

Chapter 7

References

- Altschul,S.F., Madden,T.L., Schaffer,A.A., Zhang,J, Zhang,Z., Miller,W., and Lipman,D.J. (1997) Gapped BLAST and PSI-BLAST: a new generation of protein database search programs, *Nucleic Acids Res.*, **25**, 3389-3402
- Baker,W., van den Broek,A., Camon,E., Hingamp,P., Sterk,P., Stoesser,G., and Tuli,M.A. (2000) The EMBL Nucleotide Sequence Database. *Nucleic Acids Res.*, **28**, 19-23.
- Berget,S.M. (1995) Exon recognition in vertebrate splicing. *J. Biol. Chem.*, **270**, 2411-2414.
- Berget,S.M., Moore,C. and Sharp,P.A. (1977) Spliced segments at the 5' terminus of adenovirus 2 late mRNA. *Proc. Natl. Acad. Sci. USA*, **74**, 3171-3175.
- Blencowe, B.J. (2000) Exonic splicing enhancers: mechanism of action, diversity and role in human genetic diseases. *Trends Biochem. Sci.*, **25**, 106-110.
- Boguski,M.S., Lowe,T.M., Tolstoshev,C.M. (1993) dbEST--database for "expressed sequence tags". *Nat. Genet.*, **4**, 332-333.
- Brunak,S. and Engelbrecht,J. (1991) Prediction of human mRNA donor and acceptor sites from the DNA sequence. *J. Mol. Biol.*, **220**, 49-65.
- Burge,C. (1997) Identification of complete gene structures in human genomic DNA. PhD thesis. Stanford University, Stanford, CA.
- Burge, C. (1998) Modeling dependencies in pre-mRNA splicing signals. In Salzberg,S.L., Searls,D.B., and Kasif,S. (eds.), *Computational Methods in Molecular Biology*. Elsevier Science. Amsterdam, Netherlands.
- Burge,C. and Karlin,S. (1997) Prediction of complete gene structures in human genomic DNA. *J. Mol. Biol.*, **268**, 78-94.
- Burge,C.B., Padgett,R.A, and Sharp,P.A. (1998) Evolutionary fates and origins of U12-type introns. *Mol. Cell*, **2**, 773-785.
- Burge,C.B., Tuschl,T., and Sharp,P.A. (1999) Splicing of precursors to mRNAs by the spliceosomes. In Gesteland,R.F. and Atkins,J.F. (eds.), *The RNA World*. (2nd edition) Cold Spring Harbor Laboratory Press, Cold Spring Harbor, NY.
- Burset,M., Seledtsov,I.A., and Solovyev,V.V. (2000) Analysis of canonical and non-canonical splice sites in mammalian genomes. *Nucleic Acids Res.*, **28**, 4364-4375.
- Burset,M., Seledtsov,I.A., and Solovyev,V.V. (2001) SpliceDB: database of canonical and non-canonical mammalian splice sites. *Nucleic Acids Res.*, **29**, 255-259.
- Chow,L.T., Gelinis,R.E., Broker,T.R., and Roberts,R.J. (1977) An amazing sequence arrangement at the 5' ends of adenovirus 2 messenger RNA. *Cell*, **12**, 1-8.
- Collins,C.A., and Guthrie,C. (2000) The question remains: is the spliceosome a ribozyme. *Nat. Struct. Biol.*, **7**, 850-854.

- Crispino, J., Blencowe, B.J. and Sharp, P.A. (1994) Complementation by SR proteins of pre-mRNA splicing reactions depleted of U1 snRNP. *Science*, **265**, 1860-1869.
- Crispino, J.D., Mermoud, J., Lamond, A., and Sharp, P.A. (1996) *Cis*-acting elements from the 5' splice site promote U1-independent pre-mRNA splicing. *RNA*, **2**, 664-673.
- Dietrich, R.C., Incorvaia, R. and Padgett, R.A. (1997) Terminal intron dinucleotide sequences do not distinguish between U2- and U12-dependent introns. *Mol. Cell*, **1**, 151-160.
- Dietrich, R.C., Peris, M.J., Seyboldt, A.S., and Padgett, R.A. (2001) Role of the 3' splice site in U12-dependent intron splicing. *Mol. Cell. Biol.*, **21**, 1942-1952.
- Dunham, I. *et al.*, (1999) The DNA sequence of human chromosome 22. *Nature*, **402**, 489-495.
- Durbin, R., Eddy, S., Krogh, A. and Mitchison, G. (1998) *Biological Sequence Analysis*. Cambridge University Press. Cambridge, UK.
- Graveley, B.R. (2000) Sorting out the complexity of SR protein functions. *RNA*, **6**, 1197-1211.
- Graveley, B.R. (2001) Alternative splicing: increasing diversity in the proteomic world. *Trends Genet.*, **17**, 100-107.
- Hall, S.L. and Padgett, R.A. (1994) Conserved sequences in a class of rare eukaryotic nuclear introns with non-consensus splice sites. *J. Mol. Biol.* **239**, 357-365.
- Harr, R., Haggstrom, M. and Gustafsson, P. (1983) Search algorithm for pattern match analysis of nucleic acid sequences. *Nucleic Acids Res.*, **11**, 2943-2957.
- Hastings, M.L. and Krainer, A.R. (2001) Functions of SR proteins in the U12-dependent AT-AC pre-mRNA splicing pathway. *RNA*, **7**, 471-482.
- Hurst, L.D., Brunton, C.F.A, and Smith, N.G.C. (1999) Small introns tend to occur in GC-rich regions in some but not all vertebrates. *Trends Genet.*, **15**, 437-439.
- International Human Genome Sequencing Consortium. (2001) Initial sequencing and analysis of the human genome. *Nature*, **409**, 860-921.
- Jackson, I.J. (1991) A reappraisal of non-consensus mRNA splice sites. *Nucleic Acids Res.*, **19**, 3795-3798.
- Konarksa, M.M. (1998) Recognition of the 5' splice site by the spliceosome. *Acta Biochim Pol*, **45**, 869-81.
- Kramer, A., Gruter, P., Groning, K. and Kastner, B. (1999) Combined biochemical and electron microscopic analyses reveal the architecture of the mammalian U2 snRNP. *J. Cell. Biol.*, **145**, 1355-1368.

- Kudo,M., Lida,Y. and Shimbo,M. (1987) Syntactic pattern analysis of 5' splice site sequences of mRNA precursors in higher eucaryote genes. *Comput. Appl. Biosci.*, **3**, 319-324.
- Lewin,B. (2000) *Genes VII*. Oxford University Press. Oxford, UK.
- Long,M., Rosenberg,C., and Gilbert,W. (1995) Intron phase correlations and the evolution of the intron/exon structure of genes. *Proc. Natl. Acad. Sci. USA*, **92**, 12495-12499.
- Luo,H.R., Moreau,G.A, Levin,N. and Moore,M.J. (1999) The human Prp8 protein is a component of both U2- and U12-dependent spliceosomes. *RNA*, **5**, 893-908.
- Meister,G., Hannus,S., Plottner,O., Baars,T., Hartmann,E., Fakan,S., Laggerbauer,B. and Fischer,U. (2001) SMNrp is an essential pre-mRNA splicing factor required for the formation of the mature spliceosome. *Embo J.*, **20**, 2304-2314.
- Modrek,B., Resch,A., Grassa,C. and Lee,C. (2001) Genome-wide detection of alternative splicing in expressed sequences of human genes. *Nucleic Acids Res.*, **29**, 2850-2859.
- Moore,M.J. (2000) Intron recognition comes of AGE. *Nat. Struct. Biol.*, **7**, 14-16.
- Mount,S.M. (1982) A catalogue of splice junction sequences. *Nucleic Acids Res.*, **10**, 459-472.
- Muratoglu,S., Krysan,K., Baláza,M., Sheng,H., Zákány,R., Módis,L., Kiss,I., and Deák,F. (2000) Primary structure of human matrilin-2, chromosome location of the MATN2 gene and conservation of an AT-AC intron in matrilin genes. *Cytogenet. Cell. Genet.*, **90**, 323-327.
- Pearson, W.R. (1995) Comparison of methods for searching protein sequence databases. *Protein Sci.*, **4**, 1145-1160.
- Rappsilber,J., Ajuh,P.M., Lamond,A.I. and Mann,M. (2001) SPF30, an essential human splice factor required for assembly of U4/U5/U6 tri-snRNP into the spliceosome. *J. Biol. Chem.*, in press.
- Reed,R. (2000) Mechanisms of fidelity in pre-mRNA splicing. *Curr. Opin. Cell. Biol.*, **12**, 340-345.
- Schwer,B. (2001) A new twist on RNA helicases: DExH/D box proteins as RNPsases. *Nat. Struct. Biol.*, **8**, 113-116.
- Sharp,P.A. and Burge,C.B. (1997) Classification of introns: U2-type or U12-type. *Cell*, **91**, 875-879.
- Shukla,G.C. and Padgett,R.A. (2001) The intramolecular stem-loop structure of U6 snRNA can functionally replace the U6atac snRNA stem-loop. *RNA*, **7**, 94-105.

- Staden,R. (1984). Computer methods to locate signals in nucleic acid sequences. *Nucleic Acids Res.*, **12**, 505-519.
- Staley,J.P. and Guthrie,C. (1999) An RNA switch at the 5' splice site requires ATP and the DEAD box protein Prp28p. *Mol. Cell.*, **3**, 55-64.
- Talerico,M. and Berget,S.M. (1994) Intron definition in splicing of small *Drosophila* introns. *Mol. Cell. Biol.*, **14**, 3434-3445.
- Tarn,W.Y. and Steitz,J.A. (1994) SR proteins can compensate for loss of U1 snRNP functions in vitro. *Genes Dev.*, **9**, 2704-2717.
- Tarn,W.Y. and Steitz,J.A. (1996a) A novel spliceosome containing the U11, U12 and U5 snRNPs excises a minor class (AT-AC) intron *in vitro*. *Cell*, **804**, 801-811.
- Tarn,W.Y. and Steitz,J.A. (1996b) Highly diverged U4 and U6 small nuclear RNAs required for splicing rare AT-AC introns. *Science*, **27**, 1824-1832.
- Venter,J.C. *et al.* (2001) The sequence of the human genome. *Science*, **291**, 1304-1351.
- Will, C.L., Schneider,C., Reed,R. and Lührmann,R. (1999) Identification of both shared and distinct proteins in the major and minor spliceosomes. *Science*, **284**, 2003-2005.
- Wu,Q. and Krainer,A.R. (1996) U1-mediated exon definition interactions between AT-AC and GT-AG introns. *Science*, **274**, 1005-1008.
- Wu,Q. and Krainer,A.R. (1999) AT-AC pre-mRNA splicing mechanisms and conservation of minor introns in voltage-gated ion channel genes. *Mol. Cell. Biol.*, **19**, 3225-3236.
- Wu,S., Romfo,C.M., Nilsen,T.W. and Green,M.R. (1999) Functional recognition of the 3' splice site AG by the splicing factor U2AF³⁵. *Nature*, **402**, 832-835.
- Zhang,M.Q. (1998) Statistical features of human exons and their flanking regions. *Human Mol. Genet.*, **7**, 919-932.
- Zhang,M.Q. and Marr,T.G. (1993) A weight array method for splicing signal analysis. *Comp. Appl. Biol. Sci.*, **9**, 499-509.
- Zorio,D.A.R. and Blumenthal,T. (1999) Both subunits of U2AF recognize the 3' splice site in *Caenorhabditis elegans*. *Nature*, **402**, 835-838.
- Zoubak,S., Clay,O., and Bernardi,G. (1996) The gene distribution of the human genome. *Gene*, **174**, 95-102.

Appendix A

Complete List of U12-Dependent Introns Identified in Chapter 4