Prader-Willi Syndrome: A Multidisciplinary Approach to Care

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No Conflict of Interest to Disclose

Objectives

- Identify the clinical features of PWS
- Discuss current guidelines for the management of children with PWS
- Recognize the benefits of treating PWS in multidisciplinary clinic
Introduction

- Prader-Willi syndrome (PWS) was first described by Prader, Labhart, and Willi in 1956.
- PWS is a complex genetic disorder caused by the absence of the normally active paternally expressed gene on chromosome 15q11q13.
- PWS represents the most common genetic cause of obesity.
- PWS is the first human syndrome identified with genomic imprinting.
- Estimated prevalence: 1 / 10,000 to 1 / 30,000 births.

Genetics of PWS

- Paternal Deletion - 70%
- Maternal Uniparental Disomy - 25% (UPD)
- Imprinting Defect - 5%
Diagnosis of PWS

- DNA-based methylation
  - Detects abnormal parent-specific imprinting within PWS region of chromosome 15
  - Reveals if region is maternally inherited only
  - Detects 99%
- DNA polymorphism to differ UPD/Imprinting
- Fluorescence in situ hybridization (FISH)
  - Deletion of 15q11.2–q13 (Angelman syndrome)
  - DNA-based methylation to confirm PWS

(Reprinted from Goldstone, et al 2008)

Clinical Features of PWS

Painting at the Prado by Juan Carreno de Miranda, 1680

Clinical Features of PWS

- Prenatal Characteristics
- Neonate Characteristics
- Childhood Characteristics
- Adolescent / Adult characteristics
Prenatal Features of PWS

- Decreased fetal movement
- Increased breech position
- Increased assisted delivery or cesarean section
- Birth weight, length, and BMI 15-20% less than unaffected siblings

(Miller, 2012)

Neonatal Features of PWS

- Hypotonia (nearly universal)
- Poor suck/Poor reflexes
- Decreases spontaneous arousal
- Weak cry
- May result in failure to thrive
- NG / G - tube feeding
- Genital hyperplasia
- Dyshormophism
- Strabismus

(Miller, 2012)

Childhood Features of PWS

- Dyshormophism
- Scoliosis (40-80%) varies in age of onset and severity (no relationship to GH has been found)
- Delayed motor development (average milestones reached at double the normal age)
- Delayed language (articulation abnormalities are most common)
- Learning disabilities by school age (IQ range borderline to low)
- Excessive eating begins in the preschool age group
- Obesity

(Cassidy, et al, 2011)
Childhood Features of PWS

- Sleep disturbance - abnormal sleep – wake organization
- Daytime sleepiness and sleep disordered breathing
  - Irregular REM cycles
- Growth hormone insufficiency / deficiency
- Skin picking
- Behavior and psychiatric problems:
  - Temper tantrums
  - Stubbornness, manipulative
  - Compulsive behavior
- Strabismus 60 - 70%
- Hip dysplasia 10 - 20%

(Cassidy et al, 2011; Miller, 2012)

Adult Characteristics of PWS

- Short stature
- Complications of obesity
- Delayed or incomplete puberty
- Scoliosis (can develop anytime)
- Low BMD (risk of osteoporosis)
- GH deficiency ?
- GHD found in 50% of adults
- Psychiatric / behavior issues

(Sanchez-Ortiga, 2012)

Clinical Features of PWS

(Catalotto, 2011)
Clinical Features of PWS

(Cataletto, 2011)

Genotype-Phenotype Correlations

UPD
- Longer gestational age
- Less facial characteristics
- Less likely to have hypopigmentation
- Less skill with jigsaw puzzles
- Higher verbal IQ
- Milder behavioral problems
- Higher psychosis and autism spectrum disorders

(Cassidy, et al, 2011)
Nutritional Phases of PWS

- Long held belief of two distinct nutritional phases in PWS (FTT and Hyperphagia leading to obesity)
- Collaborative study: Miller & Driscoll, 2011
- Reports seven different nutritional phrases
  Not all individuals go through all the stages
  Some adults progress to stage 4 (able to feel full)

Nutritional Phases of PWS

<table>
<thead>
<tr>
<th>Phase</th>
<th>Median ages</th>
<th>Clinical characteristics</th>
</tr>
</thead>
<tbody>
<tr>
<td>0.</td>
<td>Prenatal to birth</td>
<td>Decreased fetal movements and lower birth weight than sibs</td>
</tr>
<tr>
<td>1a.</td>
<td>0–9 months</td>
<td>Hypotonia with difficulty feeding and decreased appetite</td>
</tr>
<tr>
<td>1b.</td>
<td>9–25 months</td>
<td>Improved feeding and appetite and growing appropriately</td>
</tr>
<tr>
<td>2a.</td>
<td>2.1–4.5 yr</td>
<td>Weight increasing without appetite increase or excess calories</td>
</tr>
<tr>
<td>2b.</td>
<td>4.5–8 yr</td>
<td>Increased appetite and calories, but can feel full</td>
</tr>
<tr>
<td>3.</td>
<td>yr to adulthood</td>
<td>Hyperphagic, rarely feels full</td>
</tr>
<tr>
<td>4.</td>
<td>Adulthood</td>
<td>Appetite is no longer insatiable</td>
</tr>
</tbody>
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Endocrine Concerns in PWS

- Short Stature
- GH Insufficiency / Deficiency
- Hypogonadism
- Central Adrenal Insufficiency (CAI)
- Hypothyroidism
- Impaired Glucose Tolerance
Short Stature/GH Insufficiency

- Present by the second decade in the absence of GH therapy
- Average untreated heights:
  - 155 cm for males
  - 148 cm for females
- Data of 300 PWS children with decreased GH secretion - GH peaks <10 ug/liter in 70%
- High serum IGF 1 levels (72% >2SD) despite lower dosing
- IGF1 / IGF-BP3 ratios remain stable
- Aim of treatment is to improve growth during childhood
- Stimulation testing maybe helpful

(Burman, 2001)

Short Stature/GH Insufficiency

Recommendations:

- Early intervention*
- GH decreases fat mass and increased muscle mass
- Beneficial affects on: weight gain, appetite, and improved cognitive function?
- Improved HC, HT, BMI, improved lean muscle mass, delay of fat accumulation, body proportions, acquisition of gross motor skills, language acquisition and better cognitive scores
- Reported improved behaviors with less deterioration

(Cassidy, et al, 2011)

Hypogonadism

Genital Hypoplasia

Males:
- Penis may be small
- Scrotum is hypoplastic
- Scrotum is poorly rugated and pigmented
- Unilateral or bilateral cryptorchidism (80-90%)

Females:
- Genital hypoplasia is often overlooked.
- Clitoris and labia, especially the labia minora are generally small from birth
Hypogonadism
Incomplete / Delayed puberty

- Premature adrenarche ~ 15 - 20 %
- Delay/ incomplete puberty (30's)
  - Females: amenorrhea or oligomenorrhea
- Little is known about sexual activity Infertility
- Few incidences of reproduction have been reported
  (Eldar-Geva, et al, 2011)

Hypogonadism
Recommendations:
- Trial of hCG is recommended for infants with undescended testes
- Replacement of sex hormones
- Studies are not available on replacement therapy
- The decision to treat females is a personal decision of the family. If desired:
  - Females 11 - 12 years
  - Males: 12 - 13 years
  (Cassidy, 2011; Goldstone, 2008)

Hypogonadism
Recommendations:
- Low dose estrogen (transdermal patch) with escalating dose for 2 years or until menarche
  - Then combined estrogen – progesterone OCP or transdermal patch
- Low dose testosterone (transdermal patch or gel) escalating doses every 3 - 6months to allow testosterone to get to normal level or hCG injections
  (Cassidy, 2011; Goldstone, 2008)
Central Adrenal Insufficiency (CAI)

- In 2008, following an overnight single dose metyrapone testing CAI in 60% of the children with PWS
  - Cause of incidence of sudden death?
- Two studies found normal cortisol responses to low and high ACTH
- CAI is uncertain
- No consensus among endocrinologists as to need to do CAI evaluation on everyone with PWS

(dela Lind van Wijngaarden, 2008; Nyunt, 2010; Farholt, 2011)

Central Adrenal Insufficiency (CAI)

Recommendations:

- Counsel patient families of the symptoms of adrenal insufficiency
- Education about the best action and treatment should symptoms occur

Hypothyroidism

- Normal or low TSH value and a low free T4 level as been documented in up to 25% of individuals with PWS
- Mean age of diagnosis and treatment 2 years
Impaired Glucose /Diabetes Mellitus

- Up to 25 % of PWS adults (particularly with obesity)
- Mean age of onset age 20 years
- Large French study PWS cohorts 2 – 18.8 yrs  
  - Revealed impaired glucose tolerance in 4 % of individuals (mean age 10.2 years)
- No DM in those less than 20 years

(Diene, 2010)

Additional Concerns of PWS

Sleep Disturbance
- Abnormal sleep – wake organization
- Daytime sleepiness and sleep disordered breathing (central and obstructive sleep apnea, reduced REM latency, abnormal response to hypercapnia)
- Irregular REM cycles
- Narcolepsy with rapid onset of REM sleep and decrease in non REM sleep instability
- Appears as early as infancy

Additional Concerns of PWS

Sleep Disturbance Recommendations:
- Sleep study to assess for obstructive apnea
- Baseline before starting GH
- Six weeks after starting GH
- Anytime new symptoms occur with suspicion on / off GH
- Evaluation and treatment of sleep study / symptoms
- T&A and / or Pulmonary referral
- CPAP / BiPAP / O₂
Discuss Current Treatment Guidelines for PWS

Current Treatment Guidelines for PWS

- Early diagnosis (accredited genetic testing)
- Multidisciplinary teams
- Growth hormone treatment
- Vigorous control of food environment
- Understanding of behavioral / psychiatric aspects
- Transition to adulthood
- Increased availability of group homes
- Recognize the distinction between underlying behavioral problems seen in PWS and acute psychiatric illness
- Family education and support

Current Treatment Guidelines for PWS

Childhood

- Strict supervision of daily food intake (BMI < 30)
- Consultation with dietician with close follow up
- Lock kitchen cabinets and refrigerator
- Firm limit setting
- Sleep evaluation and treatment
- T&A or CPAP / O2 / BiPAP)
- GH treatment
- Educational planning – Individual classroom aid
- Social skills training
- Speech therapy for articulation problems
- Treatment of increased salvia if needed / Routine dental exams
Current Treatment Guidelines for PWS

Adolescents / Adult
- Management of Hyperphagia
- Prevention of obesity the same as with children
- GH therapy
- Replacement of sex hormones if desired
- Consideration of sex education and contraception
- Transition to group home

Uncertainties in Management of PWS
- Best age to start growth hormone therapy
- IGF 1 levels and how often to measure
- Role of growth hormone in transition to adulthood
- Indications and best regimen for sex steroid replacement
- Pathophysiology of hyperphagia and hence therapeutic possibilities
- Pathophysiology of psychiatric and psychological illness and hence therapeutic possibilities

Recognize the Benefits of a Multidisciplinary Clinic for PWS
Recognize the Benefits of Multidisciplinary Clinic for PWS

- Emphasize early intervention
- Proactive approach to assessment of and treatment for the possible complications that can be associated with PWS
- Will assess growth and development of newborns through young adult
- Determines age appropriate and individual treatment plans for your child
- Evaluates bone health, scoliosis and reproductive health
- Directs growth hormone therapy and other endocrine replacement hormones

Nationwide Children’s Hospital
Columbus, Ohio

History of PWS Clinic

- PWS Clinic Started in 2009
- Joint Endocrine and Genetic
- Patients Included:
  - Predominately PWS
  - Russell - Silver syndrome (RSS)
  - Noonan syndrome
- Initially staffed by:
  - Dr. Dana Hardin, Pediatric Endocrinologist
  - Dr. Joan Atkin, Genetics
  - Nurse Coordinator / Genetic Counselor / RD
NCH PWS Clinic Requirements

- Confirmed Diagnosis of PWS (Methylation testing)
- Age of PWS Patients
  - Newborn to 23 years
  - Transition to Adult Care after age 23 years
- Frequency of Clinic
  - Second Friday of Every Month
  - Fourth Friday of every other month

PWS Clinic Today

- Endocrinologist
- Genetics (physician and counselor)
  - Age < 3 years (each visit)
  - Age > 3 years (every 6 months)
  - Adolescent (annually)
- Nutritionist
- Psychologist
- Social Worker
- Endocrine Fellow
- Endocrine Nurse Coordinator

Dedicated specialties outside PWS clinic

- ENT
- Pulmonary
- Sleep Clinic/ Sleep Lab
- Speech Pathologist
- Dermatology
- Urology
- GI
- Ophthalmology
- Dentist
PWS Clinic Today

Non PWS Specific Outside PWS Clinic Visit
- Orthopedic
- Orthodontist
- Surgery
- Neurology (Seizure Clinic)

How the Clinic Works
- Nurse Coordinator arranges all clinic appointments
- Clinic has 4 rooms
  Schedule 4 patients – 10 minutes apart
- Schedule average of 8 patients per clinic session
- One provider goes in with each patient
- New Patient Appointment scheduled as first patient of the clinic- Genetics sees NP first
- NP visits last ~ 2 hours
- Follow up visits are planned for 1 ½ hours

A Typical PWS Clinic Visit

Behind the scenes!
- “Gigi”
  - Administrative support
  - Calls families with appointments
  - Reminder calls / mails cards / faxing
  - Prepares patient board before clinic
  - Girl Friday
A Typical PWS Clinic Visit

**Registration**
“Dana”
- Welcomes families to their clinic visit
- Updates demographics
- Copies insurance card / court / legal documents
- Consent
- Ensures patients have an outpatient code
- Gives stickers!

A Typical PWS Clinic Visit

**Medical Assistant**
- Accurate weight, height / length
- BP, HC
- Rooms the patient
- Updates tracking board
- Flu vaccine
- HgbA1c (POCT)
- Helps with distraction when needed

A Typical PWS Clinic Visit

**Endocrine Team**
- Dr. David Repaske
- Dr. Manmohan Kamboj
- Dr. Kathryn Stephens
- Katie Anglin, MSN, RN
- Early intervention and comprehensive management of the complications of PWS
- GH therapy
A Typical PWS Clinic Visit

PWS Nurse Coordinator:
- Clinic Coordination and clinic flow
- Medicine reconciliation
- GH teaching / up to date prior authorizations/ appeals / GH trouble shooting
- Collaboration of PWS specialty appointments
- Ensures follow-up of all referrals are done
- Ensures ordered testing is done and results reported
- Follow up from PWS conference orders
- Database of PWS patients / procedures

A Typical PWS Clinic Visit

Integrated Psychology
“David”
- Behavioral, educational, and advocacy needs
- Monitors social, cognitive, emotional development
- Behavior prevention
- Adjustment to developmental delays
- Full psychological evaluation, as needed

A Typical PWS Clinic Visit

Social Worker:
“Marcie”
- Risk / safety, economic resources / needs
- Emotional / social issues
- Family support
- Insurance issues
- Transportation
- School issues
- Children Services contact
A Typical PWS Clinic Visit

**Genetics:**

“Dr. Joan Atkin & Teresa”

- PWS type specific: Diagnosis and Early intervention
- Complete developmental assessments
- Complete physical assessment
- Genetic counseling
- Genetic letter

A Typical PWS Clinic Visit

**Registered Dietician**

- Comprehensive diet evaluation and nutrition plan
- Food diary (between visits)
- Food aggression / sneaking
- Special “birthday” recipes
- School letters regarding
- Rewards for the kids

PWS Clinic Post Conference

- All PWS team members attend
- One hour (2-3 PM)
- All patients are discussed by every discipline / provider
- Issues are identified, discussed and elaborated
- Comprehensive management and follow up plan developed
- PWS nurse coordinator ensures follow through with the comprehensive management plan
- Discussion of PWS findings (i.e. sensitive skin, frequent asthma diagnosis)
- Special speakers for teaching
PWS Case Study 08 / 11 / 12

Chief complaint:
- 5 years and 4 months old male
- PWS (Deletion) methylation testing
- Foster mother and birth father at the visit
- Child came in WC- he was not walking

Summary of Past Medical History:
- Developmental delays (crawled at one year, walked 5 years
- G - tube until 18 months (his brother pulled out)
- GH started at 6-8 months and stopped ~ 3 years old

Summary of Past Medical History continued:
- Body mass index (BMI) > 99% for age
- Off GH > 2 years
- Abnormal EEG
- Uncircumcised
- Undescended testicles
- Developmental / Speech delays
- Last sleep study was obtained at age 3

Birth History:
- Mother was 34 year old G4 P2->4AB0
- Received prenatal care
- Known exposure to chemotherapy for MS prior to awareness of pregnancy
- No tobacco, alcohol, drugs, or other known teratogens
- Pregnancy was complicated by twin pregnancy
- Fetal activity was normal and growth parameters showed symmetric growth
**Birth History continued:**
- Multiple US (twins)
- Planned cesarean section secondary to twin gestation
- Birth Weight 6 lbs 7 oz / fraternal twin weight 7 lb 6 oz
- Hypotonia while hospitalized in the NB period
- Deletion PWS was diagnosed in NICU
- Home on G-tube feedings

**Social History:**
- Fraternal twin (autism)
- Father 40 years old (with anxiety)
- Mother living in MS facility
- Lives with foster family and fraternal twin brother
- No smoking in the house
- 8 adopted children of the foster family, most with special needs
- 5 at home; age range 11-18 years old
- First visit with foster mother and biological father

**Endocrinology Template**

**Feeding / Diet:**
800 Kcal diet. Twin is seeking food.

**GH:**
Not on GH since age 3 years and 6 months.
Sleep study obtained at age 3 years, external.
Denies snoring or gasping while sleeping.

**Behavior / Therapy / Developmental:**
Speech therapy through county services. Wait list for PT / OT through county. Planned special needs preschool in the fall. Denies skin picking, behavior issues, bedwetting. Started walking age 5 years. In WC today.

**Ophthalmology:**
Occasional strabismus (R eye). No eye exam done.
PWS Case Study 08 / 11 / 12

Dental:
Poor enamel and dentition, noted thick saliva. Dentist (< 6 months ago) would like to pull teeth if every under GA.

Urology:
Undescended testes. Scrotal ultrasound by PCP 5/2012 did not identify testes within the scrotum or inguinal canal. Appointment with Urology 8/2012.

Neurology:
Abnormal EEG by PCP; pending Neurology appointment.

Physical Exam:
BP 120/58 — Ht 112 cm — Wt 49.5 kg; BMI 39.46 kg/m². G-tube site is not closed.

PWS Case Study 08 / 11 / 12

Genetics
• Full pedigree taken by genetic counselor
• Obese
• Dysmorphic features noted
• Bitemporal narrowing
• Almond shaped eyes
• Pre-auricular skin tag
• Typical PWS features
• Tapered fingers and toes
• No scoliosis
• Striae
• Complete deletion PWS genetic letter for family and case worker

PWS Post Conference 08 / 11 / 12

Endocrine / Genetic Plan:
• Obtain previous sleep study
• Plan on starting GH @ 0.24 mg / kg / week
• Refer to Ophthalmology for right eye strabismus
• Refer to Physical Medicine for fitting devices
• Refer to Surgery to help close G-tube site
• Refer to Urology for bilateral undescended
• Refer to Neurology for abnormal EEG
• Refer for speech therapy
• Social work involvement
PWS Post Conference 08 / 11 / 12

Nutritional Plan:
- Letter for foster parent and Children’s Services clarifying need for a caloric restricted diet
- Skim milk or calorie-free beverages
- Increased physical activity
- Securing food sources at all times
- Portion control
- Colored plates with "Go" (green) and "No" (red) food choices using the Stop - Light Method
- Toy during meals to include child while controlling intake

PWS Post Conference 08 / 11 / 12

Social Work Plan:
- Support in negotiating resources with Children Services
- Facilitate BCMH application
- Help with paperwork for preschool
- Provide information to Children’s Services regarding PWS and medical needs
- Did not see Psychologist this visit

PWS Case Study 10 / 26 / 12

Feeding / Diet:
Sticking to 800 Kcal diet. Rarely sneaking food.

GH:
Sleep study report from 07/03/12. Severe OSA. No central sleep apnea noted. O₂ desats into 70's. No abnormal EEG. Snoring. Recommend ENT.

Behavior / Therapy / Developmental:
Speech PT / OT in special needs pre-school
Getting behavior modification therapy in the home.
Denies skin picking, behavior issues, or bedwetting.
Recently started walking. Walked to clinic today.
Fitting devices on.

Ophthalmology:
No strabismus found. Glasses ordered, getting today.
PWS Case Study 10 / 26 / 12

**Dental:**
No new issues.

**Urology:**
Urology evaluation done in Sept with repeat testicular US. No testes in the scrotum or inguinal canal. Possible hCG stimulation.

**Neurology:**
Neurology appointment moved to Dec.

**Psychosocial:**
Biological father is living in homeless shelter and did not come to this visit due to anxiety issues. Full day preschool due to severe delays.

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PWS Case Study 10 / 26 / 12

**Physical Exam:**
VS normal; BMI 32.99 kg/m²
Leaking G - tube site; Testes not palpable.

**Psychology:**

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PWS Post Conference 10 / 26 / 12

**Endocrine/ Genetic Plan:**
- Sleep study appointment
- hCG ordered (testosterone and inhibin B)
- Repeat testicular US after hCG
- Follow up with urology after hCG
- Surgery appointment to close G - tube site
- Obtain bone age reading

**Psychology Plan:**
- DP-3 and CBCL to be completed and mailed back
- Use distraction for minor tantrums
- “Commands while touching”
**PWS Case Study Between Visits**

11 / 02 / 12 **Bone age**
Chronological age 5 years and 6 months
Skeletal age 10 years
*Impression:* Bone age advanced more than 5 SD above the mean. Endocrinologist reading, puts him at – 3SD for height corrected for BA (used phalanges as better predictor)

11 / 05 / 12 **Sleep study**
Snoring 100% of the time with severe OSA, worse in REM sleep with O₂ desats into the 50’s. No usual EEG spikes. Less severe in the right lateral decubitus position. REFER to ENT

11 / 09 / 12 **ENT appointment**
11 / 14 / 12 T&A, DL bronch, and turbinate reduction
In ICU for post op O₂ concerns
11 / 18 / 12 Put on CPAP 5 (O₂ sats 60s)
11 / 22 / 12 I & D of G-tube site
12 / 01 / 12 G-tube site surgically closed
12 / 24 / 12 hCG stimulation begins
3,000 units twice per week for 8 doses
Draw total testosterone 3 days after hCG

**Inhibin B**

<table>
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<tr>
<th>REF. RANGE</th>
<th>12/31/2012 11:15</th>
<th>01/03/2013 12:06</th>
<th>01/04/2013 10:00</th>
<th>01/05/2013 10:00</th>
</tr>
</thead>
<tbody>
<tr>
<td>PMSG T4</td>
<td>0.7-10.0 μg/L</td>
<td>1.1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>TSH</td>
<td>0.3-5.0 μIU/mL</td>
<td>3.765</td>
<td></td>
<td></td>
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<tr>
<td>TOTAL TESTOSTERONE</td>
<td>3-50 ng/dL</td>
<td>57</td>
<td>98</td>
<td>142</td>
</tr>
</tbody>
</table>

**FREE T4 Latest Range:**
0.7-2.1 ng/dL

**TSH Latest Range:**
0.6-4.5 μIU/mL

**TESTOSTERONE Latest Range:**
2-30 ng/dL

**HCG Stimulation Begins:**
3,000 units twice per week for 8 doses
Draw total testosterone 3 days after hCG
PWS Between Visits

01 / 25 / 13 Scrotal US
Small bilateral testes are identified, within the inguinal regions. Neither in the scrotal sacs. Left testes: 1.6 x 0.7 x 1.0 cm (total volume 0.6 mL). Right testes: 1.9 x 0.8 x 1.1 cm (total volume 0.9 mL)

Of note: the left testicle appears positioned within the anterior subcutaneous fat.

PWS Case Study 03 / 08 / 13

Endocrine Interval History:
GH:
Hold
Sleep Study:
Repeat sleep study February 2013 showing AHI of 4 but was much improved from prior to surgery. Per pulmonary okay to restart growth hormone once a download of the CPAP machine documents use. Pulmonary is considering a repeat sleep study after his next CPAP clinic visit.

ENT:
Follow up this morning; no new concerns

PWS Case Study 03 / 08 / 13

Feeding / Diet:
Religiously sticking to a 800 calorie diet. Foster mother reports he is rarely sneaking food, but will occasionally sneak if food is left out. Since being seen in PWS clinic for the first time in 8/2012, has successfully continued to lose weight. Has lost 1.3 kg since his last visit.

Behavior / Therapy:
Currently enrolled in Speech therapy, PT / OT in special needs pre – school. Expressed concern over articulation of certain sounds including "J" and "K"; although does not have a hypernasal quality to speech. Continues with behavior modification. Family Denies skin picking, behavior issues, bedwetting.
PWS Case Study 03 / 08 / 13

**Ophthalmology:**
No new concerns. Wearing glasses.

**Dental:**
Dental hoping to pull several broken teeth in conjunction with upcoming testicular surgery.

**Urology:**
Inhibin B slightly low (some Sertoli cell function). HCG stim test with 8000 mcg hCG given twice weekly x 4 weeks started on 12/24/12 and complete on 1/21/13 with weekly testosterone 27, 59, 143, 250, showing excellent response indicating good Leydig cell function. Penile growth w/ hCG.

PWS Case Study 03 / 08 / 13

**Urology continued:**
Discussion regarding testicle removal verses orchidopexy – endocrinologist and urologist.

**Neurology:**
History of abnormal EEG obtained by primary care physician (slowing) but no history of seizure activity. Appointment scheduled with Neurology in 12 / 2012; was unable to attend due to family emergency and needs to reschedule.

**Cardiology:**
Evaluated due to history of severe OSA. No concerns for pulmonary hypertension and no follow up needed.

PWS Case Study 03 / 08 / 13

**Psychosocial:**
Family is planning reunification of the twins with biological parents. Parents are now living in apartment. Plan to reunite twins with biological parents in March 2013. Court case scheduled for next week. Foster mother wants to stay involved as medical liaison for appointments.

**Nutrition:**
Written individual diet plan
Discussed nutrient verses caloric density
Emphasize portion sizes / portion control
Case Study Conclusion

PWS Case Study Epilog

03 / 15 / 13 Decision made to perform orchiopexy
Surgery date pending

03 / 23 / 13 CPAP download received and he is
wearing mask 100 % of the time

03 / 26 / 13 Update from case worker that
biological family is moving forward on
reunification after last court date

03/ 29 / 13 Apply for GH therapy

Requirements for Success

• Dedicated and interested providers
• Role of the clinic nurse coordinator is key
• Post Conference:
  ✓ To tie up loose ends
  ✓ Make new plans for treatment and follow up
  ✓ Discuss complex situations
  ✓ Accept input from different providers
• Community / family support / PWS of Ohio
PWS Research


Conclusion: Adenotonsillectomy, while effective in most children with PWS who demonstrate mild to moderate OSA, may not be curative in children with severe OSA. An increase in central apneas can occur in some children with PWS postoperatively, and it is important to repeat PSG after surgery. Further studies are necessary to determine optimal treatment for some children with PWS and sleep-disordered breathing.

Family Resources


References


References


Retrieved from http://www.eje-online.org/content/162/2/37772.

References


References


References

Prader-Willi Syndrome: A Multidisciplinary Approach to Care

Questions

Thank You!
Consent was obtained for all pictures.