

Swyer syndrome, also known as 46 XY complete (pure) gonadal dysgenesis (CGD), is a rare congenital disorder of sex development. People with Swyer syndrome, however, have an XY genetic makeup, or genotype, but develop a vagina, uterus, and Fallopian tubes instead of a penis and scrotum. This is referred to as a sex reversal, which is also evident in their overall physical appearance, also referred to as phenotype, resulting in unambiguous female external genitalia.