Aging in Prader-Willi Syndrome

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Snapshot of Adult PWS Today

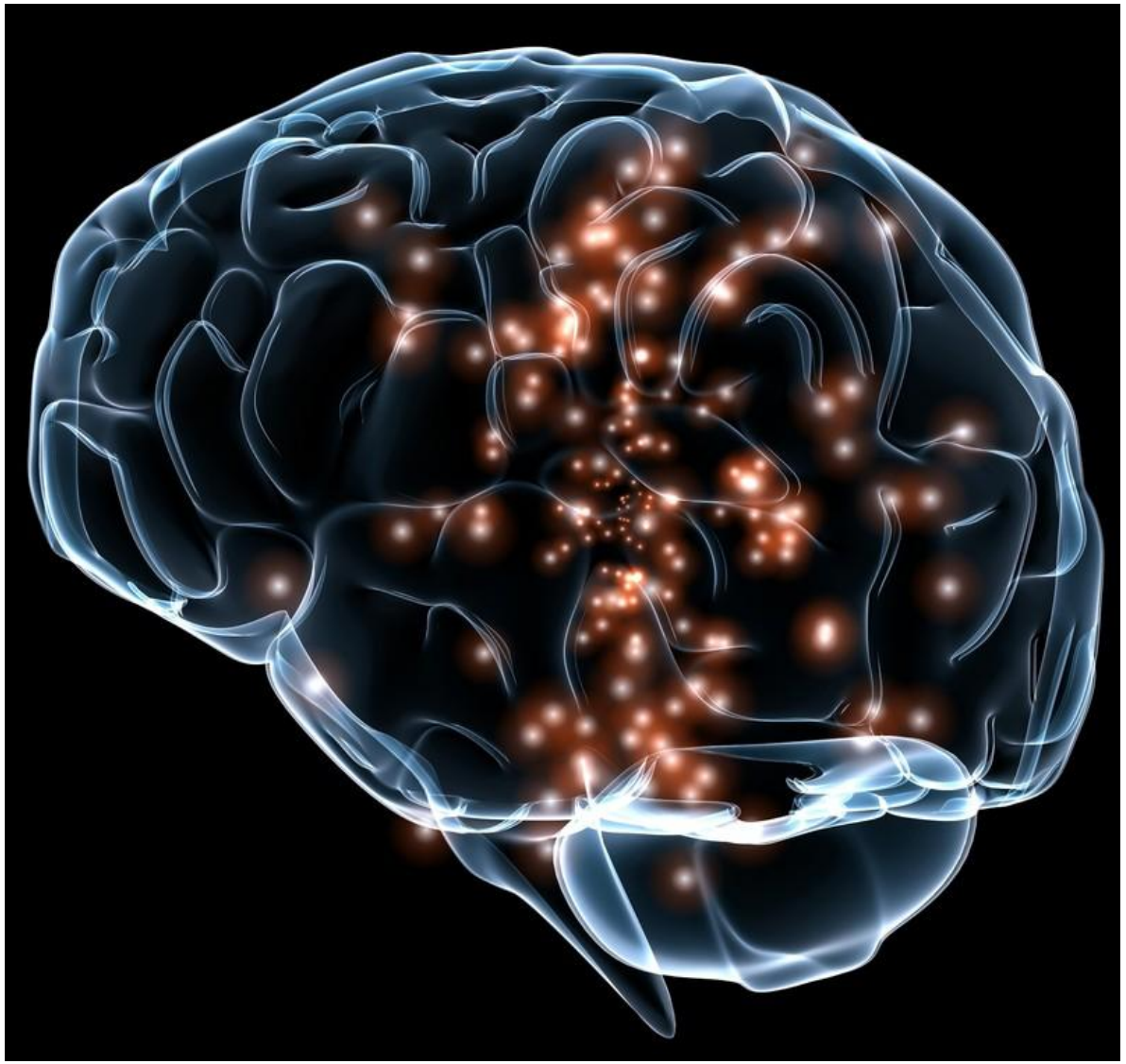
- Life expectancy improving; still reduced vs. general population globally
- Top risks: respiratory disease, cardiometabolic disease, GI emergencies
- Bone health, sleep disorders, and behavioral/cognitive changes matter for function

Drivers of Morbidity in Later Life

- Respiratory: OSA, hypoventilation—optimize CPAP/BiPAP & airway care
- Hyperphagia-related risks persist: choking, gastric dilatation/perforation
- Cardiometabolic: Type II diabetes, hypertension, heart failure

Premature Aging

- Neuroimaging studies show ↑ “predicted brain age” in adults with PWS
- Shorter leukocyte telomere length suggests early vulnerability to neurodegenerative changes
- Clinical observations: memory lapses, confabulation; dementia not a hallmark but monitor
- Functional implications: planning, independence, caregiver supports



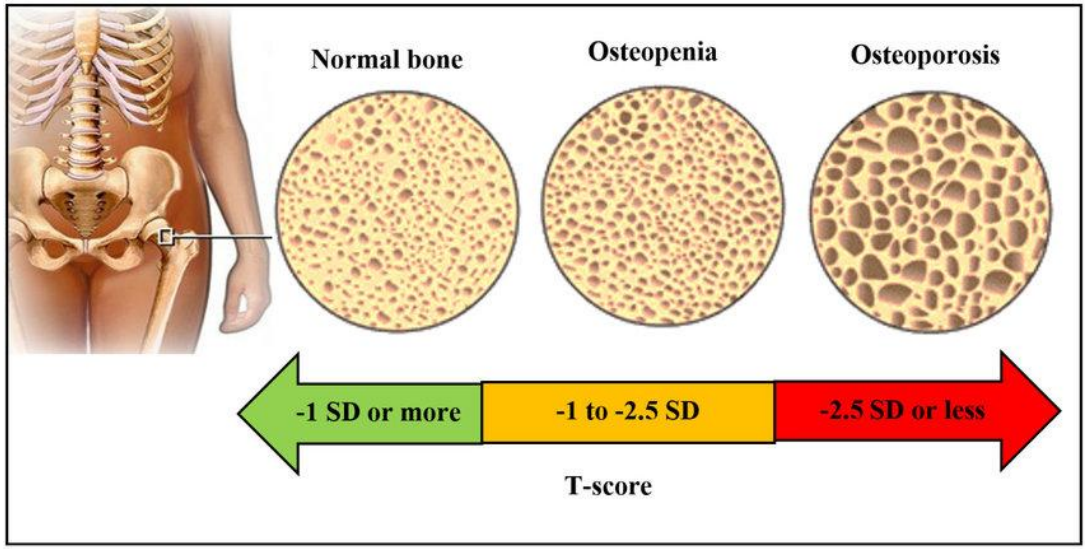
Biological systems affected

- Decreased mitochrondrial efficiency- meaning, just like a machine that hasn’t been cleaned or oiled in a long time
- The hypothalamus governs growth hormone, thyroid, adrenal, and gonadal function. Long-term dysregulation due to loss of PWS region genes can lead to: Low anabolic hormone levels Altered body composition including loss of muscle mass and obesity. These endocrine features resemble premature or accelerated aging
- When sleep and body clocks don’t work right because of missing genes, the body can’t repair itself as well, gets more inflammation, and starts to age faster

Body Composition

- Low fat-free mass and reduced resting energy expenditure
- Strength & balance decline → falls, mobility limits
- Interventions: resistance training, protein adequacy, vitamin D

Bone Health Across the Lifespan



- High vertebral & non-vertebral fracture burden; screen with DEXA
- Contributors: hypogonadism, hypotonia, reduced loading/activity
- weight-bearing/resistance exercise; fall prevention
- low muscle mass + hormone deficiencies + obesity = risk of fractures.

Sleep & Aging

- High prevalence of OSA and hypoventilation—reassess over time
- Therapies improve daytime function and may reduce cardiometabolic risk
- Support adherence: mask fit, desensitization, tracking

Endocrine in Mid-/Late Adulthood

- Hypogonadism common → bone & body composition effects
- Adult GH deficiency: consider individualized evaluation/management hormones + optimizing vitamin D/calcium + activity matter.
- Re-check thyroid and adrenal axis when clinically indicated—avoid over/under-treatment

Emerging Therapy & Trajectory

- Vykat XR (diazoxide choline XR) FDA-approved for hyperphagia (≥4y) in 2025
- Potential long-term impact: weight, cardiometabolic burden, caregiver load
- Prioritize real-world monitoring: efficacy, AEs (edema, glycemia), QoL outcomes
- Ongoing clinical trials

Prevention Priorities

Blood Tests (yearly)

- Hemoglobin and blood cell count
- Na+, K+, creatinine, liver enzymes
- Fasting blood lipids
- Hemoglobin A1c and fasting blood glucose
- Vitamin D , calcium
- Thyroid function testing (TSH, free T3, free T4)
- Sex hormone profile (LH, FSH, Estrogen in women and fasting Testosterone in males)
- IGF-1: for those treated with growth hormone, confirming dose and compliance

Other recommendations

- Ophthalmology examination - every 2 to 3 years
- Dental examination and hygiene - twice yearly or more often if needed
- Hearing evaluation - in those with poor speech and those over 50 years of age
- Mental state - If there has been a sudden and persistent change in mental state or behavior assess for the presence of abnormal mental beliefs or experiences, suggestive of the onset of mental illness
- Echocardiography - every 2 to 3 years particularly if obese

Thinking of the future

- Ongoing well rounded medical care
- Guardianship
- Placement decisions
- Financial
- End of life decisions
- Staying connected

Questions