

102. CLONING AND CHARACTERISATION OF TAMMAR *ATRX* AND *ATRY*

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ATRX (on the human and mouse X chromosome) is responsible for X-linked α -thalassaemia and mental retardation. Male patients with mutations in *ATRX* display gonadal dysgenesis, suggesting that *ATRX* has a fundamental role in the testis development pathway. In the tammar wallaby (*Macropus eugenii*), there are two distinct homologous *ATRX* loci, one on the X chromosome (*ATRX*) and one on the Y (*ATRY*). *ATRX* and *ATRY* were cloned from the tammar by a combination of RT-PCR, 3' and 5' RACE walking and genomic library screening. Tammar *ATRX* is highly homologous to human and mouse *ATRX*, whereas tammar *ATRY* is homologous in most regions to tammar *ATRX*. The full sequence of *ATRX* is about 10 kb in length, whereas *ATRY* appears to be only about 7.5 kb. Using RT-PCR, marsupial *ATRX* is detected in a wide range of tissues, but is absent from the developing testis, whereas *ATRY* is expressed exclusively in the developing and adult testis (1). Antibody to *ATRY* is not yet available, but immunolocalisation of *ATRX* with a mouse antibody that will recognise both the *ATRX* and Y showed *ATRX* protein present in the testis and ovary from day 23 of pregnancy to 3-4 days before birth (well before gonadal sex differentiation) to the adult. During testis differentiation, *ATRX* immunostaining was localised in the Sertoli cells, the Leydig cells and in the germ cells suggesting that *ATRY* may play a role in the differentiation of the testis.

(1) Pask, A., Renfree, M.B. and Graves, J.A.M. (2000). *Proc. Natl Acad. Sci.* **97**, 13198–13202.