PAX3

Nomenclature/External Links

The transcription factor PAX3 regulates neural tube closure and the early development of myoblast and neural crest lineages, including melanocytes. The complex regulation that PAX3 exerts over early neural crest development appears to maintain the stem cell properties of neural crest cells. PAX3 also activates *MITF* and *TYRP1*, thus regulating later stages of melanocyte development. In adult melanocyte stem cells, PAX3 appears to simultaneously allow melanocyte stem cells to remain undifferentiated via *DCT* inhibition, yet committed to the melanocyte lineage by activating *MITF*. *PAX3* haploinsufficiency is associated with Waardenburg syndrome types 1 and 3 and with the Splotch mouse mutant. PAX3 expression is frequently seen in melanoma, and a chromosomal rearrangement resulting in fusion of *PAX3* and the transcription factor *FOXO1A* is seen in alveolar rhabdomyosarcoma. Links are provided below to relevant sequence and gene entries at NCBI, Mouse Genome Informatics (MGI), and the UCSC Genome Browser.

Official Symbol: *PAX3 (Human), Pax3 (Mouse)*

Official Gene Name: Human: paired box 3 (Entrez Gene)
Mouse: paired box gene 3 (Entrez Gene)

Other Names: Sp, Pax-3, splotch, CDHS, HUP2, MGC120381
MGC120382, MGC120383, MGC120384, MGC13477
WS1

Human/Mouse Loci: Human: Chr. 2q35;
NM_181457 NP_852122: PAX3
NM_000438 NP_000429: PAX3a
NM_013942 NP_039230: PAX3b
NM_181458 NP_852123: PAX3d
NM_181459 NP_852124: PAX3e
NM_181461 NP_852126: PAX3g
NM_181460 NP_852125: PAX3h
NM_001127366 NP_001120838: PAX3i
UCSC Genome Browser view (linked to PAX3d; other PAX3 isoforms also annotated)

Mouse: Chr. 1 at 39.79 cM;
NM_008781 NP_032807: Pax3 transcript variant 1 / isoform a
NM_001159520 NP_001152992: Pax3 transcript variant 2 / isoform b
MGI entry: MGI:97487
UCSC Genome Browser view