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**Aging in the Adult with Prader-Willi Syndrome – A Focus on Physical and Cognitive Health**

*By Barb Dorn, R.N., B.S.N., Verona, Wisconsin*

*and Elizabeth Moser, M.S.W., L.C.S.W., Oconomowoc, Wisconsin*

*(This is the first in a series of three articles about aging in adults with PWS.)*

As adults with Prader-Willi syndrome (PWS) grow older, we are learning more about some of the common health issues many are facing. Researchers are starting to investigate the aging process in persons with PWS. It can be challenging to distinguish between health concerns that are inherent to PWS versus concerns that we all face as we age. Family history is also contributory to many health concerns and must also be considered.

This article will highlight some of the more common physical and cognitive health issues being identified by researchers as well as professionals who are working with this aging population. According to Eiholzer and Lee (2006), some of the main medical problems being reported in adults with PWS include physical disability (resulting in mobility issues), respiratory insufficiency, osteoporosis and scoliosis. The major cause of overall death in adults with PWS was respiratory insufficiency triggered by acute or chronic lung infections – not cardiopulmonary disease as originally hypothesized.

During the 2010 International Prader-Willi Syndrome Organization conference in Taiwan, researchers from The Netherlands shared findings from their study of 102 adults with PWS ages 18-66 years. Sinnema, Maaskant, et al. reported a majority (56%) were obese. In addition, the following health conditions were common: leg edema (56%), erysipelas - an acute streptococcal infection of the skin (38%), constipation (38%), diabetes mellitus (17%), osteoporosis (16%), pneumonia (14%) and hypertension (9%). There was also a high incidence of psychiatric episodes.

According to Rena Mills, R.N., Health Services Coordinator for Prader-Willi Homes of Oconomowoc (PWHO), which provides residential services to over 80 adults with PWS, premature aging seems to be a concern. By the time their residents are 40, many have medical problems like those who are 60. Mills also identified many of the same health problems as mentioned above – osteoporosis/osteopenia, constipation, intestinal blockages, pneumonia, diabetes, cellulitis, low sodium levels and high/low blood pressures.

Changes in cognitive function are also being examined to determine if the early onset of dementia may be an additional concern for adults with PWS.

Studies have shown that adults with Down syndrome (DS) are more likely to develop Alzheimer-type dementia than their peers without DS (Silverman, Zigman, Krinsky-McHale & Schupf, 2008). Still other research suggests the prevalence of dementia in adults with intellectual disabilities (ID) who do not have DS is higher than the general

population (Strydom, Chan, Fenton, et al., 2010). What does this mean for aging adults with PWS?

In March 2009 the State of Science on Dementia in older adults with Intellectual Disabilities reported, “aging and cognitive functioning has been studied in so few of the ID syndromes other than DS.” Sinnema and her research team from The Netherlands contributed to this critically-needed knowledge base with their 2010 case study of a 58-year-old woman with PWS. The resulting assessment scores supported the presence of dementia.

PWHO currently supports 27 (33%) individuals with PWS in their 40s and 2 (2%) in their 50s; one woman was diagnosed with dementia at age 54. An assessment procedure was developed to measure cognitive changes as they age, so the possible onset of dementia can be detected and early treatment implemented. This data will also be a future resource for understanding how aging influences the cognitive process in PWS.

Knowing what physical and mental health issues are common allows us to initiate preventive measures and plan appropriately for future care needs. The second article in this series will focus on five of the most common health issues being seen in aging adults with PWS along with strategies to prevent and/or minimize their effects. The third will focus on dementia research, assessment and treatment. This collaborative series will provide insight into how aging affects adults with PWS and the approaches to help to ensure they can remain healthy, productive members of their communities.

*July August 2011*

## **Physical Health Issues and the Aging Adult with Prader-Willi Syndrome**

*By Barb Dorn, R.N., B.S.N. Nurse Clinician,*

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*(second in a series of 3 articles that share information about aging in adults with PWS with a focus on health.)*

Many physical aspects of Prader-Willi syndrome impact body systems that can result in chronic health problems with increased risk of complications as a person ages. Obesity impacts most (if not all) aspects of health. Low muscle tone and spine deformities can contribute to respiratory and mobility issues. Hormonal abnormalities, low activity levels and nutritional deficiencies can result in low bone density and an increased risk of fractures. Sedentary lifestyle may contribute to lower extremity swelling and poor circulation. Diabetes is common, contributing to problems with eyesight, wound healing and other vascular issues. Constipation and other bowel problems may result from diet and low fluid intake as well as poor bowel habits.

**The following are five of the most common health concerns being seen in adults with PWS as they age.**

Prevention and management suggestions are also included.

**Respiratory Insufficiency** is the major cause of death in adults with PWS, triggered by acute or chronic lung infections – not cardiopulmonary disease as originally believed (Eiholzer and Lee 2006). Spine deformities including scoliosis and kyphosis (“hunchback” deformity) can impact lung expansion and put a person at greater risk for

respiratory compromise. Evaluation and treatment of sleep apnea, which can increase morbidity and mortality, is important, especially when an adult is undergoing a surgical procedure. Aggressive measures should be taken to prevent and manage lung infections and chronic lung conditions. Prompt evaluation of upper respiratory infections may necessitate initiation of antibiotics, encouragement of fluids, and/or assurance that the adult is kept moving and expanding their lungs. Follow-up with medical specialists is important. Discussions with health care professionals should take place on the administration of annual influenza immunization as well as the pneumonia vaccine.

**Low Bone Density, Degenerative Changes and Mobility Issues** can affect the aging population in general, with those with PWS at greater risk. The normal aging process results in decreased hormone production, arthritic changes and a combination of other risk factors that contribute to mobility issues and increase the risk of falling. Today, the use of growth hormone and supplementation of estrogen and testosterone in adults with PWS is more common, along with calcium and Vitamin D supplements and a greater focus on proper nutrition in an effort to prevent osteoporosis. Closer monitoring of bone density and the use of appropriate treatment is becoming more prevalent. Weight-bearing activities as well as medications are being tried to treat osteoporosis. Osteoporosis should be documented before initiating these medications, many of which have significant side effects. An increasing number of adults with PWS are undergoing orthopedic surgeries to treat degenerative joint problems. Keeping a person mobile is critical. When an adult with PWS becomes dependent upon a wheelchair, their risk of developing other medical emergencies (blood clots in legs and/or pulmonary emboli) is greatly increased.

It is important to create environments that reduce the risk of falls, such as guardrails installed in bathrooms and other areas, removal of throw rugs and proper lighting (especially at night). Canes and walkers may be needed to stabilize balance. Proper fitting shoes with a good tread can also help. All medications should be assessed for their impact on balance. Vision should be checked, and because bifocal lenses can alter depth perception and increase the risk of falling, two pairs of glasses may be preferable. A one-story home or home with ramps and other measures to assist with mobility should be considered.

**Peripheral Vascular Problems**, including ankle and lower extremity swelling and problems with blood pressure, are being reported in many aging adults with PWS. These types of circulation problems are often seen in diabetes. Vascular problems also impact wound healing and can result in leg ulcers and cellulitis. Skin picking problems can become very severe. Preventing circulation problems is optimal. Walking is one of the best activities to promote lower extremity circulation. Prolonged sitting should be avoided. When sitting, make sure feet are resting on the floor or stool – avoid “dangling legs”. In severe cases, special stockings may be prescribed. However, caution must be used to make sure these stockings do not impair circulation further or cause problems with wound healing. In some cases, these stockings have been used as a barrier to skin picking. Prompt attention to any cuts or sores in lower extremities and feet should take place. Avoid open-toe shoes-- especially thong-type sandals--to prevent injury to skin. These types of shoes may also cause balance and stability problems. Nail care is imperative and should be done by a health care provider if the adult has diabetes. Hydration is important to assist with maintenance of blood pressure. Weight loss, exercise, lower salt intake, frequent monitoring of BP measurements are all important. Medication to lower elevated BP may be needed.

**Complications of Diabetes** can often be severe and impact every body system. According to the American Diabetes Association (ADA), the aging population, especially those who are overweight, may develop Type 2 diabetes. High levels of glucose in the bloodstream damages blood vessels throughout the entire body—resulting in vision problems, kidney disease, and heart and vascular problems including stroke, high blood pressure and heart disease. Diabetes also impacts circulation and impairs healing – especially in lower extremities. Adults with PWS can be at increased

risk to develop diabetes, especially if they are overweight. Many persons with PWS are diagnosed with diabetes at a young age. This can add years of damage to blood vessels. Screening, early diagnosis and treatment are important. The ADA recommends screening with a blood test called Hemoglobin (Hgb) A-1C. This should be done 1-2 times a year depending on risk factors. Annual blood lipid and cholesterol testing is suggested. Diet and weight management is imperative. An active lifestyle including exercise is also important. Once diagnosed with diabetes, the adult with PWS should adhere to any diet and/or medication regime that is prescribed.

**Changes in Bowel Habits – Constipation** is another health problem that many adults face as they grow older. Persons with PWS of all ages seem to struggle with gastrointestinal issues; constipation is common. Many of the same interventions used to manage other health issues also help this problem. Exercise, an increased activity level, adequate fluid intake, high fiber foods are just a few suggestions. Probiotic foods and supplements have also been beneficial. In many cases, daily fiber and/or laxative supplementation may be needed. Staff may need to monitor the frequency and consistency of a resident's stool. Higher incidences of intestinal obstruction and ileus (slowing or stoppage of the intestine) have also been reported. Caution should also be used when using medications that slow the intestine – especially medicines used to treat diarrhea. Diarrhea can be a symptom of intestinal blockage.

*Many of the issues are the same as all of us age. The goal is to minimize problems so aging adults can maintain a higher quality of life.*

*Sept/Oct. 2011*

## **Cognitive Changes in the Aging Adult with Prader-Willi Syndrome**

*By Elizabeth Moser, M.S.W., L.C.S.W. - Psychotherapist with Prader-Willi Homes of Oconomowoc, WI*

*(This is the third in a series of three articles that shares information about aging in adults with PWS with a focus on dementia.)*

With the increased awareness of Prader-Willi syndrome (PWS) leading to early diagnosis and treatment, life expectancy has grown dramatically over the years. When I first began working with adults with PWS in 1989, I learned that many did not live past their teen years due to obesity-related medical complications. Back then, I worked with 15 people diagnosed with PWS, most in their early twenties.

Twenty-plus years later, I am thankful to still work with many of those individuals, as we step into our forties together. We are entering the uncharted years of aging in PWS with the aim to achieve the same outcome as the frontier movements of awareness--early detection and treatment--to ensure continued quality of life.

This article explores how aging may influence cognitive processes in PWS and how we can prepare for future needs.

### **What is Dementia?**

According to Wikipedia, *“dementia is a serious loss of cognitive ability in a previously unimpaired person, beyond what might be expected from normal aging.” There are several types of dementia, but the most common is Alzheimer’s (AD), which occurs in “50-80 percent of dementia cases.” It is a progressive disease with symptoms getting worse over time (www.alz.org).*

### **Dementia Prevalence:**

In the general population, Alzheimer-type dementia usually occurs after the age of 65 (alz.org). Dementia studies

are not as abundant in the area of intellectual disability (ID) syndromes compared to the general population studies, but in March 2009, the State of Science on Dementia released a comprehensive review of studies from 1997-2008 relating to aging and intellectual disabilities.

This 2009 review reported that “the prevalence of dementia (particularly AD) among the ID population may differ from the general population, at least in specific subgroups such as Down syndrome (DS)” (Zigman, Schupf, Haveman, & Silverman, 1997). Later studies showed that the prevalence of dementia in the ID population was 6.1% in those aged 60 and over, which is comparable to the percentage in the general population (Janicki and Dalton 2000). The same study showed that adults with DS had much higher rates of dementia--56% for those 60 and older.

#### **The mean age of dementia onset:**

- General population: 67 years
- Intellectual Disability excluding DS: 67.2 years
- Intellectual Disability with DS: 52.8 years
- Intellectual Disability with PWS: Unknown

#### **PWS Research Overview:**

The State of Science on Dementia cited one PWS study by Sinnema, Maaskant, Van Schrojenstein, et. al. in 2008, which evaluated 74 individuals with PWS ages 18-63 years old and reported no cases of dementia.

At the IPWSO conference in Taiwan last year, several studies examining aging in PWS were presented. Whittington and Holland reported the results of their study Recent Mortality Rates and Risk of Dementia in PWS. They found that out of the 26 individuals who were 40 years and older: 22 with no signs of dementia, one case of mild-moderate dementia, one potential case of mild dementia and evidence of cognitive decline in one person.

Sinnema and her research team from the Netherlands presented a case study of a 58-year-old woman with PWS. The assessment scores supported “the presence of dementia in very late stages.”

The researchers agree that more studies are needed to understand how aging will affect older persons with PWS.

#### **Preparing for the Future:**

Prader-Willi Homes of Oconomowoc (PWHO) currently supports 81 adults in residential care: 27% are in their 30s, 33% are in their 40s, and 2% in their 50s. One woman was diagnosed with dementia almost three years ago at age 54.

According to the article Stopping Alzheimer’s Before it Starts posted on [www.medicinenet.com](http://www.medicinenet.com), “the prevention process should begin at approximately age 40. That is because on the average, Alzheimer’s disease begins 30 years before the first symptoms appear.” Because it is unknown when onset may occur, PWHO has taken the approach of evaluating cognitive changes over time with the aim of early detection and treatment.

#### *The fundamentals of this procedure are as follows:*

1. All clients have a mental status baseline score from “The Short Portable Mental Status Questionnaire” (SPMSQ).

Date of assessment and cognitive range (normal mental functioning, mild, moderate or severe cognitive impairment) corresponding with score on the questionnaire are documented.

2. All clients thirty-five years and older complete the SPMSQ annually. If there is a change in range, further evaluation occurs (i.e. retesting, additional assessments, psychotherapist consult, psychiatrist consult, neurologist consult).

3. Clients that present with any significant behavioral and/or mental health changes complete the SPMSQ to assist in determining the possible cause(s) of decline in functioning.

4. If cognitive range declines and other medical or mental health causes have been ruled out, more in-depth assessments that measure cognitive and daily functioning are administered.

To date, the assessment scores have shown no indication of early onset of dementia in the 30s or 40s.

In diagnosing dementia and evaluating potential risk for early onset, an important contributing factor is family history.

### **Treatment:**

*The National Institute on Aging (NIA) has the following statement on their Web site, [www.nia.nih.gov](http://www.nia.nih.gov):*

AD is a complex disease, and no single “magic bullet” is likely to prevent or cure it. That’s why current treatments focus on several different issues, including helping people maintain mental function, managing behavioral symptoms, and slowing AD.

Visit [www.webmd.com](http://www.webmd.com) under Alzheimer’s Disease: Daily Care of the Alzheimer’s Patient for support recommendations. One of the most important suggestions for caregivers is “to understand and act according to your own physical and emotional limitations. Be sure to take care of yourself, and allow yourself periods of rest and relaxation.” Other factors in caring for somebody with AD are exercise, nutrition, socialization, structure, routine, and visual cues. As a PWS community, we are already one step ahead of managing the symptoms of dementia, as the approaches are similar to the ones effective in supporting persons with PWS.

In addition to daily supports, medication has also been used to manage and potentially slow down AD. The State of Science on Dementia reported, “Donepezil is the most commonly used anti-dementia drug used in intellectual disabilities to treat dementia, and there is some evidence for rivastigmine.” The woman diagnosed with dementia residing at PWHO has had a positive response to receiving donepezil (Aricept).

As treatment strategies are planned, it is important to remember that dementia is a progressive disease in that “lost skills cannot be regained” ([www.webmd.com](http://www.webmd.com)). It is crucial to incorporate and continue with activities that are physically and mentally stimulating per individual’s preference.

### **Summary:**

More studies are needed to understand how the aging process will affect persons with PWS. There is not enough information to make any conclusive statements about how or when dementia will present.

Based on my review of the literature and observations at PWHO, there is no evidence to suggest that dementia in PWS is comparable to Down syndrome with the potential age of mid-30s onset. Further research is needed to evaluate whether or not the onset could occur earlier than other ID syndromes and the general population.

Like PWS, dementia has no cure, but with early detection and the necessary supports in place, we can provide the best opportunity for the physical, mental and emotional well-being of the individual.

As we make this venture into unknown territory together, we will continue to do what the PWS community does best--unite forces--by sharing experiences, contributing to the knowledge base, applying what is successful and growing together--stronger and more informed than the day before.

Nov/Dec 2011

## Book Review

# PRADER-WILLI SYNDROME: GROWING OLDER

*By Terrance James, Ph.D.,*

*Reviewed by Barb Dorn, R.N.*

Prader-Willi Syndrome: Growing Older begins to explore a topic that is becoming very pertinent—our aging population of adults with PWS. James interviewed parents, caregivers and 14 adults with PWS ages 40+ years who reside in western Canada. He covers a variety of topics that impact them, including services, genetics, health, residential supports, and employment as well as their aging parents and/or caregivers. He also focuses on quality-of-life issues and shares the personal views and experiences of these adults.

The first few chapters help readers gain a historical perspective on the changes and advances in the field of developmental disabilities and PWS. “Younger” readers will learn to appreciate the advances in genetics and diagnosis of PWS. (Most adults over 40 did not, and many still do not, have genetic confirmation of the diagnosis). James briefly touches upon the many areas impacted by the aging process, such as cognition, behavior, physical health, mental health. He shares research resources from all over the world that support his findings.

James devotes one chapter to aging parents – an emotional topic near and dear to all parents of adults with PWS. He touches on several concerns about the future including family, residential and funding concerns of government programs. Even though most references are for Canadian programs, it was interesting to learn that we share many of the same challenges and worries.

My favorite part is the chapters about the personal life experiences of 14 older adults. They share their accomplishments and what has helped to improve their quality of life. It is enlightening to read about the various models of residential support and great to see how many are taking advantage of volunteer opportunities in their communities to keep them connected to social and cognitive stimulation when employment was no longer a full time option.

Although the book focuses on aging adults with PWS in Canada, the topics, issues and concerns cross national boundaries. It is an initiative toward the research, discussion and attention that are needed to address this important topic.