Treatment of Dysphagia in Children with Rare Disorders:

Problem Solving, Collaboration and Treatment Planning in the Unknown

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Introduction

- In practice for 10 years
- Clinical experience in every area of speech-language pathology (thanks to Covid-19 this now includes telehealth!)
- Currently work at a multidisciplinary private practice in Jackson, MI
- No relevant disclosures to share
- Informed waiver of consent was obtained from all patients that served as case studies for today's seminar.

Learning Objectives

- To understand the associated challenges when treating dysphagia in children with rare disorders
- To know where to obtain reliable information on rare disorders
- To develop treatment planning ideas for diagnoses when we have little research or established practices to draw from
- Identify other professionals on the treatment team with whom to collaborate and/or recommend their services

Agenda

- Definition of a rare disorder
- Reliable sources on rare disorders
- The challenges when working with children who have rare disorders
- Presentation of case studies
- Assessment by answering the following questions in the chat box:
  - What do you think the rare disorder is in the child's medical history?
  - Who do you think is on the treatment team?
  - What are your treatment planning ideas for this child?

What is a rare disorder?

According to Genetic and Rare Diseases Information Center (GARD):

In the United States, a rare disease is defined as a condition that affects fewer than 200,000 people in the US.

Genetic and Rare Diseases Information Center (GARD)

Website: https://rarediseases.info.nih.gov/

- A program of the National Center for Advancing Translational Sciences (NCATS) and funded by two parts of the National Institutes of Health (NIH).
- Provides the public with access to current, reliable, and easy to understand information about rare or genetic diseases
- Information is available in English and Spanish
- Diseases are alphabetically organized each with its own web page including information, resources, clinical studies and treatments
- Information is draw from a variety of reliable sources
- If you cannot find the information you are looking for you can contact a GARD information specialist

Orpha.net

- The portal for rare diseases and orphan drugs
  - www.oprha.net
- Established in France by INSERM (French National Institute for Health and Medical Research)
- Gathers and organizes information on rare disorders
  - Clinical signs and Symptoms
  - Classification
  - Genes
  - Disability
  - Encyclopaedia for patients and professionals

National Organization for Rare Disorders (NORD)

- https://rarediseases.org/
- A 501(c)(3) patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them
- Rare disease information
- Resources for patients and families
- Resources for organizations, clinicians and researchers
- Rare disease advocacy on local, state and national levels

General Challenges in Treating Pediatric Dysphagia

- Pediatric feeding and swallowing is in its infancy therefore there is a lack of research and evidence based practice
- Multidisciplinary care is essential. Not all settings allow for easy collaboration with other members of the medical team
- High levels of paternal/caregiver stress; as professionals we are susceptible to taking on that stress
- Most clinicians do not feel adequately trained in their graduate programs to treat pediatric dysphagia
  - Rely on CEUs and mentorship which is not always easily accessible

Challenges in Treating Children with Rare Disorders

Bavisetty et. al (2012) examined steps taken in US and Europe to meet the healthcare needs of children with rare disorders

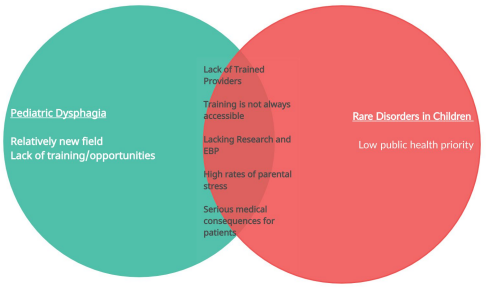
- In the United States, we do not have any national surveys aimed specifically at children with rare diseases.
- In the US, only recently have rare disorders become a public health priority
- 1983 Orphan Drug Act
  - Previously, pharmaceutical companies had little market incentive to develop treatments for patients with rare diseases.The act provides tax benefits and grants for testing and market exclusivity of treatments

Challenges in Treating Children with Rare Disorders

- Valdez et. al (2016) outlined the challenges for public health providers when treating rare diseases
  - Diagnoses are difficult and delayed
  - Case definitions for surveillance are lacking
  - ICD-10 codes are lacking or poorly defined
  - Underlying molecular or physiologic mechanisms are unknown
  - Specialized and coordinated medical care is in short supply; treatments can be complex
  - Standards of care for treatment and rehabilitation are not evidenced-based because of lacking research done on a small scale

Challenges in Treating Children with Rare Disorders

- Longitudinal data collection is scarce
- Development of new drugs and treatment is fragmented and slow
- Screening strategies lack efficiency
- Scope and capacity of most registries and databases are limited
- Caregivers face unique challenges
  - Lack of ways to measure their needs which include: understanding the rare disorder; working with health professionals; emotional issues; financial needs
- Most research on caregiver needs was conducted outside of the United States



Case Study #1: J Birth and Early Feeding History

- 4 month 11 day old male; adjusted age 3 months 11 days, possibly 2 months 14 days
- Born at 32 weeks, but may have been 29 weeks . This is unknown due to poor prenatal care.
- J has been in the care of his foster family since birth. His foster mother has cared for other infants with dysphagia.
- **Parental Concerns:** poor latch; prolonged feeding times; poor PO intake requiring supplemental NG tube feeding; history of aspiration and resultant pneumonia; poorly coordinated suck-swallow-breath; poor weight gain.

J Medical History

- Premature birth
- Low birth weight: 3 lbs 3 ounces, dropping to 2 lbs 14 ounces the next day
- Exposure to narcotics and other drugs in utero
- 62 day NICU stay after birth: 7 failed attempts at weaning from CPAP
- Bronchopulmonary dysplasia (BPD)
- Chronic Lung Disease (CLD)
- Heart murmur
- Poor GI motility
- Sandifer Syndrome
- Hypertonia
- Cerebral Palsy
- Medications: Diuril (FM feels this prevents recurrent pneumonia due to aspirating saliva); Prevacid
- MBSS 2 weeks prior to evaluation revealed: weak latch; poorly coordinated suck-swallow-breath; delayed swallow initiation; aspiration of thin via level 2 nipple.
- Recommended Diet: IDDSI Level 2 thickened with rice cereal via Level 2 nipple; positioned upright for all feeding

J Medical History

- Outpatient Evaluation Findings:
  - Beckman Oral Motor Evaluation Protocol revealed:
    - Reduced strength and ROM of the upper lip
    - Reduced buccal ROM on the left and right
    - Lingual ROM WNL
    - Reduced durational jaw movement
  - Poorly coordinated suck-swallow-breath; audible suck
    - 3-4 sucks; long pause where formula would pool in the mouth
  - Postural stability: hypertonicity; arching the back
  - Supplemental Nutrition via NG tube which FM is trained to place and remove as needed
  - Expresses hunger, eagerly accepts bottle

Sandifer Syndrome

Orpha.net's Definition: Paroxysmal dystonic movement disorder occurring in association with gastro-oesophageal reflux, and, in some cases, hiatal hernia.

Clinical Description:

- Onset usually occurs during infancy or early childhood.
- The dystonic movements are characterised by abnormal posturing of the head and neck (torticollis) and severe arching of the spine. The theory is this is to seek relief from the pain associated with GERD.
- Episodes usually last for between 1-3 minutes and can occur up to 10 times a day, although they are usually associated with the ingestion of food.

Sandifer Syndrome and Feeding

- A literature review by Mindlina (2020) revealed Sandifer Syndrome is often misdiagnosed because it initially presents more neurologically (ie., convulsion, seizure) with minimal GI symptoms.
- Sdravou et. al (2019) conducted a literature review examining 20 studies to learn more about the feeding skills of children with diseases of the upper gastrointestinal tract:
  - Several clinical studies reveal that children with gastrointestinal diseases, **especially those with GERD** and eosinophilic esophagitis, are more likely to present with feeding problems that have a negative impact on development, growth, and psychosocial dynamics.

Sandifer Syndrome and Feeding

- Postural changes impact oral and pharyngeal phase
  - Think about how sudden arching of the spine and hyperextension of the neck would affect a child's labial seal, bolus containment and postural stability while feeding.
  - J presented with limited labial ROM (rigid, tight upper lip) when experiencing dystonic movements. This impacted his labial seal resulting in increased effort and fatigue during feeds.

Case Study #1: J's Treatment Team

- **Gastroenterology (GI)**
  - The first priority in treating pediatric dysphagia is managing any underlying medical conditions.
  - For J, this was addressed with medication, a formula change, and a recommendation to remain upright during and after feeds.
  - Infants who have GERD often develop aversions to eating. They begin to associate pain with eating.
- **Physical Therapy/Occupational Therapy**
  - Managing hypertonicity

Case Study #1: J's Treatment Team

- **Speech-Language Pathology**
  - Diet modifications
  - Bottle and nipple recommendations
  - Positioning to minimize risk of aspiration, GERD symptoms and maximize efficiency
  - Recommending repeat instrumental exam when clinically indicated
- **Registered Dietitian-specializing in pediatrics**
  - Formula to meet nutritional needs for growth and minimize GERD symptoms (i.e, hypoallergenic formula such as Elecare)

Case Study #1 J: Treatment Planning

- Oral motor exercises to promote increased labial and buccal range of motion
  - Beckman Oral Motor Interventions
- Parent Education
  - Importance of following through with MBSS recommendations: thickening, bottles, nipples and positioning
  - Importance of not changing modality, consistency or positioning without repeat MBSS
  - Signs and symptoms of aspiration
  - Impact of GERD on feeding; importance of making sure GERD is well managed
  - Feeding development
  - Impact of multiple medical diagnoses on feeding

Case Study #1 J: Treatment Planning

- Recommending repeat instrumental examination as indicated
- Trialing nipples and bottles to maximize safety and efficiency
  - Often nipples and bottles are recommended per MBSS findings. These recommendations should be followed unless the child is struggling with them. An instrumental examination is essential in guiding treatment, but it is snapshot of child that is medically complex and changing all the time.
- Compensatory strategy training including positioning to maximize safety, efficiency and minimize GERD and aspiration
- Interprofessional collaboration
- Constantly monitoring all aspects of medical status: GI, respiratory, growth, nutrition

Case Study #2 R Birth and Early Feeding History

- 2 year old female
- Delivered full term without complication
- Frequently hospitalized as an infant with projectile vomiting and diarrhea
- **Parental Concerns:** Not chewing and swallowing developmentally appropriate textures and consistencies; chews and then spits food out; continues to drink from a bottle; poor growth/weight gain

R Medical History

- Severe torticollis as an infant
- Malformation on the right side of the jaw
  - No hx of trauma to the jaw; questioning impact of torticollis on jaw development
- Delayed fine motor skills
- Sensory processing concerns
- Food Protein Induced Enterocolitis Syndrome (FPIES)
- Food allergies
  - Rice, soy, sweet potato, bananas, berries, dairy
- PE tube placement
- No history/parental report of pharyngeal phase dysphagia

R Medical History

Outpatient Evaluation Results:

- Poor tongue-tip lateralization resulting in poor bolus formation and mastication
- Reduced durational jaw movement and difficulty chewing on the right side of the mouth, where jaw is malformed resulting in R frequently spitting out partially chewed pieces of food
- Parents reported the following behaviors while eating: turning head away; throwing bowl; aversion to being seated in feeding chair
- R' s cognition, speech and language development were within normal limits.

Food Protein Induced Enterocolitis Syndrome (FPIES)

NORD's clinical description of FPIES includes:

- Allergic reaction to food that affects gastrointestinal system
- Profuse vomiting and diarrhea within 2-6 hours of ingesting offending food
- Symptoms can be severe and cause acute dehydration and hypovolemic shock. Hypovolemic shock is a condition where the plasma is too low in the blood, the organs do not get enough blood and oxygen.
- Rice, milk and soy are most common triggers, but can be caused by a wide variety of food proteins. (Refer to R's very random list of food allergies)
- May outgrow by age 2 or3, in some cases allergies persist
- Exact underlying cause is unknown
- Genetic component is unknown

FPIES

- Due to low incidence, the estimate in the United States is 0.28%, of the general population, FPIES is often misdiagnosed or diagnosis is delayed
- Atopic conditions such as atopic dermatitis, asthma or hay-fever
- Genetic component is unknown
  - R's 7 week old sister was demonstrating many s/s of FPIES
- R was not diagnosed until she was 9 months old, after multiple hospitalizations and FPIES episodes
- Diagnosed by allergist-immunologist

FPIES and Feeding

Chehade et. al (2019) reviewed the signs and symptoms of children with non-IgE-mediated (reactions are delayed and involve other parts of the immune system) food allergic gastrointestinal disorders and intervention strategies for these children presenting with feeding difficulties

- Common feeding problems associated with FPIES:
  - Fear of eating
  - Selective eating
  - Restricted diet can lead to limited exposure to a range of flavors and textures resulting in impaired oral motor skills and sensory processing related to food

FPIES and Feeding

  - Caregivers can have difficulty facilitating and modeling fun,positive interactions with food as the result of anxiety and vigilance around food
- Elements of the following therapy approaches used:
  - Ellyn Satter's Division of Responsibility (sDor)
  - Food chaining : further studies with this population required
  - Sequential Oral Sensory (SOS): anecdotal evidence of positive outcomes; further research is needed.
  - Multidisciplinary approach: many studies published on efficacy of multidisciplinary treatment in children with feeding difficulties, including those with FPIES.
    - Multifactorial problems require multidisciplinary solutions

Case Study #2: R's Treatment Team

- Allergist-Immunologist
  - Throughout episode of care R was able to expand her food inventory under the guidance of her allergist. Only foods approved by the allergist were introduced in therapy. All foods were brought from home.
- Gastroenterology
  - Manage constipation and other GI issues
- Occupational Therapy
  - Sensory processing issues and impact on feeding
- Speech Language Pathology
  - Oral Motor skills
  - Sensory food exploration/feeding therapy

Case Study #2: R's Treatment Team

- Behavior Analyst and SLP
  - Consulted regarding transition from bottle
    - Drank from a straw and open cup in treatment
    - Would tantrum, become aggressive with mother and siblings when offered developmentally appropriate cups at home
    - This was very important as pediatric dental surgery and plastic surgery were being consulted regarding malformation of jaw; would not be a candidate for surgery until she was off of bottle and pacifier
- Registered Dietitian
  - Hypoallergenic formula for nutritional needs
  - Meeting nutrition needs within allowed foods

Case Study #2: R's Treatment Team

- Pediatric Dentistry, Dental Surgery, Plastic Surgery
  - Unrelated to FPIES, malformation of right side of jaw impacted mastication
  - Plastic surgeon did not believe torticollis impacted jaw development; thought it was more likely the result of unknown trauma.
    - This information was not well received, mother felt accused
  - **Interesting areas for further research:**
    - Correlation between FPIES and torticollis? Similar to dystonia seen in Sandifer Syndrome?
      - R was my patient prior to J. I was not aware of the association of dystonic movements and GI symptoms.
      - Correlation between severe torticollis and jaw development?

Case Study #2 R: Treatment Planning

- Oral motor exercises and activities to promote lingual lateralization and durational jaw movement for improved chewing and bolus management
- Compensatory strategy training: chewing on left side due to malformation on the right side of the jaw
- Parent Education:
  - Impact of FPIES and GI issues, constipation on feeding
  - All foods had to be brought from home and approved by allergist
- Messy play and food exploration within allowed foods
- Collaboration with OT regarding R's sensory processing needs to optimize success in feeding therapy
- Working with SLP-BCBA to manage behaviors associated with transition from bottle

Case Study #3 B Birth and Early Feeding History

- 23 month old male
- Initial evaluation completed by another clinician; I began working with him within 2 months
- Born at 41 weeks at home but hospitalized for breathing difficulties and failure to thrive
- **Parental Concerns:** Limited oral nutrition, oral aversions and PEG tube dependence

Case study #3 B: Medical History

- G-tube dependence; 5 ml of water by mouth via syringe two times a day
- Rubinstein-Taybi syndrome (RTS)
- Eosinophilic Gastrointestinal Disease (EGID) - food allergies
- Significant global delays in communication, pre-linguistic skills, gross and fine motor development; sensory processing
- Oral aversion (especially to syringe of water)
- Ankyloglossia (tongue tie) corrected with lingual frenectomy
- Oropharyngeal dysphagia. MBSS completed prior to outpatient evaluation revealed silent aspiration, penetration of mildly thick consistencies, and intermittently delayed swallow initiation. (IDDSI Level 4) and moderately thick liquids (IDDSI level 3)

Case Study #3 B: Medical History

Outpatient Evaluation Results:

- Labial closure and turning head away, when presented with puree at his lips, consistent with parental report of oral aversion.
- Accepted puree to lips one time using a tongue pumping motion to retrieve trace amount.
- High, arched hard palate observed.

Rubinstein-Taybi Syndrome

NORD's clinical description of Rubinstein-Taybi Syndrome:

- Rare Genetic Disorder affecting many organ systems
- Growth Delays
- Craniofacial Dysmorphism
  - Straight nose with a broad nasal bridge; downslanting eyelid openings
- Cognitive impairment with average of IQ of 36-51
- Feeding difficulties/dysphagia
- Breathing difficulties
- Prone to frequent upper respiratory illness

Rubinstein Taybi Syndrome

- Abnormalities of the mouth and jaw including : high, arched palate, underdeveloped mandible, maxilla, thin upper lip, micrognathia (abnormally small lower jaw); retrognathia (lower jaw is displaced further back)
- Broad, angulated thumbs and great toes
- Hypotonia
- Hyperreflexia
- Gait problems
- Constipation
- One third of infants diagnosed with RTS have a congenital heart defect
- Abnormalities of the respiratory system: lung lobulation (lungs divided into extra, small sections); walls of larynx may be weak and collapsible; sleep apnea
- Male infants with RTS may have abnormalities of the genitourinary tract

Rubinstein Taybi Syndrome and Feeding

Noble et. al (2007) completed a case study of two males with Eosinophilic Gastrointestinal Disease and Rubinstein Taybi Syndrome

- Case study of two older children with RTS who had EGID that presented as dysphagia.
- They diagnosed the EGID, managed it with dietary changes and medication and the dysphagia resolved.
  - Arvedson & Brodsky (2002) list Rubinstein Taybi Syndrome as a genetic syndrome associated with dysphagia in their text *Pediatric Swallowing and Feeding: Assessment and Management*
    - Infants have a weak suck; swallowing is poorly coordinated; frequent vomiting during infancy

Case Study #3 B: Treatment Team

- Occupational therapy
  - Address the sensory processing issues that are barriers to participation in feeding therapy:
    - Hyperresponsivity with stimulation to head, face lips and within oral cavity
    - Hyperresponsivity to touching wet textures
    - Fine motor skills to pick up food and utensils for increased interaction with foods
- Physical Therapy
  - Hypotonia impacting postural stability for feeding
  - Gross motor development precedes fine motor development (i.e, oral motor movements)

Case Study #3 B: Treatment Team

- Gastroenterology
- Pulmonology
- Speech Pathology: outpatient therapy and repeat modified barium swallow studies as clinically indicated

Case Study #3 B: Treatment Planning

- Systematically desensitize B and use sensory-based problem solving
  - Facilitate food exploration with pureed food on his tray
  - Getting B more comfortable with stimulation to the head, face and lips with goal of being able to complete oral motor exercises and use compensatory strategies
  - Treatment provided in front of a mirror
  - Approach from the side rather than head on
  - Monitoring for sensory responses and moving up and down feeding hierarchy consistent with Sequential-Oral-Sensory (SOS) approach to feeding.

Case Study#3 B: Treatment Planning

- Adaptive utensils
  - Beckman EZ spoon: flat shape reducing oral motor demands; small surface area at the tip provided less stimulation when presented to cheeks, lips or intraorally ; changed conditioning cue (i.e, aversion begins at the sight of the spoon typically presented) for feeding because it does not look like a typical spoon https://talktools.com/products/easy-spoon?variant=28452672585
- Parent Education
  - Disconnecting tube feeding 1 hour before sessions, to decrease vomiting with permission from physician **(implemented by evaluating clinician)**
  - Food logs to document vomiting and food acceptance **(implemented by evaluating clinician)**

Case Study #3 B: Treatment Planning

- Parent Education
  - Food exploration
    - Puree and hard munchable on tray to increase visual tolerance, interaction and messy play
  - Activities to facilitate oral motor skills ***as tolerated***
    - Oral motor tools
    - Facial towling
    - Hard Munchables consistent with SOS approach
  - Sensory-based problem solving
    - Following B's lead and not forcing noxious stimuli

Case Study #3 B: Treatment Planning

- Parent Education
  - Keep all interactions surrounding food warm, positive and fun
  - Impact of medical status on feeding
- Interprofessional Collaboration
  - Staying abreast of GI, pulmonary and general medical status
  - Gross, visual motor and fine motor skills

Case study #4 E Birth and Early Feeding History

- 4 year 3 month old female
- Born at 38 weeks gestation: induced due to premature ventricular contraction (PVC); weighed 5 lbs 11 ounces
- Jaundice and low blood sugar at birth due to hypoglycemia requiring 6 hour NICU stay
- Poor latch while breastfeeding; discharged to home without intervention; readmitted to the hospital days later
- Fed breast milk via baby bottle
- Baby cereal introduced at 4 months and E accepted. Physician advised against as it was earlier than typically recommended 6 months. Reintroduced at 7 months and has not accepted since.
- **Parental Concerns:** Only source of nutrition is Pediasure via baby bottle

Case Study#4 E: Medical History

- Ankyloglossia without frenectomy
- Aspiration at 4 months old
- G-tube placed at 4 months and removed at 12 months old
- MBSS has been attempted recently: all trials refused
- Chronic constipation
- Tetrasomy 18 P
- Spastic Diaplegic Cerebral Palsy
- Developmental delay
- Abnormal gait/toe walking
- Food allergies: eggs and peanuts (requires epipen)
- Recurrent ear infections
- Moderate hearing loss in right ear
- Sensory Processing disorder
- GERD
- Dystonia
- Muscle weakness
- Oral Aversion
- Abnormally small mouth
- High, arched palate

Case Study#4 E: Medical History

- Profoundly inmpaired speech and language
- Esotropia (inward turning of the eye). Corrected with surgery.
- Slow growth: less than 30 lbs, not on the growth chart

Outpatient Evaluation Results:

- Self-feeds Pediasure via baby bottle; hyper-extends neck to approximate nipple with highly-arched palate
- Does not remain seated in a feeding chair for PO intake. Parental report "has bottle where ever"; throws bottle when she is done
- Good postural support in adaptive stroller

Case Study #4 E: Medical History

Outpatient Evaluation Results

- Hyperresponsivity to clinician donning gloves and sitting next to her
- Observed sensory seeking behaviors: crawling rapidly around evaluation room; throwing items; gagging herself with her fingers; mouthing items (biting pages from books); parental report of sensory seeking behaviors includes crawling over high surfaces resulting in broken wrist
- Severe oral phase dysphagia, oral aversion and feeding disorder

Tetrasomy 18p

NORD's description of Tetrasomy 18p

- Very rare; 40-60 documented cases in medical literature
- Affects males and females equally; symptoms vary from case to case
- Affects many parts of the body
- Low birth weight, feeding problems, tendency to vomit
- Abnormalities of the head and craniofacial area
  - Fibrous joint (sagittal suture) between the bones that form the sides of the skull (parietal bones) may close prematurely (dolichocephaly)
  - Microcephaly (abnormally small head)
  - Facial asymmetry
  - Micrognathia

Tetrasomy 18p

- Craniofacial Abnormalities
  - Epicanthal folds (vertical skin folds) on the sides of the nose or covering the inner corners of the eyes
  - Cleft Palate
  - Ocular hypotelorism (eyes that are abnormally close together)
  - Gingival Hypertrophy (overgrown gums)
- Skeletal Abnormalities
  - Scoliosis (curvature of the spine)
  - Kyphosis (front to back curvature of the spine)
  - Abnormally small hip bones and hip abnormalities
  - Malformations of the hands and feet: fingers and toes that overlap; toes/fingers that are abnormally long or bent; webbed fingers and toes

Tetrasomy 18p

- Skeletal Abnormalities
  - Flat feet
  - Single deep crease along the palm of the hands (simian crease)
  - Absence of the horizontal ridges that are normally on the skin on the far end of the fingers (distal flexion ridges)
  - Distinctive body shape, consisting of an abnormally thin build, narrow chest and shoulders, and unusually prominent bones and/or muscles

Tetrasomy 18p

- Clusters of nerve fibers in the spinal cord (pyramidal tract) that help to regulate voluntary and reflex muscle activity may not function appropriately.
  - Hypertonia (increased muscle tone)
  - Hyperreflexia (increased reflex reactions)
  - Abnormal reflex consisting of flexing and relaxing foot (ankle clonus)
  - Spasticity
  - Abnormal gait
  - Clonus (involuntary flexing and relaxing of muscles)
- Kidney abnormalities
- Severe cognitive impairment
- Speech and language impairments
- Behavior abnormalities

Tetrasomy 18p and Feeding

According to the Chromosome 18 Registry and Research Society:

- Newborns with Tetrasomy 18p often have problems at birth or shortly thereafter.
  - Most common problem is nursing/feeding difficulties.
  - Problems latching onto the breast or bottle
  - Poorly coordinated suck-swallow-breathe
  - Some infants may vomit frequently after eating.
- Gastrointestinal problems are fairly common
  - The most common concern is constipation.
  - Reflux occurs somewhat frequently.

Tetrasomy 18P and Feeding

- In recent years, several individuals have been diagnosed with eosinophilic esophagitis (EoE).
- Inflammatory disease that affects the esophagus.
- Some symptoms include feeding difficulties and failure to thrive, reflux that doesn't respond to therapy; difficulty swallowing, nausea, and vomiting.
- Limited research available
  - Most briefly mentioned feeding difficulties in infancy
  - Lacking information on feeding difficulties later on in childhood

Case Study #4 E Treatment Team

- Physical Therapy
  - Postural stability
  - Seating
- Occupational Therapy
  - Significant sensory processing needs

Case Study #4 E Treatment Team

- SLP-BCBA
  - Consult regarding difficult to decipher behaviors: gagging herself; throwing all items both preferred and non-preferred outside of food.
- Gastroenterology
- Genetics
- Neurosurgery
- Neurology
- Allergist
- Physical medicine and rehabilitation
- Orthopedics
- Otolaryngology
- Audiology
- Registered Dietitian

Case Study #4 E Treatment Planning

- Co-treatment with occupational therapy
  - Address general sensory issues with the goal of increasing food acceptance and eventually exercise and activities to improve oral motor function
    - Work in the gym, brushing before feeding activities
    - Stimuli to the head, face, cheeks and lips using sensory-based problem solving
    - Safety!
- Goal #1: remaining seated in appropriate feeding chair in therapy sessions at at home for at least some PO intake
  - Adaptive stroller with tray

Case Study #4 E Treatment Planning

- Food exploration using sensory based problem solving; SOS approach to feeding
- Parent Education and Home Programming
  - Scheduling initially only allowed for one session per week
  - Intensive feeding program recommended, but is not an option for this family at this time.

Case Study #4 E Treatment Planning

- Parent Education and Home Programming
  - Building parental rapport and "buy in"
    - Family Dynamics
      - Single mother
      - Toddler siblings
      - E's significant needs
      - Displeased with previous feeding therapy experience
      - Skeptical if anyone can help E's feeding

Case Study #4 E Treatment Planning

- All recommendations had to be doable for the family
  - Encouraged E's mother to communicate openly if any recommendations were not feasible or ineffective
    - Agreed to 1-2 bottles of Pediasure a day in feeding chair
    - Messy play and child-lead food exploration
    - Sensory-based problem solving; not forcing noxious stimuli
    - E seated at table during family meals with pureed foods on her plate
- Interprofessional Collaboration
  - SLP/BCBA
  - Staying abreast of changes in medical status and impact on feeding
  - Contacting physicians and specialists as necessary

Summary

- Do your best to collect accurate and reliable information on rare diagnoses
  - What is consistent with this disorder that would impact feeding?
  - Are there any treatments that research shows are effective for these populations?
- ***We treat symptoms, not diagnoses***
  - What do we see?
  - What can we treat and address as speech language pathologists?
  - If we cannot treat it, who can? How do we collaborate with them or get them on the team?
- If you have any additional questions or would like follow up regarding any information I've shared today please contact me via email at slprachelf@therapyjackson.com

References

Arvedson, J. C., & Brodsky, L. (2002). *Pediatric Swallowing and Feeding*. Singular Thomson Learning.

Bavisetty, S., Grody, W. W., & Yazdani, S. (2013). Emergence of pediatric rare diseases. *Rare Diseases*, *1*(1), e23579. https://doi.org/10.4161/rdis.23579

Chehade, M., Meyer, R., & Beauregard, A. (2019). Feeding difficulties in children with non–IgE-mediated food allergic gastrointestinal disorders. *Annals of Allergy, Asthma & Immunology*, *122*(6), 603–609. https://doi.org/10.1016/j.anai.2019.03.020

The Chromosome 18 Registry & Research Society. (n.d.). *The Chromosome 18 Registry & Research Society*. Retrieved February 27, 2021, from https://www.chromosome18.org/tetrasomy-18p/

References

Diseases | Genetic and Rare Diseases Information Center (GARD) – an NCATS Program. (n.d.). Genetic and Rare Diseases Information Center. Retrieved February 27, 2021, from https://rarediseases.info.nih.gov/diseases

Mindlina, I. (2020). Diagnosis and management of Sandifer syndrome in children with intractable neurological symptoms. *European Journal of Pediatrics*, *179*(2), 243–250. https://doi.org/10.1007/s00431-019-03567-6

NORD - National Organization for Rare Disorders. (n.d.). *Home*. Retrieved February 27, 2021, from https://rarediseases.org/

References

NORD - National Organization for Rare Disorders. (2019, December 13). *Food Protein-Induced Enterocolitis Syndrome*. https://rarediseases.org/rare-diseases/food-protein-induced-enterocolitis-syndrome/

NORD - National Organization for Rare Disorders. (2015, February 11). *Chromosome 18, Tetrasomy 18p*. https://rarediseases.org/rare-diseases/chromosome-18-tetrasomy-18p/

NORD - National Organization for Rare Disorders. (2018, June 20). *Rubinstein Taybi Syndrome*. https://rarediseases.org/rare-diseases/rubinstein-taybi-syndrome/

Noble, A., Drouin, E., & Fauré, C. (2007). Eosinophilic Esophagitis and Gastritis in Rubinstein-Taybi Syndrome. *Journal of Pediatric Gastroenterology and Nutrition*, *44*, 498–500.

References

Reserved, R. A. U. I.-. (2007, March). *Orphanet: Sandifer syndrome*. Orpha.Net. https://www.orpha.net/consor/cgi-bin/Disease_Search.php?lng=EN&data_id=10994&Disease_Disease_Search_diseaseGroup=Sandifer-syndrome&Disease_Disease_Search_diseaseType=Pat&Disease(s)/group%20of%20diseases=Sandifer-syndrome&title=Sandifer%20syndrome&search=Disease_Search_Simple

Sdravou, K. (2019). Children with diseases of the upper gastrointestinal tract are more likely to develop feeding problems. *Annals of Gastroenterology*, 217–233. https://doi.org/10.20524/aog.2019.0348

Valdez, R., Ouyang, L., & Bolen, J. (2016). Public Health and Rare Diseases: Oxymoron No More. *Preventing Chronic Disease, 13*, https://doi.org/10.5888/pcd13.150491