## Open Peer Review on Qeios

## PAX8 wt Allele

National Cancer Institute

## Source

National Cancer Institute. <u>PAX8 wt Allele</u>. NCI Thesaurus. Code C97688.

Human PAX8 wild-type allele is located in the vicinity of 2q13 and is approximately 63 kb in length. This allele, which encodes paired box protein Pax-8, plays a role in the regulation of thyroid-specific gene transcription. Mutation of the gene is associated with congenital hypothyroidism non-goitrous type 2, thyroid dysgenesis, thyroid follicular carcinomas and atypical follicular thyroid adenomas. A chromosomal translocation t(2;3)(q13;p25) of this gene and the PPARG gene is associated with a subset of human thyroid follicular carcinomas.