

Identification of Transcription Initiation Sites of Retinal Expressed Genes

*V.Roni*¹, *B.Wissinger*¹, *W.Raffelsberger*², *A.Mukhopadhyay*³. ¹Molecular genetics laboratory, University Eye Hospital Tuebingen, Tuebingen, Germany; ²Genetics and Molecular and Cellular Biology, IGBMC, Strasburg, France; ³Department of Human Genetics, University Medical Centre Nijmegen, Nijmegen, The Netherlands.

Keywords:

664 retina, 523 gene/expression, 712 transcription

Purpose: The aim of this study is the identification of transcription initiation sites of two retinal expressed genes RDH12 and SLC24A2. RDH12 gene codes for a photoreceptor specific retinal-dehydrogenase and is implicated in Leber congenital amaurosis. SLC24A2 codes for a potassium-dependent sodium-calcium exchanger in cone photoreceptor.

Methods:

We used available public database information, namely EST sequence and cross-species comparisons to perform in silico assembly and analysis of 5' transcript termini. In addition RNA from human retina was used for Cap Finder RACE experiments to study and characterize the ultimate 5' terminus of the genes of interest.

Results:

The assembly of ESTs present in the public database let us to define additional transcribed sequences extending to the 5' of the ref seq entries of the two genes. With the RACE experiment we were able to confirm these sequences. Moreover we also found additional exons that were not present in the databases. The new model for RDH12 contains three new untranslated exons 5' to the previously known sequence, one of which occurs in two different splice variants. The new model for SLC24A2 predicts two sites of transcription initiation as well as two additional exons that are alternatively spliced.

Conclusions:

In silico and experimental analysis of the transcripts proved to be essential for the ultimate mapping of the initiation site. This information constitutes the basis for further exploration of the promoter and others cis-regulatory sequences at the 5' end of the gene.

Commercial Relationship:

V. Roni, None; **B. Wissinger**, None; **W. Raffelsberger**, None; **A. Mukhopadhyay**, None.

Support:

European Union Research Training Network 'RETNET' MRTN-CT-2003-504003