Physical Therapy Management with Emphasis on Quadriceps Strength Training for Chronic Patellar Dislocations in a Female Adolescent with Kabuki Syndrome: A Case Report

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Background

• Kabuki Syndrome (KS) has a current estimated prevalence at 1 in 32,000.
  • KS is a rare, but highly recognizable genetic disorder manifested in the craniofacial, skeletal, neurological, and cardiovascular systems/regions of the body.

• Patellar dislocations are common in children with KS due to the presence of hypotonia causing muscle imbalances and joint laxity.
  • Most commonly, treatment of patellar dislocations are done conservatively through exercise. Surgical interventions (lateral release, medical imbrication, medial patellofemoral ligament reconstruction, or bone realignment) are considered with continued dislocations.

Purpose

Physical therapy management approach with a focus on addressing chronic patellar dislocations through quadriceps strengthening in a 12 year-old female diagnosed with Kabuki Syndrome.

Case Description

• 12 year-old female in middle school diagnosed with Kabuki Syndrome.
  • Chronic patellar dislocation occurring bilaterally
  • Functional limitations and impairments in strength, range of motion, gait, balance, and functional squatting in the lower quarter.
  • Procedural interventions included therapeutic exercises with an emphasis on functional strengthening of the quadriceps. Functional exercises specific to the patient’s everyday tasks and demands in and out of school activities were chosen.
  • Patient was treated in an outpatient orthopedic clinic for 12 sessions spanning a 6 week period.
  • Knee Outcome Survey (Activities of Daily Living and Sports Activities Scale) was administered at admission and discharge.

Examination

• Decreased bilateral knee flexion
  • Impaired lower extremity strength
  • Bilateral foot pronation, calcaneal valgus (supported with orthotics); genu valgum and squinting knees
  • Unable to participate in all school gym classes
    • Generalized hypotonia
    • Toe-out gait & increased sway in balance

Outcomes

Following 12, hour long treatment sessions spanning a 6 week period, the following outcomes were observed:

Sample Interventions

PT focused on functional quadriceps strengthening involving a spin bike, GTS total gym, and agility ladder. Each week the resistance was increased as long as there was no report of pain and proper form was used.

Discussion

Performance of functional strengthening exercises over 6 weeks of physical therapy, and in a blocked practice format improved: Strength and range of motion in all muscles and joints tested, and increased percent function as seen by her results on the Knee Outcome Survey (KOS).

Using the KOS as a reference, she demonstrated improvements in: the presence of pain, ascending and descending stairs, squatting, stopping and starting quickly, jumping, and cutting and pivoting on either leg.

Future research is needed on the recurrence of patellar dislocations among individuals with KS following PT.