

PAX3

Expression Pattern

Human

RT-PCR analysis showed the **PAX3 isoform *PAX3a* is expressed only in skeletal muscle, esophagus, and cerebellum, while *PAX3b* is seen in most tissues** ([Tsukamoto et al., 1994](#)).

Two new isoforms of *PAX3* were identified, *PAX3g* and *PAX3h*, bringing the total number of *PAX3* isoforms to seven. RT-PCR analyses showed that ***PAX3c* and *PAX3d* show the most widespread and highest expression of all *PAX3* isoforms**. Expression of these *PAX3* isoforms was analyzed in a variety of neural crest-derived cancer cell lines, including cutaneous melanoma, ocular melanoma, and small cell lung carcinoma ([Parker et al., 2004](#)).

Using immunohistochemistry, early human embryos at Carnegie stages (CS) 12-18 were analyzed in detail for the expression of the neural crest gene regulatory network proteins SOX9, SOX10, PAX3, PAX7, HNK1, AP2Alpha, MSX1/2, and P75NTR. **PAX3 expression was seen in both premigratory (as early as CS12) and migrating neural crest cells, correlating well with PAX3 expression patterns seen in chick and mouse model organisms**. PAX3 expression was also seen in somites, dermomyotome, and the ventricular zone of the neuroepithelium, all tissues not derived from the neural crest. The migrating human neural crest populations expressing PAX3 include dorsal root ganglia, presumptive melanoblasts, and the maxillary process ([Betters et al., 2010](#)).

Using immunohistochemistry and RT-PCR analysis, human PAX3 expression was examined in normal adult melanocytes, nevi, primary melanomas, and metastatic melanomas. PAX3 is expressed in all 3 of these cell types. In normal melanocytes and nevi, PAX3 was co-expressed with MITF, and a variety of combinations of marker coexpression patterns indicated that normal dermal/follicular melanocytes exist in a range of developmental stages. Normal melanocytes show PAX3 coexpression with BCL2L1 while nevi and melanomas show PAX3 coexpression with MCAM, suggesting PAX3 may play roles in normal melanocyte cell survival and melanoma migration ([Medic and Ziman, 2010](#)).

Using immunohistochemistry, PAX3 expression was examined in normal and cancerous human tissues. **Widespread PAX3 expression was seen in normal melanocytes, nevi, and in both primary and metastatic melanomas** ([He et al., 2010](#)).

Mouse

RNAse protection and Northern blot analysis showed that mouse *Pax3* mRNA is highly expressed from E9-E12, then expression decreases until it is absent by E17. In situ hybridization analysis showed *Pax3* mRNA expression beginning at E8.5 and E9 in the **dorsal neural groove and neural tube, and in distinct regions of the diencephalon, midbrain, and hindbrain**. *Pax3* expression continued in the **dorsal developing spinal cord** from E10-E14. During these stages, *Pax3* expression was also seen in neural crest derivatives, including dorsal root ganglia, craniofacial mesectoderm, somatic mesoderm and limb mesenchyme ([Goulding et al., 1991](#)).

A Cre transgenic *Pax3* line, where Cre expression was directed by the proximal 1.6 kb *Pax3* promoter, revealed that PAX3 is expressed in **neural crest cells that give rise to enteric ganglia** ([Lang et al., 2000](#)).

Immunohistochemistry analysis of PAX3 protein expression in mouse showed **expression in melanocyte precursors migrating through the mesenchyme at E12.5, and in the epidermis and in developing hair follicles at E16.5**, in cells that were also positive for DCT and/or c-KIT ([Lacosta et al., 2005](#)).

Because the previously generated *Cre-Pax3* transgenic mice did not show Cre expression in all tissues that normally express *Pax3* ([Lang et al., 2000](#)), a new Cre transgenic *Pax3* line was generated, in which Cre was targeted to exon 1 of *Pax3*. This line showed phenotypes similar to those of *Splotch* mice, and Cre recombinase-mediated reporter gene expression in all expected tissues. This Cre line also revealed new cells that are descendants of *Pax3*-expressing cells in the **urogenital system and epithelial cells in the colon** ([Engleka et al., 2005](#)).

Crossing of the *Pax3Cre* transgenic line generated by ([Engleka et al., 2005](#)) with a newly generated Cre-reporter line (containing a fusion of LacZ and GFP along with a nuclear localization signal at the ROSA26 locus) allowed **enhanced resolution and identification of *Pax3*-expressing tissues** ([Stoller et al., 2008](#)).

Because the alternative PAX3 transcripts PAX3c and PAX3d were the primary forms expressed in cutaneous malignant melanoma, the expression of these transcripts was analyzed during embryonic development of mouse melanoblasts. RT-PCR showed that both *Pax3c* and *Pax3d* were expressed at E11.5, but *Pax3d* was downregulated in skin at E20. Immunohistochemistry showed both isoforms in melanoblasts at E12.5, but by E15 most *Pax3c*-positive cells were in developing hair follicles and most *Pax3d*-positive cells were throughout the epidermis. By E20, *Pax3c* expression persists within the hair follicle, while *Pax3d* expression is gone. Following depilation-induced anagen, *Pax3c* expression was seen at 24hr - 60hr timepoints, while *Pax3d* was seen only at 12hr and 48 hr timepoints. These results suggest different functional roles for these two isoforms during melanoblast development and during adult melanogenesis. Also, both isoforms are seen in the brain and skeletal muscle throughout embryonic development ([Blake and Ziman, 2005](#)).

MGI expression database links

Data summary: Data summary page for all *Pax3* expression data at the Mouse Genome Informatics (MGI) website.

Assays: MGI index of published assays containing murine *Pax3* expression data.

Images: MGI index of published images of murine *Pax3* expression patterns.