

PAX3

Cloning

Initial *PAX3* cloning provided partial sequence information for exons 2-4 of human *PAX3* (first named *HuP2*), and identified the paired domain ([Burri et al., 1989](#)). Identification of *PAX3* mutations in Waardenburg syndrome 1 (WS1) provided further *PAX3* sequence information ([Baldwin et al., 1992](#), [Tassabehji et al., 1992](#)). Subsequently 5' and 3' sequence information ([Hoth et al., 1993](#)) and genomic structure ([Tassabehji et al., 1994](#)) for human *PAX3* was provided.

Mouse *Pax3* was originally cloned from an E8.5 embryonic library, and encodes a 479 amino acid protein ([Couling et al., 1991](#)).

***PAX3* contains 10 exons and displays complex splicing, as at least 7 differently spliced *Pax3* isoforms are generated.** *PAX3a* and *PAX3b* are truncated after exon 4, and lack the homeodomain and transactivation domain ([Tsukamoto et al., 1994](#)). *PAX3c* contains exons 1-8, and terminates within intron 8 ([Goulding et al., 1991](#)), while *PAX3d* contains exons 1-9, and terminates within intron 10. The intronic splicing seen in *Pax3d* shows evolutionary conservation among vertebrates. *Pax3e* contains exons 1-10, with no inclusion of introns. *Pax3f*, reported only in mouse, is missing exons 6-8 and terminates within intron 9 ([Barber et al., 1999](#)). Comparative sequence analysis showed that the splicing sequence necessary to splice intron 9 correctly is present only in human sequence. *Pax3g* is a truncated form of *Pax3d*, and *Pax3h* is a truncated form of *Pax3e*; both of these lack a portion of the transactivation domain in exon 8 ([Parker et al., 2004](#)). Additionally, a glutamine residue may be either present or absent at the 5' end of exon 4 in each isoform; this is noted by the addition of Q+ or Q- to *PAX3* isoform nomenclature, respectively ([Barber et al., 1999](#)).