

The Italian National Survey for Prader-Willi Syndrome: an Epidemiologic Study

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Twenty-five medical centers and the Prader-Willi Syndrome (PWS) Association collaborated on a study which attempted to identify all people with genetically confirmed diagnosis of PWS living in Italy. Investigators of the participating centers contacted PWS subjects and/or their family, filled in a specially developed form with the required data and forwarded this information by email. The study identified 425 subjects (209 males and 216 females, between the ages of 0.4 – 46.7). Two hundred thirty-eight patients had del15, 104 had UPD15, 4 demonstrated a translocation affecting chromosome 15 and 79 showed a positive methylation test. There were fewer subjects found over the age of 35, probably due to the low rate of identification of older PWS patients as well as the high mortality rate. There were a greater number of male children and adolescents with PWS whilst, amongst adults, there were more females. As expected, the majority of subjects with PWS were obese, especially in adult life. Nevertheless, it is noteworthy that 26% of patients aged between 6 and 17 were normal weight. A total of 212 subjects had received GH treatment, of which 141 were still receiving therapy, while the remaining 71 had stopped. In children and adolescents (233 cases), 89 subjects had never undergone GH therapy. Eighteen PWS patients had died in the past 20 years. Obesity-related cardiovascular and respiratory diseases were the cause of death, both during childhood and after 18 years of age. Three children died suddenly whilst undergoing GH therapy. Respiratory infection and cardiac illness were the causes of death in two cases. There was no definitive cause of death found in the third case. Overall, there was no increase in number of deaths during GH treatment, suggesting that GH administration in patients with PWS, as a group, does not increase the risk of death.

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