

Health Issues in the Aging Adult with Prader Willi Syndrome

Contributed by Barb Dorn, RN, BSN

Introduction

In 2024, I began researching health issues in the aging adult with Prader-Willi syndrome (PWS). I learned that there was very little specific information on this topic. I did find a few articles that documented clinical evidence of early signs of aging. On the topic of dementia, I found one study that identified dementia in some individuals with PWS, along with an article that shared a case study of a woman with PWS who had dementia. Because the number of adults with PWS that had been studied was so low, it was difficult to know if these studies had broader implications. At this time, unlike Downs syndrome, there does not appear to be a genetic connection to dementia.

I developed and distributed a survey to help identify what health issues parents, guardians and residential caregivers were seeing in adults with PWS, ages 30 years and older. I received 89 surveys from parents and guardians and 9 from residential care givers. The residential care providers shared information for 21 individuals with PWS. All ranged in age from 19 years (1 person) and 30 years through 60+ years. Fifty were males and sixty were females. Questions focused on signs of early aging, the National Council of Aging's 10 most common chronic conditions for adults over the age 65 years, as well as anecdotal reports of additional health concerns and chronic illnesses being reported in aging persons with PWS. Many of our aging adults have not benefited from long term use of hormone replacement including growth hormone therapy. Some were diagnosed at a later age and have not benefitted of the many research and treatment advances. The health issues that aging individuals are facing today do vary between individuals but there are some commonalities.



Susan – Age 76

The Most Common Health Issues

The following health conditions were identified as the most prevalent and concerning in aging adults with PWS by parents, guardians and caregivers. These concerns are summarized, addressing several factors along with comments and resources. I focused on the most frequently reported issues.

Health Concern	Risk Factors in PWS	Screening / Monitoring	Prevention & Management	Other
Dental Issues <ul style="list-style-type: none"> • Extreme dental wear (typically from grinding) • Cavities • Abscessed teeth • Limited Access to Care 	<ul style="list-style-type: none"> • Abnormal saliva • Poor dental habits • Grinding – severe wear & gastric regurgitation • Poor enamel – diet & gastric regurgitation • Diet – Food high in acid include – soft drinks, sport drinks, juices, coffee. 	<ul style="list-style-type: none"> • Dental exams at least every 6 months • Monitor enamel – watch for translucency, darkening or discoloration of the teeth • Pain or cold sensitivity (may not always be reported in persons with PWS) 	<ul style="list-style-type: none"> • Dental exams with sealants and fluoride treatments at least every 6 months • Prompt evaluation of any enamel erosion • Use of bite guard • Restoration if possible • Strong emphasis on dental care every day – incentive programs. • Drink water. Limit soft drinks, sport drinks, juices and coffee in moderation. Dilute juices with water. • Swish mouth out with water or brush teeth 	<ul style="list-style-type: none"> • It is challenging to obtain dental care for the adult who is on Medicaid / Medical Assistance. • Utilize Dental Schools as well as schools that train Dental Hygienists. • Investigate Free Clinics or free dental events • Some Special Olympics organizations offer free clinics.

			<p>after drinking or eating items high in acid</p> <ul style="list-style-type: none"> Limit drink or food high in acid especially in the evening. 	<ul style="list-style-type: none"> Check state Dental Associations for “Donated Dental Services.” Investigate Medicare Advantage programs. Some provide dental coverage.
Health Concern	Risk Factors in PWS	Screening / Monitoring	Prevention & Management	Other
Dental Issues continued			<ul style="list-style-type: none"> Use a straw to keep acid away from teeth. Brush with fluoride toothpaste. Talk to a dentist about daily fluoride <u>mouthwash</u> if there is a history of cavities (Use as a rinse; do not swallow). If needed – apply with Q-tip. 	<ul style="list-style-type: none"> Private payment / individual dental insurance programs through the dental professional (monthly payment program) <p>RESOURCE: https://www.pwsausa.org/wp-content/uploads/2024/06/Dental-Health-in-Children-and-Adults-with-PWS.pdf</p>
<p>Slow Gastrointestinal Functioning</p> <p>Choking</p> <ul style="list-style-type: none"> High risk for aspiration and death 	<ul style="list-style-type: none"> Hyperphagia & fast eating Obesity Poor oral motor control Low muscle tone in entire GI tract Binge episode Thick saliva /Dry mouth Repeated episodes of pneumonia (aspiration) 	<ul style="list-style-type: none"> Monitor eating Evaluate repeated episodes of choking or coughing after eating Swallow Study 	<ul style="list-style-type: none"> Cook all vegetables to soften – avoid raw veggies. Add broth or liquids to ease with swallowing “Pace and Chase” – follow all food with sip of water. Cut food in to small pieces Serve small quantities at one time. Supervise person during eating Know Heimlich procedure Thickening of fluids 	<p>RESOURCE: https://pediatricsnationwide.org/2020/07/31/c choking-education-an-important-element-of-care-for-prader-willi-syndrome-families/</p>
<p>Slow Gastrointestinal Functioning</p> <ul style="list-style-type: none"> Gastroparesis <p>Many reported cases across all ages</p>	<ul style="list-style-type: none"> Binge eating Lack of vomiting HIGH fiber diet 	<ul style="list-style-type: none"> Know signs that require urgent medical evaluation – abdominal distention or pain, or vomiting Referral to gastroenterologist 	<ul style="list-style-type: none"> Report and monitor binge episodes Prompt treatment of acute illness Small feedings – chew foods well AVOID high fiber foods and fats – encourage lower fiber, softer diet Sit up 1 hour after eating &/or walk Medication may be ordered 	<p>Always have GI chart available</p> <p>RESOURCE: GI-Algorithm-Chart-2022.pdf <p>(pwsausa.org)</p></p>

<p>Slow Gastrointestinal Functioning</p> <ul style="list-style-type: none"> • Constipation <p>Lifelong health issue</p>	<ul style="list-style-type: none"> • Inadequate intake of water/fluids • Inadequate fiber • Low activity level • Medications • Other health problems – low thyroid 	<ul style="list-style-type: none"> • Daily monitoring for constipation - watch for abdominal distention • Rule out other causes of intestinal slowing – thyroid, colon cancer • Rumination / regurgitation should be evaluated by a health care professional. • <u>Urgently evaluate</u> nausea, vomiting and abdominal distention. 	<ul style="list-style-type: none"> • Keep moving and active. • Make the time – develop schedule for having a BM. (Best after eating or exercising) • Adequate fluid intake • Consult MD for use of laxatives. • A "bowel program" and plan is often needed. <p>Medication Caution – anti-diarrheal & pain medications can cause severe slowing of intestines.</p>	<p>Bristol stool chart helps identify if treatment is needed.</p> <p>RESOURCE: Colorectal Function in Adults w PWS - Constipation1471-230X-14-63.pdf</p>
Health Concern	Risk Factors in PWS	Screening / Monitoring	Prevention & Management	Other
<p>Prediabetes / Diabetes</p> <p>Prediabetes – elevated blood sugar that isn't high enough to be diagnosed as Type 2 Diabetes. Frequently no symptoms.</p> <p>FASTING BLOOD SUGAR RANGES: NORMAL: 70-99 PREDIABETES: 100-125 TYPE 2 DIABETES – 126+</p>	<ul style="list-style-type: none"> • Obesity • Poor diet management – high carbohydrate, saturated fats & processed foods • Age – over the age of 45 years • Low activity level • Diagnosis of Prediabetes • Low thyroid • Medications - steroids 	<ul style="list-style-type: none"> • Signs of increased thirst should be evaluated by health care professional. • Annual screening – fasting blood sugar, A1C, metabolic panel • Monitor weight daily • Monitor blood sugars as recommended by health care professional. • Kidney blood testing – creatinine, BUN • Blood lipids – cholesterol, triglycerides, HDL, LDL • Annual eye exams 	<ul style="list-style-type: none"> • Weight management • Consultation with a nutritionist / dietician • Increase activity • Medications including insulin may be necessary • See endocrinologist (doctor who specializes in diabetes) • Consultation with diabetes nurse educator 	<p>Diabetes is a serious health condition that impacts almost every organ in the body – kidney, eyes, blood vessels and heart.</p> <p>Often difficult to diagnose diabetes in persons with PWS when there is increased hunger, thirst and weight gain</p>
<p>Respiratory Issues</p> <ul style="list-style-type: none"> • Accounts for 50% of deaths 	<ul style="list-style-type: none"> • Poor muscle tone • Sleep disordered breathing – sleep apnea • Obesity • Low activity levels • Daytime sleepiness 	<ul style="list-style-type: none"> • Sleep study • Consultation with a sleep specialist • Daily weight – weight management 	<ul style="list-style-type: none"> • Evaluate excessive daytime sleepiness • Prompt treatment of respiratory infections • Weight loss • Keep moving • CPAP 	<p>RESOURCE: Respiratory-problems-2022.pdf (pwsausa.org)</p>
<p>Weight Gain - Obesity</p> <ul style="list-style-type: none"> • More are maintaining a healthy weight. • Key in preventing health complications. 	<ul style="list-style-type: none"> • Decrease in calorie needs – common in all as we age • Lower activity levels. • Mobility issues • Hypothyroid 	<ul style="list-style-type: none"> • Daily weight • Thyroid monitoring • Nutrition consultation 	<ul style="list-style-type: none"> • Low calorie / high nutrition diet • Food security • Keep active and moving • Walking – any distance. • Medication for thyroid replacement. 	<p>Obesity contributes to:</p> <ul style="list-style-type: none"> • Respiratory problems • Bone degenerative changes & mobility issues • Circulation / Cardiac • Diabetes • Bowel / Intestinal issues

<p>Low Bone Density – Osteopenia & Osteoporosis</p> <hr/> <p>What is Osteopenia and Osteoporosis?</p> <ul style="list-style-type: none"> • Our bones' structure becomes more porous as we age. • BMD (bone mineral density) scores drop <hr/> <p>OSTEOPENIA – BMD -1 to -2.5 OSTEOPOROSIS – BMD – 2.5 or lower (high risk of fracture)</p>	<ul style="list-style-type: none"> • Decrease in hormone production – GH, estrogen, testosterone • Arthritic changes • Vitamin & Mineral deficiencies – Vit. D, Calcium • Poor Muscle tone • Medications – SSRI's • Age 	<ul style="list-style-type: none"> • Hormone level testing -GH, sex hormone, thyroid function levels. • Bone density testing – DEXA scan • Vitamin & mineral level monitoring – calcium, phosphorous, magnesium, parathyroid hormone, serum 25 Vitamin D 	<ul style="list-style-type: none"> • Hormone replacement • Bone density monitoring • Medication treatment of osteoporosis • Vitamin & mineral supplementation • Dietary consulta – find ways to add calcium & vitamins to diet. • Weight bearing exercise and activities – walking, climbing, biking. • Evaluate medications – meds used for seizures/ mood stabilization (SSRI's) / acid reducers can lower calcium absorption (risk vs benefit) 	<p>An endocrinologist is typically the specialist in managing hormone replacement for osteopenia and osteoporosis. GH therapy has been found to have a positive impact on bone density in the adult.</p> <p>RESOURCE: Osteoporosis-Evaluation-and-Therapy-in-PWS.pdf pwsausa.org</p>
Health Concern	Risk Factors in PWS	Screening / Monitoring	Prevention & Management	Other
<p>Cardiac Issues High Cholesterol, Hypertension, Fluid Retention /Swelling (Oedema) & Deep Vein Thrombosis (DVT)</p> <ul style="list-style-type: none"> • Lower extremity swelling in both legs may be obesity hypoventilation (Gourash) • Lower Extremity swelling & redness in one leg – may be DVT 	<ul style="list-style-type: none"> • Obesity – poor weight management • Low activity / sedentary life style • Diet • Low GH • Sleep apnea 	<ul style="list-style-type: none"> • BP monitoring • Daily weight • Ongoing monitoring with PCP • Endocrine Consult • Lab testing – Lipids, GH • Sleep study • Work up for DVT – vascular ultrasound, venogram, D-dimer blood test 	<ul style="list-style-type: none"> • DASH diet – limits salt, sugar and saturated fats. • Exercise – walking • Weight management • Medication management • CPAP / Oxygen if needed • Referral to lymphedema specialist / Wraps or sleeves to reduce swelling • Anticoagulant therapy as part of high-risk surgery • **If DVT or Pulmonary Emboli is diagnosed, anticoagulation therapy 	<p>Deep vein thrombosis (DVT) (blood clot in the lower extremity) Increases risk for a pulmonary embolus (PE) – blood clot in lung.</p> <p>GH therapy found to have a positive impact on cardiac health in the adult.</p> <p>RESOURCES: Edema in PWS Linda Gourash pwsausa.org https://praderwillinews.com/news/pws-patients-more-prone-to-have-blood-clotting-problems-study-shows/</p>
<p>Mobility Issues – Spine Deformities</p> <p>Scoliosis and Kyphosis most common spine deformities</p> <p>Scoliosis – lateral curvature of spine Kyphosis – posterior curvature</p>	<ul style="list-style-type: none"> • Decrease in hormone production – GH, estrogen, testosterone • Arthritic changes • Vitamin & Mineral deficiencies – Vit. D, Calcium • Poor muscle tone / posture 	<ul style="list-style-type: none"> • Hormone level testing -GH, sex hormone, thyroid function levels. • Bone density testing – DEXA scan • Vitamin & mineral level monitoring – calcium, phosphorous, Vit. D, magnesium, parathyroid hormone • Spine x-rays 	<ul style="list-style-type: none"> • Hormone replacement • Bone density monitoring • Use of walker • Exercise that strengthens core and balance. • Spine support and attention to posture • Grab bars, good shoes, avoid clutter on floor, shower chair 	<p>Keep person with PWS mobile. Once becomes wheelchair dependent –increases many more health risks – blood clots, pulmonary emboli, muscle loss, weight gain.</p>

of the spine - "humpback"	<ul style="list-style-type: none"> Gait issues – high risk for falling 		<ul style="list-style-type: none"> In severe cases - wheel chair Home assessment Ranch home OT/PT referral 	
Worsening Mental Health <ul style="list-style-type: none"> Depression Low Energy Dementia <p>Several medical conditions can contribute to changes in mental health and energy levels. Low energy should not be labeled as "PWS behaviors".</p>	<ul style="list-style-type: none"> Life long struggle with mental health issues Hormonal changes – thyroid, menopause, low testosterone Diabetes and cardiac issues – can cause fatigue Constipation Lack of meaningful activities and/or job Isolation Limited physical activity 	<ul style="list-style-type: none"> Physical exam by PCP to rule out medical issues, including dementia Referral to neurologist, geriatrician or mental health professional <u>Cognitive Screening Tools:</u> CAMDEX- DS: assessment tool for those w/ Down Syndrome ABDQ- adaptive behavior dementia questionnaire which measures changes in adaptive behaviors 	<ul style="list-style-type: none"> Maintain communication with mental health professional. Medication adjustment Treatment of any medical problems Daily monitoring of bowel movements. (see more suggestions under constipation) Maintain structure and predictability. Daytime activities and social interactions Exercise Maintain routine – minimize change 	<p>Planning for Retirement – what does this look like?</p> <ul style="list-style-type: none"> Keep socially connected Physically active Weight under control Management of health issues <p>RESOURCE: PWS Mental Health Guidebook (fpwr.org)</p>
Health Concern	Risk Factors in PWS	Screening / Monitoring	Prevention & Management	Other
Worsening Mental Health cont. <ul style="list-style-type: none"> Depression Low Energy Dementia <p>Dementia may begin with low energy and apathy but advances to loss of basic skills.</p>	<ul style="list-style-type: none"> Few cases of dementia in PWS have been reported Often difficult for persons with PWS to communicate their feelings and needs 	<ul style="list-style-type: none"> DLD- dementia questionnaire for people with learning disabilities that focuses on changes in behavior and activities of daily living over time 	<ul style="list-style-type: none"> If dementia is noted – may need to increase oversight and assistance with daily living skills. 	

IMPORTANT RESOURCE IN EDUCATING FAMILY MEMBERS, HEALTH CARE PROFESSIONALS AS WELL AS YOURSELF IS THE PWS MEDICAL ALERT BOOKLET: [MedicalAlertsBooklet-GIChart-2022.pdf \(pwsausa.org\)](https://www.pwsusa.org/medical-alerts-booklet-gi-chart-2022.pdf)

We continue to learn more about the aging process and issues that are facing individuals with PWS. However, just like all of us, we are individuals first and the aging process can be a very individual experience. Some age gracefully; while others face great challenges.

Aging Research in Persons with PWS – Bibliography

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