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A 68-year-old white female with Prader–Willi syndrome

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Abstract

A 68-year-old white female with Prader–Willi syndrome is described. The clinical features are described and the progression of her condition is discussed and illustrated.

Keywords

Prader–Willi syndrome; cataracts; 15q11-q13 deletion; natural history

Prader–Willi syndrome (PWS) is generally characterized by infantile hypotonia, feeding difficulty, hypogonadism, hyperphagia, early onset of childhood obesity, short stature, small hands/feet, mental deficiency and minor anomalies (Cassidy, 1984; Butler, 1990; Thompson *et al.*, 1996). Approximately 70% of PWS subjects have a paternal chromosome 15q11-q13 deletion while the remaining individuals generally have maternal disomy of chromosome 15 (Nicholls *et al.*, 1989, 1998; Butler, 1990; Mascari *et al.*, 1992). Prader–Willi syndrome is the most common syndromic cause of marked human obesity and obesity is the most significant health problem in the affected individuals (Butler, 1990). Generally, individuals with PWS are thought to have a normal life span if the obesity is controlled through diet restriction and increased physical activity. Because of a paucity of reports of older individuals with PWS, the natural history, onset and type of age related problems are not clearly understood. Therefore, we describe a 68-year-old white female with PWS, to our knowledge the oldest PWS person reported.

NA was born full-term to a 30-year-old mother and 31-year-old unrelated father. The family history was unremarkable. She had hypotonia at birth and feeding difficulties during infancy. Developmental delay was noted during infancy and obesity developed in early childhood. She attended elementary school through grade 7 and received vocational training. She had the typical PWS facial features including a short upturned nose, down-slanted corners of the mouth, narrow forehead, light hair and skin complexion and sticky saliva (see Figure 1). She also had small hands and feet. PWS was diagnosed at 48 years of age and recently chromosome studies showed the typical 15q11-q13 deletion and 45% of her lymphocytes studied were missing an X chromosome. She lived with her parents until their death approximately 10 years ago. Since that time, she has lived with her sister or in a nursing home setting. She was relatively healthy throughout her life but developed bilateral cataracts

which were removed at 51 years of age and intraocular lenses implanted. Intermittent right exotropia developed requiring surgical intervention at 52 years of age along with open angle glaucoma. Type II diabetes mellitus was diagnosed during her sixth decade of life which has been kept under diet control along with arthritis and hypertension. She was grossly obese at that time and weighed approximately 114 kg with a maximum weight of 130 kg at 55 years of age. She required periodic hospitalizations for weight reduction during late adulthood. She has been treated for hypothyroidism for the past 15 years. She has had the typical behaviour of persons with PWS including stubbornness, rigidity, food foraging and hoarding, and skin picking. She enjoys working with jigsaw puzzles and crafts. She now weighs 99 kg (>95th centile) and is 150 cm tall (<5th centile) and lives in a nursing home. She wears a size 6 extra-wide shoe. She has remained alert and has not had mental regression.

In conclusion, there have been no problems recognized in this patient that would be different than other similar obese individuals at comparable ages in the general population. Other health care providers are encouraged to report older PWS subjects in order to describe and better understand the natural history of PWS subjects particularly as they become older. This information will be helpful in the health care management of older Prader–Willi syndrome subjects and for discussion with family members.

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FIGURE 1.
Frontal photographs of the 68-year-old individual with Prader–Willi syndrome taken at different ages