

Tbx3

T-box transcription factor TBX3 is a protein that in humans is encoded by the TBX3 gene.

T-box 3 (TBX3) is a member of the T-box gene family of transcription factors which all share a highly conserved DNA binding domain known as the T-box. The T-box gene family consists of 17 members in mice and humans that are grouped into five subfamilies, namely Brachyury (T), T-brain (Tbr1), TBX1, TBX2, and TBX6. Tbx3 is a member of the Tbx2 subfamily which includes Tbx2, Tbx4 and Tbx5. The human TBX3 gene maps to chromosome 12 at position 12q23-24.1 and consists of 7 exons that encodes a 723 amino acid protein (ENSEMBL assembly release GRCh38.p12).

The **neuroendocrine** system consists of a heterogeneous collection of **neuropeptidergic** neurons in the brain, among which hypothalamic **KNDy neurons** represent an indispensable cell subtype controlling **puberty** onset. Although **neural progenitors** and **neuronal precursors** along the **cell lineage** hierarchy adopt a cascade diversification strategy to generate hypothalamic neuronal heterogeneity, the cellular logic operating within the lineage to specify a subtype of **Neuroendocrine cells** remains unclear. As human genetic studies have recently established a link between **TBX3** mutations and delayed puberty onset, we systematically studied **Tbx3**-derived neuronal lineage and Tbx3-dependent neuronal specification and found that Tbx3 hierarchically established and maintained the identity of KNDy neurons for triggering puberty. Apart from the well-established lineage-dependent fate determination, they uncovered rules of interlineage interaction and intralinear retention operating through neuronal differentiation in the absence of Tbx3. Moreover, they revealed that human TBX3 mutations disturbed the phase separation of encoded proteins and impaired transcriptional regulation of key neuropeptides, providing a pathological mechanism underlying TBX3-associated puberty disorders ¹⁾

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Shi X, Zhuang Y, Chen Z, Xu M, Kuang J, Sun XL, Gao L, Kuang X, Zhang H, Li W, Wong SZH, Liu C, Liu L, Jiang D, Pei D, Lin Y, Wu QF. Hierarchical deployment of Tbx3 dictates the identity of hypothalamic KNDy neurons to control puberty onset. Sci Adv. 2022 Nov 18;8(46):eabq2987. doi: 10.1126/sciadv.abq2987. Epub 2022 Nov 16. PMID: 36383654.

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