Related work :

- Previous studies indicated that haploinsufficiency of the short stature homeobox (SHOX) gene leads to the occurrence of short stature and many specific skeletal anomalies in TS individuals.

- mutations in *NKX2.5*, *GATA5*, and *NOTCH1* have been identified as the causative factor in non-syndromic patients with inherited BAV.

- chromosome structural variants and potential pathogenic genes such as *TIMP3* and *TIMP1* may be associated with TS patients with congenital cardiac abnormalities.

- sex chromosome imbalance and dysregulation of certain genes on the X chromosome (such as *FMR1, PDIAPH2*, and *BMP15*, etc.) may result in accelerated oocyte atresia, leading to gonadal dysgenesis later in life.

- haploinsufficiency of a lymphatic gene is related to the development of lymphedema and webbed neck.

- haploinsufficiency of immune-associated genes on the X chromosome was reported to result in the development of autoimmune diseases, including autoimmune thyroiditis, type 1 diabetes and autoimmune enteritis.