Not Prader-Willi syndrome: successful fertility in a patient with an erroneous diagnosis for over 25 years

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We present a 29-year-old man diagnosed as Prader-Willi Syndrome (PWS). In childhood some facial dysmorphic features and learning difficulties were noted but he lacked many of the typical features of PWS with normal appetite, weight and behaviour. The diagnosis of PWS was based solely on chromosomal analysis performed in infancy.

At puberty he had poorly developed secondary sexual characteristics. He was diagnosed with hypogonadotrophic hypogonadism and commenced on testosterone replacement. MRI pituitary was normal. Further investigations revealed hypothyroidism and growth hormone deficiency following which thyroxine treatment was commenced.

He presented age 25 with fertility concerns. On review he lacked features of PWS and pituitary profile was inconsistent with expected results in PWS. Semen analysis off testosterone confirmed azoospermia. Repeat detailed genetic testing showed no evidence of PWS and an alternative diagnosis has been entertained.

He did not attend genetic counselling and underwent fertility treatment without success. Following discontinuation of fertility treatment his wife had a spontaneous pregnancy and later gave birth to a daughter.

This case highlights the importance of revisiting an established diagnosis. Despite the previous genetic confirmation, no clinical evidence supported a diagnosis of PWS. In addition to the ethical issues of inappropriate diagnosis and treatment in a patient with an unclear, potentially inheritable condition consideration must also be given to the appropriateness of offering fertility treatment. The availability of the technology does not absolve the clinician of responsibility to act in a patient’s best interest.