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46,XX Testicular Differences of Sex Development

National Cancer Institute

Source

National Cancer Institute. *46,XX Testicular Differences of Sex Development*. NCI Thesaurus. Code C127170.

Presence of testes in an individual with a 46,XX karyotype, typically associated with translocation of the SRY gene, encoding the transcription factor sex-determining region Y protein, from the paternal Y chromosome to the paternal X chromosome during gametogenesis (SRY-positive). Approximately 15-20% of individuals with 46,XX testicular DSD are SRY-negative. These individuals may have other genetic variations affecting testis determination, such as duplication of the SOX9 gene, which encodes the transcription factor SOX-9.