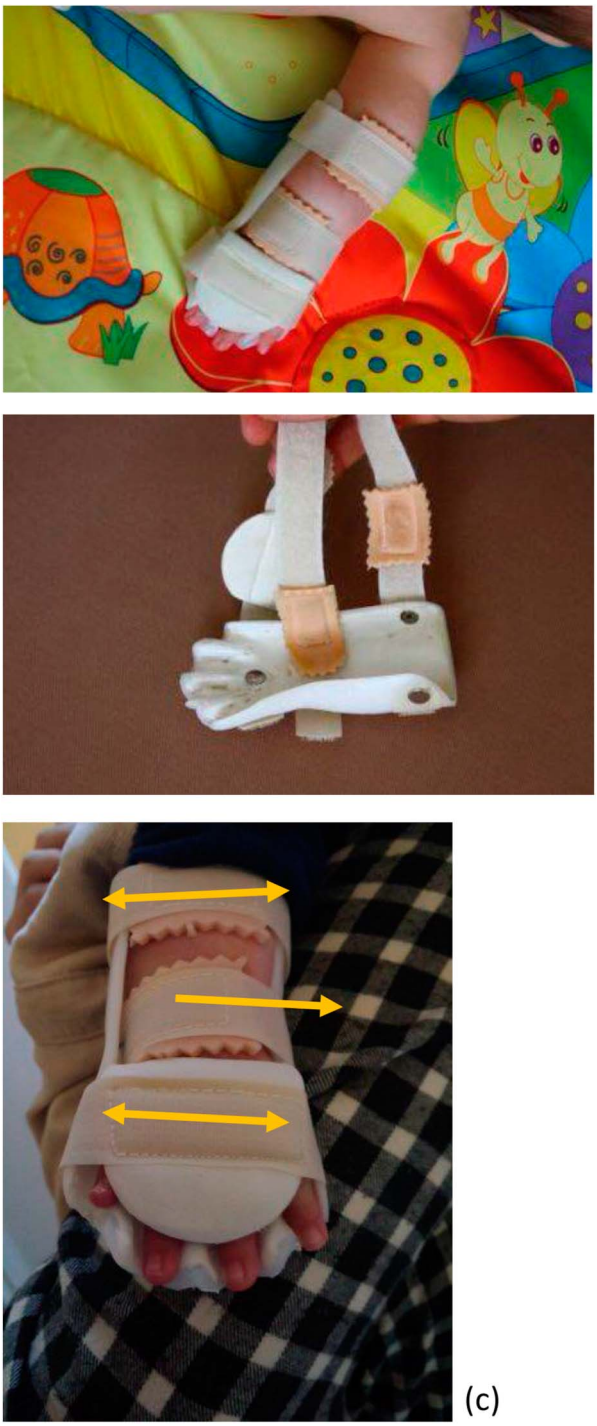

Figure 3. Resting splint positioning hand wrist and elbow (a), finger grows and positioning of the straps (b), and corrective forces given by the straps and finger pad (c).

milestones of head, trunk, and limb movements giving care for correct lying down on supine, prone, side lying (both sides) and supported sitting on the lap of the parents, such as during breast feeding of mother, as presented in Table 1.[2,11] They were given as home exercises twice in a day 5–6 times each for 15 minutes totally. The parents practiced them under the supervision of the physiotherapist (HSI) in the first session, and they were let to video them by their cellphone. In addition, exercises with explanations were given in a list. The follow-ups were done by face-to-face visits in every 3 months and by phone.

Symmetrical extension exercises were given to the trunk muscles by means of sensorimotor stimulations (Figure 2). The left site agenesis may trigger thoracolumbar scoliosis due to the loss of loading;[6] however, since the case was a new born, this was not the issue. For the preventive physiotherapy concept, we added these symmetrical extension exercises into the exercise program as a precaution against the postural changes of vertebral colon as the child grows and starts to use his right upper extremity effectively in this daily living life.

Bilateral range of motion exercises for the right shoulder and lower limb joints were given active assistive and for the wrist and fingers either passively or active assistively as the baby responded were included to the treatment plan. Splint was removed only during the hand and finger exercises. The goal of the exercise treatment was to achieve gross motor movements of the left shoulder, wrist, and fine motor movements of hand as the baby grows up and to improve the reflex movements of the arm according to the normal motor development stages of the child.[12] Such as taking the hand to mouth, chewing the fingers, grasping the parents' fingers, and touching and feeling the mother's skin.

Assessment of function and activity

Since this is a bilateral congenital upper extremity amputation case, with the absence of the left arm (agenesis) and the absence of right thumb (disgenesis) and unfunctional elbow joint with weak hook grip of the fingers on the right side, we assessed his gross and fine motor functions and activities of the baby in relation to his motor development stages.[12] The assessments were done in every 3 months during the face-to-face visits to observe functional improvements. The parents were asked to rate his daily living activities by means of the Visual Analog Scale (0–10)[13] as "0: not possible to perform," and "10: able to perform in a normal way" at the age of 1 and a half years (Table 2).

Orthotic approach of the physiotherapist

Splinting of the hand and fingers was performed with the aim of preparing the index finger for possible future pollicization. This was done in 2 consecutive procedures. First, a thermoplastic resting hand splint was applied to be used at night and during daytime sleep for 6 months (Figure 3). This splint aimed to position the wrist in a neutral position, secured with a Velcro strap that pulls it from the radial to the ulnar side to reduce the radial deviation (70°). To keep the fingers in extension without radial deviation, they were placed in their grooves and secured with a Velcro strap supported by a Plastazote piece, like a sandwich. In this way, the baby's hand was also prepared for cylindrical grip.

When the baby was 6 months old, thermoplastic resting hand splint was changed to a thermoplastic cock-up splint to widen the gap between the index and middle fingers and prepare the index finger for opposition with abduction and internal rotation (Figure 4). This splint was used for 6 months in the same system at night and during daytime sleep.

Figure 4. Thermoplastic cock-up splint to widen the gap between the index and middle fingers for opposition, trimmed from the blue lines.

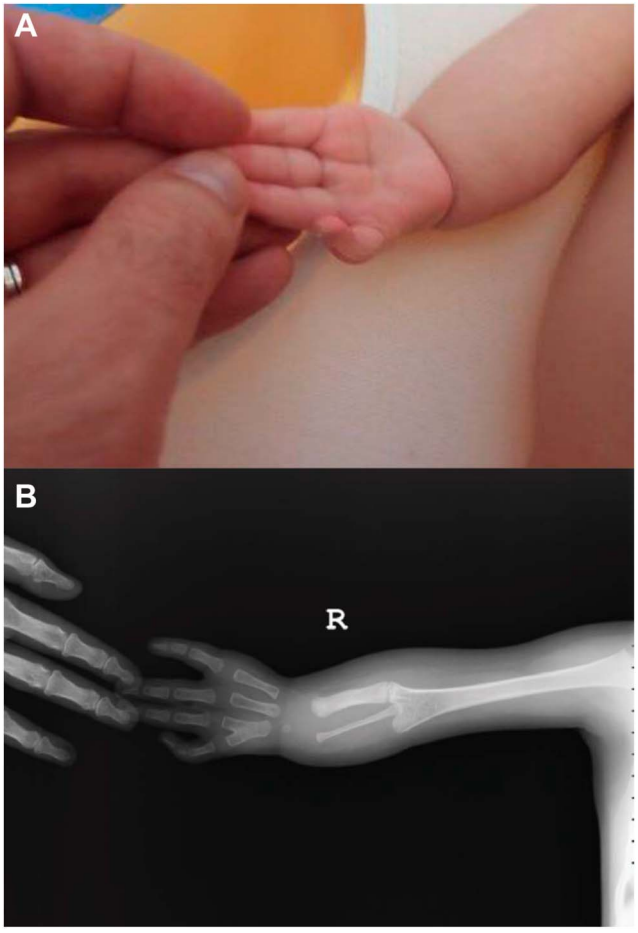

Figure 5. Extended right elbow and wrist and abducted index finger from the palmar site after positioning splint (a) and X-ray of the right arm from the dorsal site after positioning splint (b).

Outcome

At the age of 1 and a half years, he was able to hold objects, play with toys, and eat independently by holding the food (a piece of bread, biscuit, meatball, or a potato) or a spoon by his hand without any support. The activities were graded by the parents in which physiotherapist followed them in videos taken by the father in his home environment. They were (1) holding toys given to his hand according to his age as 8/10; (2) playing with toys by holding and releasing them voluntarily as 10/10; and (3) eating a biscuit or piece of bread independently as 10/10 (Table 2). Although he was able to grasp the toys and play with them by his abducted index finger, the inefficient opposition of the index finger and the synostosis of 4–5 metatarsals prevent fully functional palmar grasping (Figures 5 and 6). In addition, since the 5th finger is hypoplastic with a clinodactyly in the proximal joint, his grasping was not for a long time and lost the contact with the toys. The mother explained this as follows: "*He partially grasps and shakes the toy, but sometimes he loses contact, and it falls. We are happy that he holds the toy or our finger, this is important.*"

The mother's score for his performance in activities of daily living was 9/10 and the father's was 10/10. The father's and mother's perspectives on the effectiveness of the exercises were 9/10 and 10/10 and on the effectiveness of the splints were 9/10 and 10/10, respectively. The mother said, "*We both worked hard from the beginning, but especially he is a hard worker, so I expect more, for his future.*"

Discussion

The total loss of 1 limb and almost 40%–50% loss of hand function due to the loss of the thumb on the other side result in severe functional limitations in activities of daily living at any

Figure 6. Playing while sitting and holding a tray (a), holding a ball with abducted shoulder (b), and holding a toy with the abducted index finger by crossing the body (c).

age.[14,15] Although pollicization of the index finger, considered for aesthetic and functional purposes, is not recommended before the child is 1 year old that is expected to hold objects in pinch grip according to the normal developmental stage,[16] it was not considered in this case. Therefore, for good use of time, it was aimed to position the wrist and fingers at neutral position as extended wrist at mid-level without ulnar deviation for functional purposes with serial splinting and abduct and internally rotate the index finger to perform opposition position until the pollicization operation would contribute to the results.[3,16]

He was able to hold objects, play with toys, and eat food functionally in pinch or cylindrical grip at the age of 1 and a half years and expected to improve it. As it was stated, children with agenesis of 1 arm develop strategies throughout their lives to perform activities of daily living without a prosthesis with the support of their other side.[17] Since activity performance depends

mainly on active range of motion of the wrist and fingers rather than strength,[15] we emphasized active range of motion exercises of the wrist and fingers. However, we also exercised the trunk and right shoulder symmetrically against gravity to increase the strength of the upper trunk and shoulder muscles for the balanced movements.[18] As Needham and Nelson (2023) stated, once the infants succeed in reaching-to-grasping objects they like to practice to more that leads refinement in hand and finger movements.[19] We believe that the eagerness of the parents to motivate the baby to practice the exploratory actions of his hand and fingers in reaching out to the toys shown by the parents or grasping their fingers are also important for the improved functional activities necessary for the daily living life of the baby (Figure 5).

The parents were confident in the effectiveness of the exercises and positioning splints. However, the mother rated his performance in activities lower than the father. This may be due to her critical approach. She was also less satisfied with the effectiveness of the exercises and splints for both interventions. She explained this with a simple sentence: "Too much effort and too many expectations!" Therefore, the resilience and expectations of parents regarding the activity performance of their child with severe congenital anomalies of the upper extremities are the issues to be considered.

The limitation of this study was that we could not measure strength and mobility of the case. In addition, the challenges on assessing 1.5 years old baby in relation to the functional level of his upper extremity were also another limitation. However, we believe that the classification of the exercises given from the first week is a unique example of sensorimotor training of the child.

Conclusion

The child was able to participate in the activities according to the stages of his normal motor development as a child in 1 and a half years old. He was able to reach, grip, and release objects though he was not able to perform full opposition with his index finger. He was able to feed himself by his hand and by holding a baby spoon, sit-stand, and walk independently. The role of the physiotherapist in relation to exercise and orthotic interventions can be a feasible practical approach before the surgery.

Ethics Review and Approval

The author(s) disclosed ethics review and approval for the research described in this article: The parents have given their consent for this case study, and they were verbally informed about the procedure and accepted and signed the consent form case studies.

Funding

The author(s) disclosed that they received no financial support for the research, authorship, and/or publication of this article.

Declaration of conflicting interest

The author(s) disclosed no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Acknowledgments

We would like to thank the parents of this case for their permission to share their information. In addition, we would like to thank the manager Ms Kadriye Bulat in Ida Orthopedic, Istanbul, Turkey for their contribution.

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Supplemental material

No supplemental digital content is available in this article.

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