



The Effectiveness of Physical Therapy Interventions for a Six-Year-Old Boy with Monosomy 18p

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BACKGROUND & PURPOSE

Monosomy 18p is a rare chromosomal disorder that presents with short stature, motor developmental and speech delay, mild to moderate intellectual disability, and characteristic facial features, affecting about one per fifty thousand births.¹ There is no specified physical therapy treatment in the literature for patients with Monosomy 18p.¹ However, more common diagnoses with similar impairments, such as, hypotonic cerebral palsy (CP) can be used as a model for treatment of rare diagnoses, like Monosomy 18p.

The purpose of this case report is to explain the utilization and effectiveness of physical therapy interventions for improving gross motor abilities in a six-year-old boy diagnosed with Monosomy 18p.

CASE DESCRIPTION

Health Condition
Monosomy 18p

Body Functions/Structures
Global hypotonia
Hypermobility joints
Lower extremity muscle tightness
Generalized weakness
Decreased endurance
Repeated tactile self-stimulation

Activity (limitations)
Limited mobility beyond rolling, prone, and transferring supine to sit
Decreased ability to maintain standing, kneeling, and quadruped postures without support

Personal Factors
6 year old male
Born at 36 weeks
Craniosynostosis s/p repair
Non-verbal

Participation Restrictions
Inability to play with his friends on the playground
Unable to explore his environment

Environmental Factors
Family of low socioeconomic status
Not enrolled in school

INTERVENTIONS

Stretching
Gastrocnemius
Soleus
Hamstrings

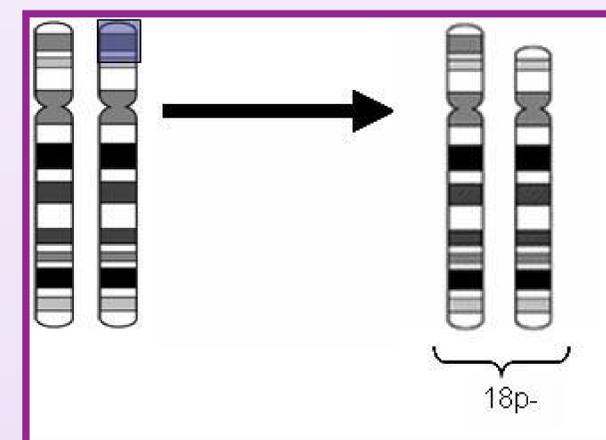
Static Postures
Quadruped
Standing

Functional Transitions
Rolling to the left
Sit to stand

Dynamic Balance
Sitting on therapy ball

CLINICAL RELEVANCE

Due to the amount of rare diagnoses that affect the pediatric population, it is a viable solution to use model diagnoses to guide physical therapy interventions for less common diagnoses that present with similar impairments. Using hypotonic CP as a model diagnosis, this case demonstrated that the utilization of physical therapy can benefit children with Monosomy 18p.



<https://www.chromosome18.org/18p/>

RESULTS

Gross Motor Function Measure-66

Initial Evaluation
33.4*

6 weeks

Discharge
37.5*

*Percent scores

12.3% change

REFERENCES

