

Hypothermia In Prader-Willi Syndrome

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The Children's Institute (TCI) has had a program for persons with Prader-Willi syndrome (PWS) since 1985 and has seen more than 400 patients ranging in age from a few months to 60 years.

Percentage overweight has ranged from 20 percent to more than 300 percent. From this population, two patients age 40 and 46 years were recognized to have a recurrent hypothermia syndrome. They both had the onset of this hypothermia in late November while living in the Mid-Atlantic area of the U.S. They had repeated episodes during the winter and experienced relief during the summer months and then had recurrence during the following late fall.

The 46-year-old man died abruptly over 4-6 hours the following October. This occurred several months after discharge from TCI, and it is unknown if hypothermia was present.

The 40-year-old female had recurrence of symptoms the next winter and still survives.

Both had been on psychoactive drug(s) that might have retarded shivering; tardive dyskinesia was previously mentioned. However, withdrawal of these medications did not prevent recurrence of hypothermia, and persisting frank tardive dyskinesia after medication withdrawal was not observed. Data was obtained from both outside medical records and direct observation at TCI.

Hypothermia syndrome began with a change in behavior followed by decreasing activity proceeding to near coma. Hypothermia (81 — 94°F), decreased blood pressure, bradycardia and slow respirations were discovered. Lab studies revealed decreased hemoglobin, low WBC, decreased platelets, hyponatremia without acidosis or hyperkalemia, and elevated renal and liver function tests. All of these changes return to normal levels over several days as the patient is rewarmed. Sepsis was frequently suspected, but blood cultures were negative. Both hypothermic patients were noted at TCI to have ear probe temperatures from 91-101°F but without the resultant syndrome of mental decline, functional deterioration and lab changes.

In order to more broadly define the problem of hypothermia in persons with PWS, nine patients were surveyed over 24 days. Six patients dropped their temperature to 92 — 95° when ambient temperatures were below 66°F, despite walking vigorously and appropriately clothes in an exercise program. Following the outdoor walk and inside the building at 72°, all body temperature were back in the range of 98-99°F. This group confirms the reports of Bray et al (Medicine 1983).

Webb (Amer J Physiol 1995) presented heat regulation as a physiologic method of handling metabolic heat rather than temperature regulations per se, i.e., heat production from metabolic activity is the primary variable, with the resultant body temperature a function of heat loss.

In the person with PWS there is hypotonia and progressive obesity with inordinate drive to get food, yet the consequence is hypoactivity, very poor muscle mass/strength, and an apparent inability to maintain body heat, especially if there is a significant environmental gradient.

In these two patients (cited in paragraph two above) episodes of profound hypothermia with multiple abnormalities were noted. When reversed, they resumed their usual PWS baseline state. Of great interest, caloric restriction of 600 calories per day combined with progressive increasing exercise in the Children's Institute PWS Program did not lead to worsening hypothermia.

Does this scenario in persons with PWS suggest a diversion of nutrients from growth and development to useless storage only partly overcome by the drive to "mass action" eating by the patient with PWS, with resulting secondary behavior and hypothalamic consequences, as envisioned by Jackson et al (CIBA Foundation Symposium 1996).

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