Annals of Clinical Case Reports

Case Report
Published: 11 Dec, 2019

Progress toward Achievement of Early Motor Milestones for a Child with Trisomy 9 Mosaicism: Case Report

*Lisa Rossman Murphy**

Department of Kinesiology and Health, Rutgers University, USA

Abstract

Background: Trisomy 9 Mosaicism (T9M) is a rare genetic disorder, with less than 100 known cases reported since 1973. Studies confirm numerous phenotypic features and anomalies of various body systems and organs. Developmentally, a wide range of function has been reported. This study documents the phenotypic presentation of a female with T9M, describes her progress toward developmental motor milestones, and offers insight for the medical community and caregivers of those affected by this condition.

Case Description: The patient was born premature with low birth weight, had a complicated medical background and presented with global hypotonia and low function. This study follows the patient from 16 to 36 months of age. The patient received physical therapy for 60 min, twice per week until age 24-months when it was increased to three-60 min sessions per week.

Outcomes: The Battelle Developmental Inventory, 2nd Edition (BDI-2) was used to assess gross motor function. BDI-2 scores at 16-months old placed the patient at a newborn level. Throughout the study, the patient contended with medical issues-reflux, hearing impairment, vision impairment. She lived with a very supportive family who wanted her to develop to her potential and understood there would be delays in her achieving motor milestones. After 36-months of age, BDI-2 scores for gross motor skills were at a 9-month level.

Discussion: Although there are many medical factors potentially affecting development in children with T9M, physical therapy with goals to increase postural control, strength, motor control and function may improve social interactions and ultimately quality of life for these patients. Caregivers and clinicians should strive to aid these children in reaching their potential.

OPEN ACCESS

Correspondence:
Lisa Rossman Murphy, Department of Kinesiology and Health, Rutgers University, USA,
E-mail: lisa.rossmanmurphy@rutgers.edu

Received Date: *04 Nov 2019*
Accepted Date: *04 Dec 2019*
Published Date: *11 Dec 2019*

Citation:
Murphy LR. Progress toward Achievement of Early Motor Milestones for a Child with Trisomy 9 Mosaicism: Case Report. Ann Clin Case Rep. 2019; 4: 1766.

ISSN: 2474-1655

Introduction

Trisomy 9 Mosaicism (T9M) is a rare chromosomal disorder in which the entire 9th chromosome appears three times in some cells of the body [1]. Since the disorder was initially described in 1973, over 60 cases have been reported in the medical literature [1]. Presentation of individuals with T9M varies greatly depending on the percentage of cells with the extra chromosome [1,2]. Common features include anomalies of the musculoskeletal, cardiopulmonary, urogenital, and central nervous systems, and developmental delays. Musculoskeletal abnormalities reported include congenital hip dislocation/dysplasia; punctuate cartilage mineralization, calcaneovalgus deformity, joint hypermobility, hypotonia, scoliosis and kyphosis [2-4]. Typically, delays in cognition, motor milestone achievement, speech and self-care are seen [2]. Developmental status data reveal a wide range of function in motor skills. Average age for sitting without support for at least three minutes was 17 months and for walking with bilateral support was 31 months. Some children used a combination of wiggling, scooting or rolling on their back as a means of mobility [2,5]. The purpose of this study is to document the phenotypic presentation of a female with T9M and her progress toward developmental motor milestones from 16 to 36 months of age, while receiving physical therapy.

Patient History

Birth to six months

The patient was born at 36 weeks *via* caesarean section and weighed 4 lbs. 12 oz. She spent 5 days in the NICU, primarily to treat mild jaundice. Due to her small size and some other irregularities, genetic testing was conducted and revealed the patient's diagnosis of T9M, with 7.5% of her cells affected. She presented with craniosynostosis, mild sensorineural hearing loss, and a narrow, high

Table 1: Gross Motor Developmental Status.

Gross Motor Skills	Status				
	Age (mos.)				
	16	22	28	34	36
Hands to midline	Achieved	Maintained	Maintained	Maintained	Maintained
Turns head to left and right	Supine Turns to left or right about 25° from midline	Supine Achieved Supported sitting-Turns to left and right about 60°	Maintained Achieved	Maintained Maintained	Maintained Maintained
Hands to feet in supine	Emerging-Brings hands to knees	Emerging-Brings right hand to right foot	Achieved	Maintained	Maintained (Removes both socks)
Pulls to sit	With slight head lag	Achieved	Maintained	Maintained	Maintained
Lifts head and chest in prone	Emerging-Lifts head 1-2 secs about 20° with assist to maintain prone positioning	Achieved	Maintained	Maintained -Pushes up 2-3 secs on extended arms with assist	Maintained-Pushes up 2-3 secs on extended arms
Roll prone to/ from supine over either side	Emerging-Rolls supine to left side	Emerging-Rolls supine to left side; rolling prone to supine over right side is emerging	Emerging- Rolls prone to spine over right side	Emerging- Rolls prone to supine over right side	Emerging- Rolls prone to supine over either side; supine to prone is emerging
Play with toy in side-lying on either side	Emerging-Plays using left hand in left side-lying	Emerging-Plays using either hand in left side-lying	Achieved	Maintained (Rolls ball reciprocally to another person)	Maintained
Pivot	No	No	Emerging to right in supine	Achieved to right in supine	Maintained-With legs advanced, can bridge and pivot to left
Sitting	Emerging- Ring sitting with support at upper trunk,	Emerging- Ring sitting with support at mid trunk,	Emerging- Short sitting with support at	Emerging- Short sitting with support at sides of	Emerging- Tailor sits briefly with propping
Tolerate sitting in adapted chair	Awaiting device	30 mins/day	1 h/day	1.5-2 h/day	>2 h/day
Tolerates standing in prone stander	Awaiting device	Awaiting device	20 mins/day	25 mins/day	30 mins/day
Maintains quadruped	No	No	No	Emerging	Briefly with support
Bear weight through legs in supported standing	No	Emerging	Achieved- Bears wt briefly at a surface with support through posterior of body and assistance at upper trunk	Maintained- Bears wt for 30 secs at a surface with support at lower trunk/ pelvis	Maintained- Bears wt for one min at a surface with support at lower trunk/ pelvis

Table 2: BDI-2.

Developmental Domain	Age at Testing (in months)	Domain Score (100 is average)	Z Score* (0.0 is average)	Raw Score	Age Equivalent (in months)
Gross Motor	16	55	-3	0	<1
Gross Motor	36	55	-3	24	9

*Represents Standard Deviations (SD) from the norm. In this case, the patient is 3 SD's below the mean score

arched palate but was able to feed by mouth. Her cardiopulmonary system appeared to be normal. She presented with bilateral hip dysplasia which was corrected with bracing. At two months old, she was diagnosed with laryngomalacia. When she was six months old, the patient was diagnosed with seizure activity. She was given Levetiracetam (Keppra) and remained on this drug throughout the study. Multiple EEG tests revealed no subsequent seizure activity.

Ten to sixteen months

At 10 months old, the patient underwent craniofacial surgery to correct the craniosynostosis. Due to a possible tear in the dura during surgery, fluid began to accumulate necessitating shunt placement (subgaleal-peritoneal); the next day, the shunt required revision to subdural-peritoneal. At 12 months old, slow weight gain led to a diagnosis of intestinal malrotation and she was also diagnosed with sleep apnea. From that time through the remainder of the study, she was on BiPAP during nights. At 14 months old, the intestinal malrotation was surgically corrected *via* a Ladd procedure with appendectomy and a G-tube/feeding button was placed. The patient proceeded to vomit often when fed and began to take Ranitidine (Zantac) and then Omeprazole (which she was taking through the end of this study), to treat the reflux. Simultaneously with the Ladd procedure, she underwent a successful supraglottoplasty to correct the laryngomalacia. The patient developed an infection in her abdomen after the procedures, necessitating a month-long hospital stay, during which the subdural-peritoneal shunt was litigated.

Twenty-four to thirty six months

The patient continued to vomit daily until about 24 months old when her parents discovered that by increasing the frequency and decreasing the amount of her feedings, the vomiting decreased, resulting in improved caloric intake, weight gain and endurance. At 30 months old, the patient received new hearing aids and wore them through the remainder of this study. (She had previously worn hearing aids briefly until ten months old when she underwent craniofacial surgery). She received her first pair of glasses to correct for astigmatism and far sightedness at 35 months of age. Throughout the duration of the study, she wore the hip abduction brace sporadically during the nighttime.

Figures 1: The patient at 22 months old.

Table 3: Initial Goals for Six months.

Initial Pt Goals for Six Months (Patient 16-22 Mos. of Age)
Roll prone to-from supine over either side
Play with a toy in side-lying with both hands, on either side
Hold head upright 5 minutes in supported sitting
Reach for a toy; using hands equally
Tolerate upright seating in an adapted chair for one hour per day
Begin standing program using a prone stander

Family

The patient lived in a single family home with her mother, father, older sister and older brother. Her parents initially wanted to see her overall health improve as she was experiencing many medical issues. Motorically, they wanted to see her begin to lift her head when held upright and to develop to her potential, even if hitting milestones according to her own schedule. (Health Insurance, Portability, and Accountability Act-HIPAA- requirements were met throughout the course of physical therapy for this patient and during the preparation of this study. A signed patient consent form was obtained prior to publication of this report).

Clinical impression

Little is known about T9M and its impact on development. The small body of existing research literature reveals a wide range of abilities in children with T9M. This patient presented with global hypotonia and very limited function. The aim of physical therapy was to improve strength and motor control and the patient's abilities toward reaching developmental gross motor milestones. Documentation of the patient's initial presentation and progress throughout the study provides further information to be added to the literature which may aid family members, medical practitioners and other caregivers of children with T9M by increasing their knowledge of the challenges and potential inherent in children with this diagnosis.

Examination

At 16 months old, the patient presented with global hypotonia. She presented with passive range of motion within normal limits, except at her ankles which presented with dorsiflexion on left at 0° to 10° and on right at 0° to 15° and bilateral pronation. In supine, she preferred to hold her hips and knees slightly flexed about 20 degrees at each joint, knee together and rotated to her left. In supine, she could actively rotate her head to left or right about twenty five degrees from midline, could bring her hands to her knees and she could lift her legs slightly off the floor as a pair. Prone positioning was difficult due to the posture of her legs and she could only tolerate it for about 30 seconds. When positioned in prone, with assistance to maintain propping on forearms, she could lift her head for 1 sec to 2 sec at a time. She preferred left side-lying and would feel for nearby toys and grab them with her left hand. The patient presented with decreased awareness of her right arm. When pulled to sit, her head did not maintain alignment with her body. In ring sitting with support at upper trunk, she could lift her head and hold it upright for up to a minute when she would hyperextend her neck and spine, requiring manual correction to resume sitting position. She could tolerate left side-sitting with assistance to maintain the position but showed irritation when positioned in right side-sitting. At this time, the patient could not bear weight through her legs in supported standing or kneeling. She had no means of communication. Her gross motor skills were much delayed (Table 1). The Battelle Developmental Inventory, Second Edition (BDI-2), a norm-referenced tool for assessing developmental milestones, was used to quantify the patient's skills in the gross motor domain (Table 2) [6].

Clinical Impression

The patient was very low functioning, able to lift her head for only 1 sec to 2 sec in prone and for about one minute in sitting with support at her upper trunk. In supported sitting, the patient would hyperextend her spinal and cervical muscles. It was not clear if this was due to a delayed primitive hypertonic extensor pattern or possibly voluntary movement in an effort to return to a supine position of vestibular comfort. Although she was generally hypotonic, there was some muscle shortening of bilateral dorsiflexors and her leg posture was flexed and drifted to her left. She could roll to her left and grab for toys in left side-lying. She had no means of exploring her environment and no means of communication. Goals were written to be achieved within six-month time frames and included tolerance for upright positioning, head control, floor mobility and functional use of hands while incorporating symmetry of body alignment (Table 3).

Intervention

Although there are no studies citing the effects of physical therapy specifically on infants with T9M, research has shown early intervention may improve motor development for the short term, especially for pre-term infants [7-10]. The patient received PT for 60 min, twice per week from 16 months of age (after recovering from the abdominal infection) until 24 months old, at which time the patient's improved general health and endurance made it possible to increase PT to 60 min, three times per week. The focus of therapy sessions was to improve head control and core strength, muscle tone, body awareness, tolerance for positioning, motor planning and ultimately motor function. Strategies employed were positioning, stretching of arms and legs, infant massage, Neurodevelopmental Treatment (NDT), Kinesiotaping* (KinesioTex Gold, Molenstraat 15, 2513BH The Hague, The Netherlands), wearing of a Stabilizing Pressure Input Orthosis (SPIO)† (SPIO, 127 SW 156th St, Burien, WA 98166) vest and AFOs, use of adapted seating and a prone stander, and parent education and training [7-20]. During the time of this study, the patient also received speech and occupational therapies.

Positioning

Tolerance to prone positioning over various surfaces (therapy ball, pillow, foam wedge, PT's legs, the floor) was encouraged with hopes that once the patient could tolerate prone more easily she would begin to push up on her forearms and lift her head, improving head control, trunk strength and upper body strength. The patient preferred to lay on her left side, so right side-lying was promoted. Improved head and trunk control were also addressed using a variety

Table 4: 1st Reassessment of PT Goals for Six Months.

1st Reassessment of PT Goals for Six Months (Patient 22-28 Mos. of Age
Roll prone to/from supine over either side-emerging
Play with a toy in side-lying with both hands, on either side-emerging
Hold head upright 5 minutes in supported sitting-achieved
Reach for a toy; using hand equally-emerging
Tolerate upright seating in an adapted chair for one hour per day-Emerging
Begin standing program using a prone stander-equipment on order
Short sit with support at mid-trunk with head control for 10 minutes-new

Table 5: 2nd Reassessment of PT Goals for Six Months.

2nd Reassessment of PT Goals for Six Months (Patient 28-36 Mos. of Age)
Roll prone to-from supine over either side-emerging
Play with a toy in side-lying with both hands, on either side-achieved
Reach for a toy; using hands equally- achieved
Tolerate upright seating in an adapted chair for one hour per day- achieved
Begin standing program using a prone stander- achieved
Short sit with support at low trunk with head control for 10 minutes-achieved
Sit in adapted chair 2 hours per day-new
Tolerate prone stander at least 10 minutes per day-new
Short sit with support at low trunk with head control for 15 minutes-new
Hold head/trunk upright when standing with upper body support at ball with assist at lower trunk for 5 minutes-new

of positions, including supported ring sitting on floor, supported sitting on a therapy ball with gentle weight shifts, and supported side-leaning, which she could do more easily to her left, so right side-leaning was emphasized. At 22 months old, the patient received a Rifton‡ (Rifton Equpment PO Box 260, Rifton, NY 12471-0260) Hi/Lo Chair and was initially tolerating sitting for 30 min per day. She received bilateral AFO's and at 26 months old began a standing program, using a prone stander. Use of the chair and stander was to benefit her skeletal development and the processes of her cardiopulmonary and gastrointestinal systems. Her parents were taught various ways to position her and were creative in developing their own. For instance, they discovered their daughter enjoyed sitting propped in the corner of the couch where she could better watch and laugh at her siblings' antics.

Stretching and massage

Gentle stretching of all extremities was employed with a goal to maintain passive range of motion, increase range of bilateral ankles and reduce the risk of muscle tightening due to lack of active motion. Areas of focus were bilateral hip flexors, hip rotators, hamstrings and dorsiflexors.

The patient's parents received instruction in stretching and full body massage and the recommendation to provide both daily, to maintain muscle length and to improve proprioception and tone [11-14]. It was also recommended to stretch and massage her legs at every diaper change as a way to incorporate the technique into a frequently recurring routine.

NDT

Facilitation techniques and manual cueing were employed with goals to improve head, neck and trunk control and to facilitate her posture in various positions (i.e. prone, side-leaning). Manual cueing was also used in an attempt to facilitate volitional movements, such as reaching and rolling. Each therapy session was conducted with functional outcomes as the focus [15]. Parents were taught hand placements and muscle cueing reflecting the patient's gross motor status and goals at that time.

Kinesiotaping

Kinestiotape was applied to the patient's abdominal area in an "x" pattern with tape running from origin to insertion of the abdominal with a goal to help elicit a contraction to assist the patient with lifting her lower trunk in supine and with core control for sitting posture [16-19]. The tape was worn for three days at a time with breaks of a few days in between wearing.

SPIO vest

The patient wore a SPIO vest in an attempt to improve core muscle activation and stabilization. When she was 17 months old, it was worn for short periods in between feedings. At 24 months of age, she wore the vest during all therapy sessions and by 27 months she was wearing it during the day except for feedings [20].

Parent education

Therapy sessions were conducted following a parent teaching model with techniques and strategies explained and taught and parents providing return demonstration. Documentation and written instructions were given to parents describing strategies/techniques to employ for carryover until the following session.

Outcome

The patient made slow but steady progress throughout her time in physical therapy. Goals were reassessed at six-month intervals (Table 4 and 5) and gross motor skills throughout the study time frame were documented (Table 1). The patient's BDI-2 scores at 36 months of age, in comparison to those at 16 months, reveal the patient gross motor skills progressed from a newborn level to that of a nine month old (Table 2). The patient showed an increase for tolerance of various positions. She initially could only tolerate prone for about 30 sec, could not tolerate right side-lean, had very little head control in supported sitting and could not bear weight through her legs. By the end of the case study, she could more easily tolerate prone, could side-lean with assistance to either side and could bear weight through her legs in supported standing. Her head control and postural strength had improved so that she was able to tolerate kneeling on her mom's lap and push up on extended arms on her mother's chest to look out of the living room window for several minutes. She had improved her tolerance to upright positioning and could sit in the adapted chair for more than two hours daily and tolerate the prone stander for 30 min daily. Episodes of pushing back into extension were no longer present. She achieved some motor milestones-rolling to side-lying and using hands for play, turning head fully to the either side in a variety of positions, removing socks, and rolling prone to supine. Most importantly, she discovered a way to maneuver on the floor toward items of interest and simple methods of non-verbal communication. She could wave "bye bye," shake her head, "No," and would tap the back of her hand to her chin indicating, "Yes," or, "More." All of these new skills enabled her to explore her environment, to communicate with her family and others and to gain some control over her situation and surroundings.

Discussion

There are relatively few documented cases of T9M. Existing literature reveals a range of ages for achieving motor milestones, i.e.

rolling prone to supine from 19 to 66 mos; sitting without support from 10 to 22 mos (Figure 1) [2,5]. This patient achieved the former at 32 mos. but had yet to achieve the latter milestone. It should be noted that this patient had extensive medical complications and feeding issues that affected her overall health, endurance and ability to have therapy. Also, her BDI-2 Domain scores and Z scores did not change from 17 mos. to 36 mos. of age; however, the BDI-2 is a norm-referenced assessment tool and although relative to other typically developing three year old this patient's scores represented severe delay, she made noteworthy gains and her progress was consistent. Age equivalents revealed she made progress from the gross motor level of a newborn to that of a 9-month old. Perhaps a more sensitive assessment tool, with a breakdown of components of early motor milestones (i.e. reaching across midline while flexing lower trunk, or flexing and rotating lower trunk in supine to initiate rolling) might have better underscored her progress and achievements. This patient tended to push back into extension when positioned upright until she was about 32 months. Through a combination of increasing weight bearing in prone positioning and the subsequent possible neuroplasticity, increasing the strength of flexor musculature and conditioning her vestibular system to the upright position, she overcame this tendency. Further research might investigate if a preference for cervical and spinal hyperextension is common among children with T9M. Also significant is that the patient's parents discovered that by decreasing the quantity and increasing the frequency of her feedings, she vomited less, improving her health, endurance, and tolerance for prone positioning. Future research might further investigate the effect of reflux on progress toward motor milestones in patients with T9M.

Acknowledgement

Thank you to the patient and her family for participation in this study.

References

1. National Organization for Rare Disorders. [Internet]. Bethesda: Mosaic Trisomy 9.
2. Rare Chromosome Disorder Support Group [Internet]. Oxted: Trisomy 9 Mosaicism.
3. Arnold GL, Kirby RS, Stern TP, Sawyer JR. Trisomy 9: Review and report of two new cases. Am J Med Genet. 1995;56(3):252-7.
4. Miryounesi M, Dianatpour M, Shadmani Z, Ghafouri-Fard S. Report of a case with trisomy 9 mosaicism. Iranian J of Med Sci. 2016;41(3):249-52.
5. Bruns DA, Campbell E. Twenty-five additional cases of trisomy 9 mosaic: Birth information, medical conditions, and developmental status. Am J Med Genet. 2015;167A(5):997-1007.
6. Newborg J. Battelle Developmental Inventory. 2nd ed. Boston, Houghton Mifflin Harcourt, MA, USA; 2004.
7. Blauw-Hospers CH, Dirks T, Hulshof LJ, Bos AF, Hadders-Algra M. Pediatric physical therapy in infancy: From nightmare to dream? A two-arm randomized trial. Phys Ther. 2011;91(9):1323-38.
8. Hughes AJ, Redsell SA, Glazebrook C. Motor Development Interventions for Preterm Infants: A systematic review and meta-analysis. Pediatrics. 2016;138(4):e20160147.
9. Lekskulchai R, Cole J. Effect of a developmental program on motor performance in infants born preterm. Aust J Physiother. 2001;47(3):169-76.
10. Spittle A, Orton J, Anderson PJ, Boyd R, Doyle LW. Early developmental intervention programmes provided post hospital discharge to prevent motor and cognitive impairment in preterm infants. Cochrane Database Syst Rev. 2015;11:CD005495.
11. Vickers A, Ohlsson A, Lacy JB, Horsley A. Massage for promoting growth and development of preterm and/or low birth-weight infants. Cochrane Database Syst Rev. 2000;2:CD0000390.
12. Badr LK, Abdallah B, Kahala L. A me`ta-analysis of pre-term infant massage: An ancient practice with contemporary applications. Am J Matern Child Nurs. 2015;40(6):344-58.
13. Choi H, Kim SJ, Oh J, Lee MN, Kim S, Kang KA. The effects of massage therapy on physical growth and gastrointestinal function in premature infants: A pilot study. J Child Health Care. 2016;20(3):394-404.
14. Juneau AL, Aita M, Heon M. Review and critical analysis of massage studies for term and pre-term infants. Neonatal Netw. 2015;34(3):165-77.
15. Raine S. The current theoretical assumptions of the Bobath concept as determined by the members of the BBTA. Physiother Theory Prac. 2007;23(3):137-52.
16. Lee YJ, Kim JY, Kim SY, Kim KH. The effects of trunk kinesio taping on balance ability and gait function in stroke patients. J Phys Ther Sci. 2016;28(8):2385-8.
17. Iosa M. The application of Kinesio Taping in children with cerebral palsy. Dev Med Child Neurol. 2015;57(1):11-2.
18. Kara OK, Usal SA, Turker D, Karayazgan S, Gunel MK, Baltaci G. The effects of Kinesio Taping on body functions and activity in unilateral spastic cerebral palsy: A single-blind randomized controlled trial. Dev Med Child Neurol. 2015;57(1):81-8.
19. Iosa M, Morelli D, Nanni MV, Veredice C, Marro T, Medici A, et al. Functional taping: A promising technique for children with cerebral palsy. Dev Med Child Neurol. 2010;52:587-89.
20. Hylton N, Allen C. The development and use of SPIO Lycra compression bracing in children with neuromotor deficits. Pediatr Rehabil. 1997;1(2):109-16.