Pediatric Genetic Conditions

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Basics & Characteristics

Prader-Willi Syndrome
- Most common cause of syndromic obesity (Irizarry et al, 2016)
- Growth hormone therapy is an alternative treatment shown to improve outcomes (Irizarry et al, 2016)

Angelman Syndrome
- Characterized by developmental delay, ataxia, happy demeanor and excessive laughter (Williams, Driscoll, Dagli, 2010)

Chromosome 18q
- Epilepsy is frequent and common characteristic of children who have chromosome 18 abnormalities (Verrotti et al, 2015)
Prader-Willi syndrome (PWS) (Irizarry et al, 2016)

Affects chromosome 15q11.2 to 13 with unknown genetic mechanism

Clinical characteristics vary with age

- Hypotonia
- Failure to thrive in infancy
- Excessive weight gain
- Obsession with good
- Short stature
- Cognitive impairment resulting in learning delays
Treatments for PWS (Irizarry et al, 2016)

- Growth hormone therapy
- Physical therapy
- Occupational therapy
- Speech therapy
- Psychiatric therapy
Angleman Syndrome  (Williams, Driscoll, Dagli, 2010)

Affects chromosome 15q11.2-q13 with variable phenotype
- Similar mutation to PWS

Microcephaly and seizures are common

Clinical features do not occur until after age 1 year

Challenging diagnosis to make because of delayed clinical features

Speech impairment or absence of speech is key in diagnosis

Cardiac abnormalities can be co-morbid
Treatment for Angelman Syndrome

Physical Therapy (Williams, Driscoll, Dagli, 2010)

Occupational Therapy (Williams, Driscoll, Dagli, 2010)

Speech Therapy (Williams, Driscoll, Dagli, 2010)

Management of gastrointestinal dysfunction (Williams, Driscoll, Dagli, 2010)

Medication for seizure management (Williams, Driscoll, Dagli, 2010)

Assistive Technology (Radstaake et al, 2012)
Chromosome 18 Abnormalities (Cody & Hale, 2015)

Affects 18p and 18q genes with variable presentations

Translocations, ring, and trisomy are all genetic mechanisms that impact chromosome 18

High risk of seizures in all types

Malformations of the skull

Clubfeet

Eye defects with visual abnormalities

Cardiac abnormalities can be present

Kidney dysfunction is common
Treatment for Chromosome 18 Abnormalities
(Verrotti et al, 2015)

- Medication for seizure management
- Hemispherectomy for seizure management
- Medication for gastrointestinal dysfunction
- Speech therapy
- Physical therapy
- Occupational therapy
**Parent’s views on genetic conditions**


Families report high levels of stress associated with caring for a child with a rare genetic condition.

Parents report that not one specific coping strategy is successful in caring for a child with a rare genetic condition.

Community support and timely information about the diagnosis and treatment were most helpful in coping with the condition.
References


References


Disclaimer

*Effective April 29, 2015, the Doctor of Physical Therapy Program at Briar Cliff University has been granted Candidate for Accreditation status by the Commission on Accreditation in Physical Therapy Education (1111 North Fairfax Street, Alexandria, VA, 22314; phone: 703-706-3245; email: accreditation@apta.org). Candidate for Accreditation is a pre-accreditation status of affiliation with the Commission on Accreditation in Physical Therapy Education that indicates that the program is progressing toward accreditation and may matriculate students in technical/professional courses. Candidate for Accreditation is not an accreditation status nor does it assure eventual accreditation.*