Functional School-Based Physical Therapy Management for a Child with Pallister-Killian Syndrome: A Case Report

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Background & Purpose

- Pallister-Killian Syndrome (PKS) is a rare genetic disorder caused by an additional short arm in chromosome 12.
- PKS affects multiple systems, which can impact a child’s development. (Figure 1)
- Common clinical manifestations include: hypotonia, visual impairment, hearing loss, coarse facial features, intellectual disability, and congenital heart defects.
- Improvements in gross motor function have resulted from physical therapy (PT) and rehabilitation involving neurodevelopmental treatment (NDT).
- Research is limited on the effects of PT interventions to promote standing and ambulation for children with PKS.

The purpose of this case report was to describe school-based PT interventions for a child with PKS, which included standing exercises, body-weight support treadmill training (BWSTT), overground gait training, and a standing program.

Timeline

2 Years Prior to Initial Evaluation:
- Started school and received PT, OT, speech therapy, and vision therapy
- Presented with hypotonia, developmental delay, auditory impairment, and visual impairment
-Received PT 5x/week involving NDT and additional interventions

Relevant Past Medical History:
- Diagnosed with Pallister-Killian Syndrome at 20 weeks gestation
- At 3 months, MRI revealed left germinal matrix hemorrhage
- At 8 months, received early intervention for delayed gross motor skills
- At 2 years old, MRI revealed mild ventriculomegaly

Week 1:
- Child presented with delayed gross motor skills
- BWSTT (2 reciprocal steps taken) Standing exercise (0:16)

Week 2:
- Overground gait training initiated (8 reciprocal steps)
- No significant improvements in standing time reported

Week 3:
- New solid AFOs
- Significant improvements during BWSTT (83 reciprocal steps)
- Standing exercise (best trial = 1:04)

Week 4:
- New solid AFOs
- Significant improvements during BWSTT (83 reciprocal steps)
- Standing exercise (best trial = 3:05)
- Overground gait training (8 reciprocal steps)

Week 5:
- Absent for 1 week due to ear infection

Week 6:
- Significant improvement during standing exercise (best trial = 3:05)
- Overground gait training (8 reciprocal steps)
- Child continued to receive physical therapy 5x/week

Interventions

- Overground gait training
- BWSTT
- Overground gait training (0 steps to 63 steps) overground gait training (0 steps to 6 steps)

Case Description

- The child was a 7-year-old male who received PT 5 days a week in school.
- Past medical history included hypotonia, global developmental delay, congenital hip dysplasia, atrial septal defect, cortical visual impairment, hearing loss, and oropharyngeal dysphagia.
- The child used bilateral solid ankle-foot orthoses (AFOs), adaptive glasses, and hearing aids.
- The child was able to ring sit independently, stand with moderate assistance, and ambulate with maximum assistance.
- The child’s level of function was classified as Gross Motor Function Classification System (GMFCS) Level V.

Outcomes

- Improvements in standing time (0:16 seconds to 3:05 minutes) were evident after 6 weeks. (See above)
- Improvements in the number of reciprocal steps were seen during BWSTT (3 steps to 63 steps) overground gait training (0 steps to 6 steps).
- Observational posture and strength in standing and ambulation were additionally seen.

Discussion & Conclusion

- A strength of this case report included the unique nature of the child’s diagnosis and clinical presentation.
- The child missed 1 week of school due to an ear infection, which lead to a total of 5 weeks of PT.
- The child was unable to tolerate the standing program.
- A standing exercise, BWSTT, and overground gait training may improve gross motor skills for children with PKS.
- Further research is warranted on the benefits of these interventions to elicit improvements in function and declines in disability for children with PKS.

References


Contact Information

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