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Prader-Willi syndrome: Are there population differences?

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Abstract

A 15½-year-old black female with features consistent with the Prader-Willi syndrome is reported. This is the second case report of a black individual and the first case of a black female with the Prader-Willi syndrome. There is an apparent paucity of blacks reported with this condition. Whether this difference is a true difference or represents under-reporting is not known. We urge reporting of individuals representing other racial groups with this disorder and suggest population studies to determine the incidence as well as the true population difference in the Prader-Willi syndrome.

Keywords

Black female with Prader-Willi syndrome; population differences

Prader-Willi syndrome was first described in 1956 (Prader et al. 1956) and subsequently there have been over 300 cases reported (Naselli et al. 1981). The syndrome is characterized by nonprogressive infantile hypotonia, onset of obesity in early childhood, mental deficiency (intelligence quotient of 20-80, with a mean of 60), short stature, small hands and feet, and hypogonadism. At present the cause(s) is unclear, although a chromosome 15q deletion has been reported in 50% of the patients (Ledbetter et al. 1982). Thus, it is of interest that among the 300 Prader-Willi cases reported, only one individual was black (Norman 1979). In addition, we are unaware of any reports of this syndrome in the African populations. Additional reports for other racial groups are obviously needed to analyze the possibility of population differences in the incidence of Prader-Willi syndrome and perhaps provide a clue to the syndrome's etiology. We report here the second case of a black individual and the first case of a black female with the Prader-Willi syndrome.

Case Report

This 15½-year-old black female was born after a 36-week gestation to a P0G1 Ab0 black female. The labor and delivery were uncomplicated. She weighed 2.07 kg (< 3%ile). The father was not a relative and the family history was unremarkable. A younger brother is of normal weight and in good health. The mother smoked 1/2 pack of cigarettes a day during the pregnancy. Medications for constipation, iron and vitamin supplements were taken

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during pregnancy. The fetal activity was considered to be poor. Hypotonia, feeding difficulties and a diminished cry were noted during the first few months of life. She had a poor suck and was lethargic in the neonatal period. She showed delayed development with rolling over at 9 months and walking at 2 years. She was underweight until about 2 years of age, when her appetite increased and she experienced a rapid and steady weight gain. By 4 years of age, her weight was considered excessive. There is a history of sneaking food. Weight control around 7 years was moderately successful but no control has been placed on the patient since the age of 11 years. There is a history of strabismus of the left eye which was surgically corrected. She has not had menarche.

Examination of the patient revealed a pleasant, obese, mentally retarded black female. She was normocephalic and had a height of 141.5 cm, a weight of 52.03 kg, and a blood pressure of 110/70. Her height is below a — 3 SD for black girls, while her weight is at the 50th percentile (National Center for Health Statistics 1973). This relationship places her weight between the 90th and 95th percentiles for her height age of 10 years based on data for U.S. black females (National Center for Health Statistics 1974). Stated another way, her ponderal index ($\text{height}/\sqrt[3]{\text{weight}}$) is between the 5th and 10th percentiles for the height age, an indication of increased laterality of body physique when compared to normals (National Center for Health Statistics 1974).

In addition, she had full cheeks and chin, almond-shaped eyes, abdominal obesity and small hands and feet (15.5 cm and 21 cm, respectively) matching those of an average black female at 9 years of age (National Center for Health Statistics 1974). The fingers appeared short in relation to the palm. The rest of the physical examination was unremarkable and in particular there was no scoliosis. She had moderate breast development as well as pubic and axillary hair growth. Blood could not be obtained for chromosome analysis.

Discussion

The patient presented in this paper had the typical characteristics associated with Prader-Willi syndrome, i.e., hypotonia improving with age, early poor feeding and lethargy, psychomotor and mental retardation, delayed sexual development, early childhood obesity, excessive appetite, short stature, small hands and feet and typical facial appearance including almond- shaped eyes. Therefore, we feel that the clinical findings support the diagnosis of the Prader-Willi syndrome.

The etiology of the Prader-Willi syndrome is unclear although an interstitial deletion of the proximal long arm of chromosome 15 has been reported (Ledbetter et al. 1982). We have focused our attention on chromosome 15 with double-blind studies and have also found the deletion in about 50% of our patients with Prader-Willi syndrome. Anthropometric measurements and clinical observations of our patients support the existence of two separate and distinguishable subgroups based on the chromosome findings of this syndrome (Butler et al. 1982). Several monozygotic twins (Naselli et al. 1981) and other family members (Hall & Smith 1972) have been reported with this syndrome, although most affected individuals are sporadic occurrences.

With regard to population differences in the Prader-Willi syndrome, there is an apparent paucity of blacks with this disorder. Whether this difference is a true difference or merely represents under-reporting is not known. If it is real, then it may be possible to recognize a factor(s) which is present in whites, and not usually in blacks, as the underlying cause of the Prader-Willi syndrome. Thus, we urge reporting of individuals with this disorder and, in particular, population studies in order to determine the incidence as well as the true population difference in the Prader-Willi syndrome.

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