

# Bibliography

- S. F. Altschul, W. Gish, W. Miller, E. W. Myers, and D. J. Lipman. Basic local alignment search tool. *J Mol Biol*, 215:403–410, 1990.
- M. Ariel, J. McCarrey, and H. Cedar. Methylation patterns of testis-specific genes. *Proc Natl Acad Sci*, 88:2317–2321, 1991.
- J. F. Bateman, S. Freddi, G. Natrass, and R. Savarirayan. Tissue-specific rna surveillance? nonsense-mediated mrna decay causes collagen x haploinsufficiency in schmid metaphyseal chondrodysplasia cartilage. *Hum Mol Genet*, 12(3):217–225, 2003.
- D. A. Benson, I. Karsch-Mizrachi, D. J. Lipman, J. Ostell, and D. L. Wheeler. Genbank: update. *Nucleic Acids Res*, 32:D23–D26, 2004.
- Y. Bergman and R. Mostoslavsky. Dna methylation: regulation of gene expression and role in immune system. *Biochim Biophys Acta*, 1333(1):F29–F50, 1997.
- P. Bertone, V. Stolc, T. E. Royce, J. S. Rozowsky, A. E. Urban, X. Zhu, J. L. Rinn, W. Tongprasit, M. Samanta, S. Weissman, M. Gerstein, and M. Snyder. Global identification of human transcribed sequences with genome tiling arrays. *Science*, 306(5705):2242–2246, 2004.
- T. J. Bloom and J. A. Beavo. Identification and tissue-specific expression of pde7 phosphodiesterase splice variants. *Proc Natl Acad Sci U S A*, 93(24):14188–14192, 1996.
- M. S. Boguski, T. M. Lowe, and C. M. Tolstoshev. dbest-database for "expressed sequence tags". *Nat Genet*, 4(4):332–333, 1993.
- M. F. Bonaldo, G. Lennon, and M. B. Soares. Normalization and subtraction: two approaches to facilitate gene discovery. *Genome Res*, 6(9):791–806, 1996.
- C. F. Bourgeois, F. Lejeune, and J. Stevenin. Broad specificity of sr (serine/arginine) proteins in the regulation of alternative splicing of pre-messenger rna. *Prog Nucleic Acid Res Mol Biol*, 78:37–88, 2004.
- K. S. Brocke, G. Neu-Yilik, N. H. Gehring, M. W. Hentze, and A. E. Kulozik. A novel algorithm for computational identification of contaminated est libraries. *Hum Mol Genet*, 80:601–613, 2002.

- C. Y. Brown, G. Z. Mize, M. Pineda, D. L. George, and D. R. Morris. Role of two upstream open reading frames in the translational control of oncogene mdm2. *Oncogene*, 18(41):5631–5637, 1999.
- C. B. Burge, T. Tuschl, and P. A. Sharp. *Splicing of precursors to mRNAs by the spliceosomes*. CSHL Press, 1999.
- J. F. Caceras and A. R. Kornblihtt. Alternative splicing: multiple control mechanisms and involvement in human disease. *Trends Genet*, 18(4):186–193, 2002.
- P. Carninci and Y. Hayashizaki. High-efficiency full-length cDNA cloning. *Methods Enzymol*, 303:19–44, 1999.
- L. Cartegni, S. L. Chew, and A. R. Krainer. Listening to silence and understanding nonsense: exonic mutations that affect splicing. *Nat Rev Genet*, 3(4):285–298, 2002.
- H. Chen, M. Centola, S. F. Altschul, and H. Metzger. Characterization of gene expression in resting and activated mast cells. *J Exp Med*, 188:1657–1668, 1998.
- F. Clark and T. A. Thanaraj. Categorization and characterization of transcript-confirmed constitutively and alternatively spliced introns and exons from human. *Hum Mol Genet*, 32:D64–D69, 2002.
- P. G. Corn and W. S. El-Deiry. Derangement of growth and differentiation control in oncogenesis. *Bioassays*, 24(1):83–90, 2002.
- E. Coward, S. A. Haas, and M. Vingron. SpliceNest: visualization of gene structure and alternative splicing based on EST clusters. *Trends Genet*, 18(1):53–55, 2002.
- C. Delaloy, J. Lu, A. Houot, S. Disse-Nicodeme, J. Gasc, P. Corvol, and X. Jeune-maitre. Multiple promoters in the *wnk1* gene: one controls expression of a kidney-specific kinase-defective isoform. *Mol Cell Biol*, 24:9208–9221, 2003.
- M. Diehn, G. Sherlock, G. Binkley, H. Jin, T. Matese, J. C. and Hernandez-Boussard, C. A. Rees, J. M. Cherry, D. Botstein, P. O. Brown, and A. A. Alizadeh. Source: a unified genomic resource of functional annotations, ontologies, and gene expression data. *Nucleic Acids Res*, 31(1):219–223, 2003.
- C. Dieterich, B. Cusack, H. Wang, K. Rateitschak, A. Krause, and M. Vingron. Annotating regulatory DNA based on man-mouse genomic comparison. *Bioinformatics*, 18(suppl 2):84–90, 2002.
- R. M. Ewing and J. M. Claverie. EST databases as multi-conditional gene expression datasets. *Pac Symp Biocomput*, pages 430–442, 2000.
- L. Florea, G. Hartzell, Z. Zhang, G. M. Rubin, and W. Miller. A computer program for aligning a cDNA sequence with a genomic DNA sequence. *Genome Res*, 8(9):967–974, 1998.

- T. H. Geerlings, A. W. Faber, M. D. Bister, J. C. Vos, and H. A. Raue. Rio2p, an evolutionarily conserved, low abundant protein kinase essential for processing of 20S pre-rRNA in *Saccharomyces cerevisiae*. *J Biol Chem*, 278(25):22537–22545, 2003.
- S. F. Gilbert. *DevBio: A companion to developmental biology*. Sinauer Associates, 2003.
- H. Granzier, D. Labeit, Y. Wu, C. Witt, K. Watanabe, S. Lahmers, M. Gotthardt, and S. Labeit. Adaptations in titin's spring elements in normal and cardiomyopathic hearts. *Adv Exp Med Biol*, 538:517–530, 2003.
- S. Gupta, M. Vingron, and S. A. Haas. T-STAG: Resource and web-interface for tissue-specific transcripts and genes. *Nucleic Acids Res*, 2005. in press.
- S. Gupta, D. Zink, B. Korn, M. Vingron, and S. A. Haas. Genome-wide identification and classification of alternative splicing based on EST data. *Bioinformatics*, 20(16):2579–2585, 2004a.
- S. Gupta, D. Zink, B. Korn, M. Vingron, and S. A. Haas. Strengths and weaknesses of EST-based prediction of tissue-specific alternative splicing. *BMC Genomics*, 5(72), 2004b.
- S. A. Haas, T. Beissbarth, E. Rivals, A. Krause, and M. Vingron. GeneNest: automated generation and visualization of gene indices. *Trends Genet*, 16(11):299–300, 2000.
- S. A. Haas, M. Hild, A. P. H. Wright, T. Hain, D. Talibi, and M. Vingron. Genome-scale design of PCR primers and long oligomers for DNA microarrays. *Nucleic Acids Res*, 31(19):5576–5581, 2003.
- A. Hanamura, J. F. Caceres, A. Mayeda, B. R. J. Franza, and A. R. Krainer. Regulated tissue-specific expression of antagonistic pre-mRNA splicing factors. *RNA*, 4(4):430–444, 1998.
- H. Hao, M. G. Tyshenko, and V. K. Walker. Dihydrofolate reductase of *Drosophila*. Cloning and expression of a gene with a rare transcript. *J Biol Chem*, 269(21):15179–15185, 1994.
- F. Hashimoto, N. Oyaizu, V. S. Karyanarayanan, and S. Pahwa. Modulation of bcl-2 protein by CD4 cross-linking: a possible mechanism for lymphocyte apoptosis in human immunodeficiency virus infection and for rescue of apoptosis by interleukin-2. *Blood*, 90:745–753, 1997.
- S. Hashimoto, T. Suzuki, H. Y. Dong, S. Nagai, N. Yamazaki, and K. Matsushima. Serial analysis of gene expression in human monocyte-derived dendritic cells. *Blood*, 94:845–852, 1999a.

- S. Hashimoto, T. Suzuki, H. Y. Dong, N. Yamazaki, and K. Matsushima. Serial analysis of gene expression in human monocytes and macrophages. *Blood*, 94:837–844, 1999b.
- K. Hibi, Q. Liu, G. A. Beaudry, S. L. Madden, W. H. Westra, S. L. Wehage, S. C. Yang, R. F. Heitmiller, A. H. Bertelsen, D. Sidransky, and J. Jen. Serial analysis of gene expression in non-small cell lung cancer. *Cancer Res*, 58:5690–5694, 1998.
- R. T. Hillman, R. E. Green, and S. E. Brenner. An unappreciated role for rna surveillance. *Genome Biol*, 5(2), 2004.
- H. Holthues and L. Vollrath. The phototransduction cascade in the isolated chick pineal gland revisited. *Brain Res*, 999(2):175–180, 2004.
- K. Irizarry, V. Kustanovich, C. Li, N. Brown, S. Nelson, W. Wong, and C. Lee. Genome-wide analysis of single-nucleotide polymorphisms in human expressed sequences. *Nat Genet*, 26:233–236, 2000.
- O. A. Itani, J. R. Campbell, J. Herrero, P. M. Snyder, , and C. P. Thomas. Alternate promoters and variable splicing lead to hnedd4-2 isoforms with a c2 domain and varying number of ww domains. *Am J Physiol Renal Physiol*, 285(5):916–929, 2003.
- X. Jin, E. Turcott, S. Englehardt, G. J. Mize, and M. D. R. The two upstream open reading frames of oncogene *mdm2* have different translational regulatory properties. *J Biol Chem*, 278(28):25716–25721, 2003.
- J. M. Johnson, J. Castle, P. Garrett-Engele, Z. Kan, P. M. Loerch, C. D. Armour, R. Santos, E. E. Schadt, R. Stoughton, and D. D. Shoemaker. Genome-wide survey of human alternative pre-mrna splicing with exon junction microarrays. *Science*, 302(5653):2141–2144, 2003.
- Z. Kan, D. States, and W. Gish. Selecting for functional alternative splices in ests. *Genome Res*, 12:1837–1845, 2002.
- A. Y. Karpova, P. M. Howley, and L. V. Ronco. Dual utilization of an acceptor/donor splice site governs the alternative splicing of the *irf-3* gene. *Genes Dev*, 14(22):2813–2818, 2000.
- S. Kuersten and E. B. Goodwin. The power of the 3'-utr: translational control and development. *Nat Rev Genet*, 4:626–637, 2003.
- S. Kurtz, J. V. Choudhuri, E. Ohlebusch, C. Schleiermacher, J. Stoye, and R. Giegerich. Reputer: the manifold applications of repeat analysis on a genomic scale. *Nucleic Acids Res*, 29:4633–4642, 2001.
- C. Lee, L. Atanelov, B. Modrek, and Y. Xing. *asap*: The alternative splicing annotation project. *Nucleic Acids Res*, 31:101–105, 2003.

- B. P. Lewis, R. E. Green, and S. E. Brenner. Evidence for the widespread coupling of alternative splicing and nonsense-mediated mrna decay in humans. *Proc Natl Acad Sci USA*, 100(1):189–192, 2003.
- Q. R. Lu, J. K. Park, E. Noll, J. A. Chan, J. Alberta, D. Yuk, M. G. Alzamora, D. N. Louis, C. D. Stiles, D. H. Rowitch, and P. M. Black. Oligodendrocyte lineage genes (olig) as molecular markers for human glial brain tumors. *Proc Natl Acad Sci U S A*, 98(19):10851–10856, 2001.
- M. Malumbres and A. Carnero. Cell cycle deregulation: a common motif in cancer. *Prog Cell Cycle Res*, 5:5–18, 2003.
- M. Matsushita, R. Yamazaki, H. Ikeda, and Y. Kawakami. Preferentially expressed antigen of melanoma (prame) in the development of diagnostic and therapeutic methods for hematological malignancies. *Leuk Lymphoma*, 44(3):439–444, 2003.
- K. Megy, S. Audic, and J. Claverie. Heart-specific genes revealed by expressed sequence tag (est) sampling. *Genome Biol*, 3(12):74.1–74.11, 2002.
- A. A. Mironov, J. W. Fickett, and M. S. Gelfand. Frequent alternative splicing of human genes. *Genome Res*, 9(12):1288–1293, 1999.
- W. S. Modi, D. D. Pollock, B. A. Mock, C. Banner, J. C. Renauld, and J. Van Snick. Regional localization of the human glutaminase (gls) and interleukin-9 (il9) genes by in situ hybridization. *Cytogenet Cell Genet*, 57(2-3):114–116, 1991.
- B. Modrek and C. Lee. A genomic view of alternative splicing. *Nat Genet*, 30:13–19, 2002.
- B. Modrek, A. Resch, C. Grasso, and C. Lee. Genome-wide detection of alternative splicing in expressed sequences of human genes. *Nucleic Acids Res*, 29(13):2850–2859, 2001.
- T. Naiki, M. Nagaki, Y. Shidoji, H. Kojima, M. Imose, T. Kato, N. Ohishi, K. Yagi, and H. Moriwaki. Analysis of gene expression profile induced by hepatocyte nuclear factor 4alpha in hepatoma cells using an oligonucleotide microarray. *J Biol Chem*, 277(16):14011–14019, 2002.
- A. Nishida, A. Furukawa, C. Koike, Y. Tano, S. Aizawa, I. Matsuo, and T. Furukawa. Otx2 homeobox gene controls retinal photoreceptor cell fate and pineal gland development. *Nat Neurosci*, 6(12):1255–1263, 2003.
- D. T. Odom, N. Zizlsperger, D. B. Gordon, G. W. Bell, N. J. Rinaldi, H. L. Murray, T. L. Volkert, J. Schreiber, P. A. Rolfe, D. K. Gifford, E. Fraenkel, G. I. Bell, and R. A. Young. Control of pancreas and liver gene expression by hnf transcription factors. *Science*, 303(5662):1378–1381, 2004.

- C. J. Phiel, V. Gabbeta, L. M. Parsons, D. Rothblat, R. P. Harvey, and K. M. McHugh. Differential binding of an *srf/nk-2/mef2* transcription factor complex in normal versus neoplastic smooth muscle tissues. *J Biol Chem*, 276(37):34637–34650, 2001.
- S. Pikkarainen, H. Tokola, R. Kerkela, and H. Ruskoaho. Gata transcription factors in the developing and adult heart. *Cardiovasc Res*, 63(2):196–207, 2004.
- A. Poliard, G. Feldmann, and D. Bernuau. Alpha fetoprotein and albumin gene transcripts are detected in distinct cell populations of the brain and kidney of the developing rat. *Differentiation*, 39(1):59–65, 1998.
- K. Polyak, Y. Xia, J. L. Zweier, K. W. Kinzler, and B. Vogelstein. A model for p53 induced apoptosis. *Nature*, 389:300–305, 1997.
- T. Putilina, C. Jaworski, S. Gentleman, B. McDonald, M. Kadiri, and P. Wong. Analysis of a human cDNA containing a tissue-specific alternatively spliced LIM domain. *Biochem Biophys Res Commun*, 252(2):433–439, 1998.
- J. C. Reubi, B. Waser, W. Vale, and J. Rivier. Expression of *CRF1* and *CRF2* receptors in human cancers. *L Clin Endocrinol Metab*, 88(7):3312–3320, 2003.
- G. C. Roberts and C. W. J. Smith. Alternative splicing: combinatorial output from the genome. *Curr Opin Chem Biol*, 6:375–383, 2002.
- S. M. Russell, R. L. Sparrow, I. F. McKenzie, and D. F. Purcell. Tissue-specific and allelic expression of the complement regulator *cd46* is controlled by alternative splicing. *Eur J Immunol*, 22(6):1513–1518, 1992.
- A. Ryo, Y. Suzuki, K. Ichiyama, T. Wakatsuki, N. Kondoh, A. Hada, M. Yamamoto, and N. Yamamoto. Serial analysis of gene expression in HIV-1-infected T cell lines. *FEBS Lett*, 462:182–186, 1999.
- C. G. Sagerstrom, B. I. Sun, and H. L. Sive. Subtractive cloning: past, present, and future. *Annu Rev Biochem*, 66:751–783, 1997.
- J. Sambrook. Adenovirus amazes at cold spring harbour. *Nature*, 268:101–104, 1977.
- J. R. Sanford, D. Longman, and J. F. Caceres. Multiple roles of the SR protein family in splicing regulation. *Prog Mol Subcell Biol*, 31:33–58, 2003.
- E. E. Schadt, S. W. Edwards, D. GuhaThakurta, D. Holder, L. Ying, V. Svetnik, A. Leonardson, K. W. Hart, A. Russell, G. Li, G. Cavet, J. Castle, P. McDonagh, Z. Kan, R. Chen, A. Kasarskis, M. Margarint, R. M. Caceres, J. M. Johnson, C. D. Armour, P. W. Garrett-Engle, N. F. Tsinoremas, and D. D. Shoemaker. A comprehensive transcript index of the human genome generated using microarrays and computational approaches. *Genome Biol*, 5(10):R73, 2004.

- A. O. Schmitt, T. Specht, G. Beckmann, E. Dahl, C. P. Pilarsky, B. Hinzmann, and A. Rosenthal. Exhaustive mining of est libraries for genes differentially expressed in normal and tumour tissues. *Nucleic Acids Res*, 27(21):4251–4260, 1999.
- D. Schmucker, J. C. Clemens, H. Shu, C. A. Worby, J. Xiao, M. Muda, J. E. Dixon, and S. L. Zipursky. *Drosophila* dscam is an axon guidance receptor exhibiting extraordinary molecular diversity. *Cell*, 101(3):671–684, 2000.
- Y. Shibata, P. Carninci, A. Watahiki, T. Shiraki, H. Konno, M. Muramatsu, and Y. Hayashizaki. Cloning full-length, cap-trapper-selected cdnas by using the single-strand linker ligation method. *Biotechniques*, 30(6):1250–1254, 2001.
- T. Shiraki, S. Kondo, S. Katayama, K. Waki, T. Kasukawa, H. Kawaji, R. Kodzius, A. Watahiki, M. Nakamura, T. Arakawa, S. Fukuda, D. Sasaki, A. Podhajski, M. Harbers, J. Kawai, P. Carninci, and Y. Hayashizaki. Cap analysis gene expression for high-throughput analysis of transcriptional starting point and identification of promoter usage. *Proc Natl Acad Sci U S A*, 100(26):15776–15781, 2003.
- C. C. Siebrands, J. M. Sanger, and J. W. Sanger. Myofibrillogenesis in skeletal muscle cells in the presence of taxol. *Cell Motil Cytoskeleton*, 58(1):39–52, 2004.
- R. Sorek and H. M. Safer. A novel algorithm for computational identification of contaminated est libraries. *Nucleic Acids Res*, 31(3):1067–1074, 2003.
- S. Stamm, J. Zhu, K. Nakai, P. Stoilov, O. Stoss, and M. Q. Zhang. An alternative-exon database and its statistical analysis. *DNA Cell Biol*, 19:739–756, 2000.
- S. W. Stevens, D. E. Ryan, H. Y. Ge, R. E. Moore, M. K. Young, T. D. Lee, and J. Abelson. Composition and functional characterization of the yeast spliceosomal penta-snrnp. *Mol Cell*, 9(1):31–44, 2002.
- G. Stoesser, W. Baker, A. Van den Broek, E. Camon, M. Garcia-Pastor, C. Kanz, T. Kulikova, V. Lombard, R. Lopez, H. Parkinson, N. Redaschi, P. Sterk, P. Stoehr, and M. A. Tuli. The embl nucleotide sequence database. *Nucleic Acids Res*, 29(1):17–21, 2001.
- D. H. Sweet, D. S. Miller, J. B. Pritchard, Y. Fujiwara, D. R. Beier, and S. K. Nigam. Impaired organic anion transport in kidney and choroid plexus of organic anion transporter 3 (oat3 (slc22a8)) knockout mice. *J Biol Chem*, 277(30):26934–26943, 2002.
- T. A. Thanaraj and F. Clark. Human gc-ag alternative intron isoforms with weak donor sites show enhanced consensus at acceptor exon positions. *Nucleic Acids Res*, 29(12):2581–2593, 2001.
- T. A. Thanaraj, S. Stamm, F. Clark, J. J. Riethoven, V. Le Texier, and J. Muilu. Asd: the alternative splicing database. *Nucleic Acids Res*, 32:D64–D69, 2004.

- O. Tureci, U. Sahin, M. Koslowski, B. Buss, C. Bell, P. Ballweber, C. Zwick, T. Eberle, M. Zuber, C. Villena-Heinsen, G. Seitz, and M. Pfreundschuh. A novel tumour associated leucine zipper protein targeting to sites of gene transcription and splicing. *Oncogene*, 21(24):3879–3888, 2002.
- J. Villard. Transcription regulation and human diseases. *Swiss Med Wkly*, 134(39-40): 571–579, 2004.
- Z. Wang, L. H. S, H. Yang, S. Gere, Y. Hu, K. H. Buetow, and M. P. Lee. Computational analysis and experimental validation of tumor-associated alternative rna splicing in human cancer. *Cancer Res*, 63(3):655–657, 2003.
- M. Weir and M. Rice. Ordered partitioning reveals extended splice-site consensus information. *Genome Res*, 14:67–78, 2004.
- D. L. Wheeler, D. M. Church, S. Federhen, A. E. Lash, T. L. Madden, J. U. Pontius, G. D. Schuler, L. M. Schriml, E. Sequeira, T. A. Tatusova, and L. Wagner. Database resources of the national center for biotechnology. *Nucleic Acids Res*, 31:28–33, 2003.
- R. Wieder, H. Wang, S. Shirke, Q. Wang, T. Menzel, N. Feirt, A. A. Jakubowski, and J. L. Gabilove. Low level expression of basic fgf upregulates bcl-2 and delays apoptosis, but high intracellular levels are required to induce transformation in nih 3t3 cells. *Growth Factors*, 15(1):41–60, 1997.
- G. S. Wilkie, K. S. Dickson, and N. K. Gray. Regulation of mrna translation by 5'- and 3'-utr-binding factors. *Trends Biochem Sci*, 28(4):182–188, 2003.
- C. L. Will and R. Luhrmann. Spliceosomal usnrnp biogenesis, structure and function. *Curr Opin Cell Biol*, 13(3):290–301, 2001.
- K. J. Worsley. An improved bonferroni inequality and applications. *Biometrika*, 69: 297–302, 1982.
- N. A. Woychik and M. Hampsey. The rna polymerase ii machinery: Structure illuminates function. *Cell*, 108(4):453–463, 2002.
- Q. Xu and C. Lee. Discovery of novel splice forms and functional analysis of cancer-specific alternative splicing in human expressed sequences. *Nucleic Acids Res*, 31: 5635–5643, 2003.
- Q. Xu, B. Modrek, and C. Lee. Genome-wide detection of tissue-specific alternative splicing in the human transcriptome. *Nucleic Acids Res*, 30(17):3754–3766, 2002.
- M. Yamamoto, T. Wakatsuki, A. Hada, and A. Ryo. Use of serial analysis of gene expression (sage) technology. *J Immunol Methods*, 250:45–46, 2001.
- C. Yan, A. Z. Zhao, J. K. Bently, and J. A. Beavo. The calmodulin-dependent phosphodiesterase gene pde1c encodes several functionally different splice variants in a tissue specific manner. *J Biol Chem*, 271(41):25699–25706, 1996.



- L. A. Zadeh. Fuzzy sets. *Information Control*, 8:338–353, 1965.
- L. Zhang, W. Zhou, V. E. Velculescu, S. E. Kern, R. H. Hruban, S. R. Hamilton, B. Vogelstein, and K. W. Kinzler. Gene expression profiles in normal and cancer cells. *Science*, 276:1268–1272, 1997.
- T. Zhang, P. Haws, and Q. Wu. Multiple variable first exons: a mechanism for cell- and tissue-specific gene regulation. *Genome Res*, 14(1):79–89, 2004.

