
Bibliography for chapter 1

1. Strachan T, Read AP. Human Molecular Genetics. 2nd edition. New York: Wiley-Liss; 1999. Chapter 7, <http://www.ncbi.nlm.nih.gov/books/NBK7587/#A664>.
2. Lander ES, Linton LM, Birren B, Nusbaum C, Zody MC et.al. Initial sequencing and analysis of the human genome. Nature. 2001 Feb 15;409(6822):860-921. Erratum in: Nature 2001 Aug 2;412(6846):565. Nature 2001 Jun 7;411(6838):720.
3. Strachan T, Read AP. Human Molecular Genetics. 2nd edition. New York: Wiley-Liss; 1999. Chapter 8, <http://www.ncbi.nlm.nih.gov/books/NBK7588/>.
4. Strachan T, Read AP. Human Molecular Genetics. 2nd edition. New York: Wiley-Liss; 1999. Chapter 5, <http://www.ncbi.nlm.nih.gov/books/NBK7567/>.
5. Brown TA. Genomes. 2nd edition. Oxford: Wiley-Liss; 2002. Chapter 7, <http://www.ncbi.nlm.nih.gov/books/NBK21136/>.
6. Ramsköld D, Kavak E, Sandberg R. How to analyze gene expression using RNA-sequencing data. Methods Mol Biol. 2012;802:259-74.
7. Resing KA. Proteomics for cell protein expression profiling. J Invest Dermatol. 2003 Jul;121(1):XI-XII.
8. Hegde PS, White IR, Debouck C. Interplay of transcriptomics and proteomics. Drug Discov Today. 2004 Jan 15;9(2 Suppl):S53-6.
9. Wang C, Gong B, Bushel PR, Thierry-Mieg J, Thierry-Mieg D, Xu J, Fang H, Hong H, Shen J, Su Z, Meehan J, Li X, Yang L, Li H, Łabaj PP, Kreil DP, Megherbi D, Gaj S, Caiment F, van Delft J, Kleinjans J, Scherer A, Devanarayan V, Wang J, Yang Y, Qian HR, Lancashire LJ, Bessarabova M, Nikolsky Y, Furlanello C, Chierici M, Albanese D, Jurman G, Riccadonna S, Filosi M, Visintainer R, Zhang KK, Li J, Hsieh JH, Svoboda DL, Fuscoe JC, Deng Y, Shi L, Paules RS, Auerbach SS, Tong W. The concordance between RNA-seq and microarray data depends on chemical treatment and transcript abundance. Nat Biotechnol. 2014 Sep;32(9):926-32.
10. Zhao S, Fung-Leung WP, Bittner A, Ngo K, Liu X. Comparison of RNA-Seq and microarray in transcriptome profiling of activated T cells. PLoS One. 2014 Jan 16;9(1):e78644.
11. Gry M, Rimini R, Strömberg S, Asplund A, Pontén F, Uhlén M, Nilsson P. Correlations between RNA and protein expression profiles in 23 human cell lines. BMC Genomics. 2009 Aug 7;10:365.
12. Ghazalpour A, Bennett B, Petyuk VA, Orozco L, Hagopian R, Mungrue IN, Farber CR, Sinsheimer J, Kang HM, Furlotte N, Park CC, Wen PZ, Brewer H, Weitz K, Camp DG 2nd, Pan C, Yordanova R, Neuhaus I, Tilford C, Siemers N, Gargalovic P, Eskin E, Kirchgessner T, Smith DJ, Smith RD, Lusis AJ. Comparative analysis of proteome and transcriptome variation in mouse. PLoS Genet. 2011 Jun;7(6):e1001393.
13. Bumgarner R. Overview of DNA microarrays: types, applications, and their future. Curr Protoc Mol Biol. 2013 Jan; Chapter 22:Unit 22.1.
14. PubMed: <http://www.ncbi.nlm.nih.gov/pubmed>
15. Schena M, Shalon D, Davis RW, Brown PO. Quantitative monitoring of gene expression patterns with a complementary DNA microarray. Science. 1995 Oct 20;270(5235):467-70.
16. Barrett T, Troup DB, Wilhite SE, Ledoux P, Evangelista C, Kim IF, Tomashevsky M, Marshall KA, Phillippy KH, Sherman PM, Muetter RN, Holko M, Ayanbule O, Yefanov A, Soboleva A. NCBI GEO: archive for functional genomics data sets--10 years on. Nucleic Acids Res. 2011, 39(Database issue):D1005-1010.

-
17. Ha KCh, Coulombe-Huntington J, Majewski J. Comparison of Affymetrix Gene Array with the Exon Array shows potential application for detection of transcript isoform variation. *BMC Genomics*. 2009 Nov 12;10:519.
 18. Barrett JC, Kawasaki ES. Microarrays: the use of oligonucleotides and cDNA for the analysis of gene expression. *Drug Discov Today*. 2003 Feb 1;8(3):134-41.
 19. Hardiman G. Microarray platforms--comparisons and contrasts. *Pharmacogenomics*. 2004 Jul;5(5):487-502.
 20. Illumina array based gene expression analysis: http://www.ohsu.edu/xd/research/research-cores/gene-profiling-shared-resource/about/upload/datasheet_gene_exp_analysis.pdf
 21. GeneChip® Human Exon ST Array: http://www.affymetrix.com/catalog/131452/AFFY/Human+Exon+ST+Array#1_1
 22. GeneChip® Gene 1.0 ST Array: http://gegf.net/storage/gene_1_0_st_technote.pdf
 23. Barnes M, Freudenberg J, Thompson S, Aronow B, Pavlidis P. Experimental comparison and cross-validation of the Affymetrix and Illumina gene expression analysis platforms. *Nucleic Acids Res*. 2005 Oct 19;33(18):5914-23.
 24. Applied microarrays: http://www.appliedmicroarrays.com/back_up/Human.html
 25. Verdugo RA, Medrano JF. Comparison of gene coverage of mouse oligonucleotide microarray platforms. *BMC Genomics*. 2006 Mar 21;7:58.
 26. Trevino V, Falciani F, Barrera-Saldaña HA. DNA microarrays: a powerful genomic tool for biomedical and clinical research. *Mol Med*. 2007 Sep-Oct;13(9-10):527-41.
 27. GeneChip scanner: http://www.affymetrix.com/estore/catalog/131503/AFFY/Scanner+3000+7G#1_1
 28. GenePix scanner: <http://www.moleculardevices.com/systems/microarray-scanners>
 29. SpotLight scanner: http://www.arrayit.com/Products/Microarray_Scanners/Microarray_Scanner_SpotLight_2/microarray_scanner_spotlight_2.html
 30. SureScan scanner: <http://www.genomics.agilent.com/en/Microarray-Scanner-Processing-Hardware/SureScan-Dx-Microarray-Scanner/?cid=AG-PT-144&tabId=AG-PR-1199>
 31. Bilban M, Buehler LK, Head S, Desoye G, Quaranta V. Normalizing DNA microarray data. *Curr Issues Mol Biol*. 2002 Apr;4(2):57-64.
 32. Quackenbush J. Microarray data normalization and transformation. *Nat Genet*. 2002 Dec;32 Suppl:496-501.
 33. GeneSpring: <http://www.genomics.agilent.com/en/Microarray-Data-Analysis-Software/GeneSpring-GX/?cid=AG-PT-130&tabId=AG-PR-1061>
 34. Genowiz: <http://www.ocimumbio.com/genowiz/>
 35. Yauk CL, Berndt ML. Review of the literature examining the correlation among DNA microarray technologies. *Environ Mol Mutagen*. 2007 Jun;48(5):380-94.
 36. MAQC Consortium, Shi L, Reid LH, Jones WD, Shippy R, Warrington JA, et.al. The MicroArray Quality Control (MAQC) project shows inter- and intraplatform reproducibility of gene expression measurements. *Nat Biotechnol*. 2006 Sep;24(9):1151-61.

-
37. Campain A, Yang YH. Comparison study of microarray meta-analysis methods. *BMC Bioinformatics*. 2010 Aug 3;11:408.
 38. Chang LC, Lin HM, Sibille E, Tseng GC. Meta-analysis methods for combining multiple expression profiles: comparisons, statistical characterization and an application guideline. *BMC Bioinformatics*. 2013 Dec 21;14:368.
 39. Kondrakhin YV, Sharipov RN, Keld AE, Kolpakov FA. Identification of differentially expressed genes by meta-analysis of microarray data on breast cancer. *In Silico Biol*. 2008;8(5-6):383-411.
 40. Goonesekere NC, Wang X, Ludwig L, Guda C. A meta analysis of pancreatic microarray datasets yields new targets as cancer genes and biomarkers. *PLoS One*. 2014 Apr 16;9(4):e93046.
 41. Lu TP, Hsu YY, Lai LC, Tsai MH, Chuang EY. Identification of gene expression biomarkers for predicting radiation exposure. *Sci Rep*. 2014 Sep 5;4:6293.
 42. Kotelnikova E, Shkrob MA, Pyatnitskiy MA, Ferlini A, Daraselia N. Novel approach to meta-analysis of microarray datasets reveals muscle remodeling-related drug targets and biomarkers in Duchenne muscular dystrophy. *PLoS Comput Biol*. 2012 Feb;8(2):e1002365.
 43. van Hal NL, Vorst O, van Houwelingen AM, Kok EJ, Peijnenburg A, Aharoni A, van Tunen AJ, Keijer J. The application of DNA microarrays in gene expression analysis. *J Biotechnol*. 2000 Mar 31;78(3):271-80.
 44. Majtán T, Bukovská G, Timko J. DNA microarrays--techniques and applications in microbial systems. *Folia Microbiol (Praha)*. 2004;49(6):635-64.
 45. Abdullah-Sayani A, Bueno-de-Mesquita JM, van de Vijver MJ. Technology Insight: tuning into the genetic orchestra using microarrays--limitations of DNA microarrays in clinical practice. *Nat Clin Pract Oncol*. 2006 Sep;3(9):501-16.
 46. Olson NE. The microarray data analysis process: from raw data to biological significance. *NeuroRx*. 2006 Jul;3(3):373-83.
 47. Affymetrix support: http://www.affymetrix.com/support/help/faqs/exon_array_design/faq_1.jsp
 48. Lipshutz RJ, Fodor SP, Gingeras TR, Lockhart DJ. High density synthetic oligonucleotide arrays. *Nat Genet*. 1999 Jan;21(1 Suppl):20-4.
 49. Genevestigator manual: https://genevestigator.com/gv/file/2014-08_GENEVESTIGATOR_UserManual.pdf
 50. Affymetrix support: http://media.affymetrix.com/support/downloads/manuals/data_analysis_fundamentals_manual.pdf
 51. Affymetrix exon array: http://www.affymetrix.com/estore/catalog/131452/AFFY/Human+Exon+ST+Array#1_1
 52. Affymetrix gene array: http://www.affymetrix.com/estore/catalog/131453/AFFY/Human+Gene+ST+Arrays#1_1
 53. Affymetrix transcriptome array: http://media.affymetrix.com/support/technical/datasheets/hta_array_2_0_datasheet.pdf
 54. Nagalakshmi U, Wang Z, Waern K, Shou C, Raha D, Gerstein M, Snyder M. The transcriptional landscape of the yeast genome defined by RNA sequencing. *Science*. 2008 Jun 6;320(5881):1344-9.

-
55. Wilhelm BT, Marguerat S, Watt S, Schubert F, Wood V, Goodhead I, Penkett CJ, Rogers J, Bähler J. Dynamic repertoire of a eukaryotic transcriptome surveyed at single-nucleotide resolution. *Nature*. 2008 Jun 26;453(7199):1239-43.
 56. Mortazavi A, Williams BA, McCue K, Schaeffer L, Wold B. Mapping and quantifying mammalian transcriptomes by RNA-Seq. *Nat Methods*. 2008 Jul;5(7):621-8.
 57. Wang Z, Gerstein M, Snyder M. RNA-Seq: a revolutionary tool for transcriptomics. *Nat Rev Genet*. 2009 Jan;10(1):57-63.
 58. Trapnell C, Roberts A, Goff L, Pertea G, Kim D, Kelley DR, Pimentel H, Salzberg SL, Rinn JL, Pachter L. Differential gene and transcript expression analysis of RNA-seq experiments with TopHat and Cufflinks. *Nat Protoc*. 2012 Mar 1;7(3):562-78.
 59. Metzker ML. Sequencing technologies - the next generation. *Nat Rev Genet*. 2010 Jan;11(1):31-46.
 60. Shendure J, Ji H. Next-generation DNA sequencing. *Nat Biotechnol*. 2008 Oct;26(10):1135-45.
 61. Trapnell C, Pachter L, Salzberg SL. TopHat: discovering splice junctions with RNA-Seq. *Bioinformatics*. 2009 May 1;25(9):1105-11.
 62. Dobin A, Davis CA, Schlesinger F, Drenkow J, Zaleski C, Jha S, Batut P, Chaisson M, Gingeras TR. STAR: ultrafast universal RNA-seq aligner. *Bioinformatics*. 2013 Jan 1;29(1):15-21.
 63. Campagna D, Albiero A, Bilardi A, Caniato E, Forcato C, Manavski S, Vitulo N, Valle G. PASS: a program to align short sequences. *Bioinformatics*. 2009 Apr 1;25(7):967-8.
 64. Li R, Yu C, Li Y, Lam TW, Yiu SM, Kristiansen K, Wang J. SOAP2: an improved ultrafast tool for short read alignment. *Bioinformatics*. 2009 Aug 1;25(15):1966-7.
 65. Langmead B, Trapnell C, Pop M, Salzberg SL. Ultrafast and memory-efficient alignment of short DNA sequences to the human genome. *Genome Biol*. 2009;10(3):R25.
 66. Lindner R, Friedel CC. A comprehensive evaluation of alignment algorithms in the context of RNA-seq. *PLoS One*. 2012;7(12):e52403.
 67. Engström PG, Steijger T, Sipos B, Grant GR, Kahles A, Räscher G, Goldman N, Hubbard TJ, Harrow J, Guigó R, Bertone P; RGASP Consortium. Systematic evaluation of spliced alignment programs for RNA-seq data. *Nat Methods*. 2013 Dec;10(12):1185-91.
 68. Griffith M, Griffith OL, Mwenifumbo J, Goya R, Morrissy AS, Morin RD, Corbett R, Tang MJ, Hou YC, Pugh TJ, Robertson G, Chittaranjan S, Ally A, Asano JK, Chan SY, Li HI, McDonald H, Teague K, Zhao Y, Zeng T, Delaney A, Hirst M, Morin GB, Jones SJ, Tai IT, Marra MA. Alternative expression analysis by RNA sequencing. *Nat Methods*. 2010 Oct;7(10):843-7.
 69. Trapnell C, Williams BA, Pertea G, Mortazavi A, Kwan G, van Baren MJ, Salzberg SL, Wold BJ, Pachter L. Transcript assembly and quantification by RNA-Seq reveals unannotated transcripts and isoform switching during cell differentiation. *Nat Biotechnol*. 2010 May;28(5):511-5.
 70. Bullard JH, Purdom E, Hansen KD, Dudoit S. Evaluation of statistical methods for normalization and differential expression in mRNA-Seq experiments. *BMC Bioinformatics*. 2010 Feb 18;11:94.
 71. Anders S, Huber W. Differential expression analysis for sequence count data. *Genome Biol*. 2010;11(10):R106.
 72. Robinson MD, Oshlack A. A scaling normalization method for differential expression analysis of RNA-seq data. *Genome Biol*. 2010;11(3):R25.

-
73. Dillies MA, Rau A, Aubert J, Hennequet-Antier C, Jeanmougin M, Servant N, Keime C, Marot G, Castel D, Estelle J, Guernec G, Jagla B, Jouneau L, Laloë D, Le Gall C, Schaeffer B, Le Crom S, Guedj M, Jaffrézic F; French StatOmique Consortium. A comprehensive evaluation of normalization methods for Illumina high-throughput RNA sequencing data analysis. *Brief Bioinform.* 2013 Nov;14(6):671-83.
 74. Trapnell C, Hendrickson DG, Sauvageau M, Goff L, Rinn JL, Pachter L. Differential analysis of gene regulation at transcript resolution with RNA-seq. *Nat Biotechnol.* 2013 Jan;31(1):46-53.
 75. Robinson MD, McCarthy DJ, Smyth GK. edgeR: a Bioconductor package for differential expression analysis of digital gene expression data. *Bioinformatics.* 2010 Jan 1;26(1):139-40.
 76. Seyednasrollah F, Laiho A, Elo LL. Comparison of software packages for detecting differential expression in RNA-seq studies. *Brief Bioinform.* 2015 Jan;16(1):59-70.
 77. Malone JH, Oliver B. Microarrays, deep sequencing and the true measure of the transcriptome. *BMC Biol.* 2011 May 31;9:34.
 78. Oshlack A, Robinson MD, Young MD. From RNA-seq reads to differential expression results. *Genome Biol.* 2010;11(12):220.
 79. Rajeevan MS, Ranamukhaarachchi DG, Vernon SD, Unger ER. Use of real-time quantitative PCR to validate the results of cDNA array and differential display PCR technologies. *Methods.* 2001 Dec;25(4):443-51.
 80. Rajkumar AP, Qvist P, Lazarus R, Lescai F, Ju J, Nyegaard M, Mors O, Børghlum AD, Li Q, Christensen JH. Experimental validation of methods for differential gene expression analysis and sample pooling in RNA-seq. *BMC Genomics.* 2015 Jul 25;16:548.
 81. Manecksha RP, Fitzpatrick JM. Epidemiology of testicular cancer. *BJU Int.* 2009 Nov;104(9 Pt B):1329-33.
 82. Thonneau P, Marchand S, Tallec A, Ferial ML, Ducot B, Lansac J, Lopes P, Tabaste JM, Spira A. Incidence and main causes of infertility in a resident population (1,850,000) of three French regions (1988-1989). *Hum Reprod.* 1991 Jul;6(6):811-6.
 83. Mansbach JM, Forbes P, Peters C. Testicular torsion and risk factors for orchiectomy. *Arch Pediatr Adolesc Med.* 2005 Dec;159(12):1167-71.
 84. Møller H, Prener A, Skakkebaek NE. Testicular cancer, cryptorchidism, inguinal hernia, testicular atrophy, and genital malformations: case-control studies in Denmark. *Cancer Causes Control.* 1996 Mar;7(2):264-74.
 85. Nieschlag, Eberhard, Behre, Hermann M., Nieschlag, Susan (Eds.). Male reproductive health and dysfunction. 3rd ed. 2010, XVII, 629 p. Chapter 2, <http://www.springer.com/978-3-540-78354-1>.
 86. Anatomy and Physiology of Male Reproduction: <https://www.cornellurology.com/clinical-conditions/male-infertility/general-information/anatomy-and-physiology-of-male-reproduction/>
 87. MedlinePlus: <http://www.nlm.nih.gov/medlineplus/ency/anatomyvideos/000121.htm>
 88. Bablok L, Dziadecki W, Szymusik I, Wolczynski S, Kurzawa R, Pawelczyk L, Jedrzejczak P, Hanke W, Kaminski P, Wielgos M. Patterns of infertility in Poland - multicenter study. *Neuro Endocrinol Lett.* 2011;32(6):799-804.
 89. Eberhard Nieschlag, Hermann M. Behre, Susan Nieschlag. *Andrology: Male Reproductive Health and Dysfunction*. Springer Science & Business Media, 13-Jan-2010. Chapter 9, page 137, <https://books.google.co.in/books?id=mEgckDNkonUC&pg=PA137&dq=normal+semen+parameters+asthenozoospermia+teratozoospermia+oligozoospermia&hl=en&sa=X&ei=GOC3VK2tGMe5u>

[ATvxYH4Dw&ved=0CCEQ6AEwAQ#v=onepage&q=normal%20semen%20parameters%20asthe nozoospermia%20teratozoospermia%20oligozoospermia&f=false.](http://www.ncbi.nlm.nih.gov/pubmed/20888888)

90. Jarvi K, Lo K, Fischer A, Grantmyre J, Zini A, Chow V, Mak V. CUA Guideline: The workup of azoospermic males. *Can Urol Assoc J.* 2010 Jun;4(3):163-7.
91. Sharif K. Reclassification of azoospermia: the time has come? *Hum Reprod.* 2000 Feb;15(2):237-8.
92. Antonio Luigi Pastore, Giovanni Palleschi, Luigi Si Ivestri, Antonino Leto and Antonio Carbone (2012). *Obstructive and Non-Obstructive Azoospermia, Male Infertility*, Dr. Anu Bashamboo (Ed.), ISBN: 978-953-51-0562-6, InTech, Available from: <http://www.intechopen.com/books/male-infertility/obstructive-and-non-obstructive-azoospermia>
93. Fedder J, Crüger D, Oestergaard B, Petersen GB. Etiology of azoospermia in 100 consecutive nonvasectomized men. *Fertil Steril.* 2004 Nov;82(5):1463-5.
94. M Rashed, N Ragab, A Shalaby, W Ragab. Patterns Of Testicular Histopathology In Men With Primary Infertility. *The Internet Journal of Urology.* 2007 Volume 5 Number 2.
95. Vernaev V, Verheyen G, Goossens A, Van Steirteghem A, Devroey P, Tournaye H. How successful is repeat testicular sperm extraction in patients with azoospermia? *Hum Reprod.* 2006 Jun;21(6):1551-4.
96. Lee JY, Dada R, Sabanegh E, Carpi A, Agarwal A. Role of genetics in azoospermia. *Urology.* 2011 Mar;77(3):598-601., Hamada AJ, Esteves SC, Agarwal A. A comprehensive review of genetics and genetic testing in azoospermia. *Clinics (Sao Paulo).* 2013;68 Suppl 1:39-60.
97. Gallego A, Rogel R, Luján S, Plaza B, Delgado F, Boronat F. AZF gene microdeletions: Case series and literature review. *Actas Urol Esp.* 2014 Jun 18. pii: S0210-4806(14)00146-6.
98. Zaimy MA, Kalantar SM, Sheikhha MH, Jahaninejad T, Pashaiefar H, Ghasemzadeh J, Zahraei M. The frequency of Yq microdeletion in azoospermic and oligospermic Iranian infertile men. *Iran J Reprod Med.* 2013 Jun;11(6):453-8.
99. Damyanova V, Dimitrova-Dikanarova D, Hadjidekova S, Savov A, Nesheva D, Rukova B, Vatev I, Toncheva D. [Genomic study in patients with idiopathic azoospermia and oligoasthenoteratozoospermia]. *Akush Ginekol (Sofia).* 2013;52(5):27-34.
100. Alhalabi M, Kenj M, Monem F, Mahayri Z, Abou Alchamat G, Madania A. High prevalence of genetic abnormalities in Middle Eastern patients with idiopathic non-obstructive azoospermia. *J Assist Reprod Genet.* 2013 Jun;30(6):799-805.
101. Vozdova M, Heracek J, Sobotka V, Rubes J. Testicular sperm aneuploidy in non-obstructive azoospermic patients. *Hum Reprod.* 2012 Jul;27(7):2233-9.
102. Zhang Y, He XJ, Song B, Ye L, Xie XS, Ruan J, Zhou FS, Zuo XB, Cao YX, Du WD. Association of single nucleotide polymorphisms in the USF1, GTF2A1L and OR2W3 genes with non-obstructive azoospermia in the Chinese population. *J Assist Reprod Genet.* 2014 Nov 6.
103. Zhang Y, Song B, Du WD, He XJ, Ruan J, Zhou FS, Zuo XB, Ye L, Xie XS, Cao YX. Genetic association study of RNF8 and BRDT variants with non-obstructive azoospermia in the Chinese Han population. *Syst Biol Reprod Med.* 2014 Nov 6:1-6.
104. He XJ, Song B, Du WD, Cao YX, Zhang Y, Ruan J, Tian H, Zhou FS, Zuo XB, Wu H, Zha X, Xie XS, Wei ZL, Zhou P. CREM variants rs4934540 and rs2295415 conferred susceptibility to nonobstructive azoospermia risk in the Chinese population. *Biol Reprod.* 2014 Aug;91(2):52.

-
105. Fedick AM, Eckert K, Thompson K, Forman EJ, Devkota B, Treff NR, Taylor D, Scott RT Jr. Lack of association of KATNAL1 gene sequence variants and azoospermia in humans. *J Assist Reprod Genet.* 2014 Aug;31(8):1065-71.
 106. Hu Z, Li Z, Yu J, Tong C, Lin Y, Guo X, Lu F, Dong J, Xia Y, Wen Y, Wu H, Li H, Zhu Y, Ping P, Chen X, Dai J, Jiang Y, Pan S, Xu P, Luo K, Du Q, Yao B, Liang M, Gui Y, Weng N, Lu H, Wang Z, Zhang F, Zhu X, Yang X, Zhang Z, Zhao H, Xiong C, Ma H, Jin G, Chen F, Xu J, Wang X, Zhou Z, Chen ZJ, Liu J, Shen H, Sha J. Association analysis identifies new risk loci for non-obstructive azoospermia in Chinese men. *Nat Commun.* 2014 May 23;5:3857.
 107. Zhao H, Xu J, Zhang H, Sun J, Sun Y, Wang Z, Liu J, Ding Q, Lu S, Shi R, You L, Qin Y, Zhao X, Lin X, Li X, Feng J, Wang L, Trent JM, Xu C, Gao Y, Zhang B, Gao X, Hu J, Chen H, Li G, Zhao J, Zou S, Jiang H, Hao C, Zhao Y, Ma J, Zheng SL, Chen ZJ. A genome-wide association study reveals that variants within the HLA region are associated with risk for nonobstructive azoospermia. *Am J Hum Genet.* 2012 May 4;90(5):900-6.
 108. Tewes AC, Ledig S, Tüttelmann F, Kliesch S, Wieacker P. DMRT1 mutations are rarely associated with male infertility. *Fertil Steril.* 2014 Sep;102(3):816-820.e3.
 109. Hojati Z, Heidari S, Motovali-Bashi M. Exon 10 CFTR gene mutation in male infertility. *Iran J Reprod Med.* 2012 Jul;10(4):315-20.
 110. Wu W, Hu Z, Qin Y, Dong J, Dai J, Lu C, Zhang W, Shen H, Xia Y, Wang X. Seminal plasma microRNAs: potential biomarkers for spermatogenesis status. *Mol Hum Reprod.* 2012 Oct;18(10):489-97.
 111. Slezak R, Szczepaniak M, Pasińska M, Czemarmazowicz H. [The analysis of CFTR mutations in men with azoospermia, oligozoospermia and asthenozoospermia]. *Ginekol Pol.* 2007 Aug;78(8):605-10.
 112. Ramasamy R, Ridgeway A, Lipshultz LI, Lamb DJ. Integrative DNA methylation and gene expression analysis identifies discoidin domain receptor 1 association with idiopathic nonobstructive azoospermia. *Fertil Steril.* 2014 Oct;102(4):968-973.e3.
 113. Sugimoto K, Koh E, Iijima M, Taya M, Maeda Y, Namiki M. Aberrant methylation of the TDMR of the GTF2A1L promoter does not affect fertilisation rates via TESE in patients with hypospermatogenesis. *Asian J Androl.* 2013 Sep;15(5):634-9.
 114. Minor A, Chow V, Ma S. Aberrant DNA methylation at imprinted genes in testicular sperm retrieved from men with obstructive azoospermia and undergoing vasectomy reversal. *Reproduction.* 2011 Jun;141(6):749-57.
 115. Abu-Halima M, Hammadeh M, Backes C, Fischer U, Leidinger P, Lubbad AM, Keller A, Meese E. Panel of five microRNAs as potential biomarkers for the diagnosis and assessment of male infertility. *Fertil Steril.* 2014 Oct;102(4):989-997.e1.
 116. Wu W, Qin Y, Li Z, Dong J, Dai J, Lu C, Guo X, Zhao Y, Zhu Y, Zhang W, Hang B, Sha J, Shen H, Xia Y, Hu Z, Wang X. Genome-wide microRNA expression profiling in idiopathic non-obstructive azoospermia: significant up-regulation of miR-141, miR-429 and miR-7-1-3p. *Hum Reprod.* 2013 Jul;28(7):1827-36.
 117. Wang C, Yang C, Chen X, Yao B, Yang C, Zhu C, Li L, Wang J, Li X, Shao Y, Liu Y, Ji J, Zhang J, Zen K, Zhang CY, Zhang C. Altered profile of seminal plasma microRNAs in the molecular diagnosis of male infertility. *Clin Chem.* 2011 Dec;57(12):1722-31.
 118. Christman MS, Gudeman SR, Nork JJ, Walters RC, L'Esperance JO, Crain DS. Operating characteristics of follicle-stimulating hormone in azoospermic men. *Fertil Steril.* 2014 May;101(5):1261-5.

-
119. Goulis DG, Iliadou PK, Tsametis C, Gerou S, Tarlatzis BC, Bontis IN, Papadimas I. Serum anti-Müllerian hormone levels differentiate control from subfertile men but not men with different causes of subfertility. *Gynecol Endocrinol*. 2008 Mar;24(3):158-60.
 120. Drabovich AP, Dimitromanolakis A, Saraon P, Soosaipillai A, Batruch I, Mullen B, Jarvi K, Diamandis EP. Differential diagnosis of azoospermia with proteomic biomarkers ECM1 and TEX101 quantified in seminal plasma. *Sci Transl Med*. 2013 Nov 20;5(212):212ra160.
 121. Batruch I, Smith CR, Mullen BJ, Grober E, Lo KC, Diamandis EP, Jarvi KA. Analysis of seminal plasma from patients with non-obstructive azoospermia and identification of candidate biomarkers of male infertility. *J Proteome Res*. 2012 Mar 2;11(3):1503-11.
 122. Drabovich AP, Jarvi K, Diamandis EP. Verification of male infertility biomarkers in seminal plasma by multiplex selected reaction monitoring assay. *Mol Cell Proteomics*. 2011 Dec;10(12):M110.004127.
 123. Bai J, Fu SH, Cai LL, Sun L, Cong YL. [Identification of differential proteins in the seminal plasma of healthy fertile and non-obstructive azoospermia men by shotgun proteomic strategy]. *Zhonghua Nan Ke Xue*. 2010 Oct;16(10):887-96.
 124. Malcher A, Rozwadowska N, Stokowy T, Kolanowski T, Jedrzejczak P, Zietkowiak W, Kurpisz M. Potential biomarkers of nonobstructive azoospermia identified in microarray gene expression analysis. *Fertil Steril*. 2013 Dec;100(6):1686-94.e1-7.
 125. Malcher A, Rozwadowska N, Stokowy T, Jedrzejczak P, Zietkowiak W, Kurpisz M. The gene expression analysis of paracrine/autocrine factors in patients with spermatogenetic failure compared with normal spermatogenesis. *Am J Reprod Immunol*. 2013 Dec;70(6):522-8.
 126. Okada H, Tajima A, Shichiri K, Tanaka A, Tanaka K, Inoue I. Genome-wide expression of azoospermia testes demonstrates a specific profile and implicates ART3 in genetic susceptibility. *PLoS Genet*. 2008 Feb;4(2):e26.
 127. Yang B, Yuan JL, Gao XK, Liu HL, Qin WJ, Shao C, Liu F, Kang FX. [Expressions of cadherin molecules CDH18 and PCDH17 in human azoospermic testes]. *Zhonghua Nan Ke Xue*. 2009 Dec;15(12):1081-4.
 128. Yang B, Yuan JL, Gao XK, Wang H, Shao C, Liu HL, Chen BQ, Qin RL, Shao GX, Kang FX. [Expression of COX10 in human non-obstructive azoospermia testes]. *Zhonghua Nan Ke Xue*. 2009 Jul;15(7):599-603.
 129. Aslani F, Modarresi MH, Soltangharaee H, Akhondi MM, Shabani A, Lakpour N, Sadeghi MR. Seminal molecular markers as a non-invasive diagnostic tool for the evaluation of spermatogenesis in non-obstructive azoospermia. *Syst Biol Reprod Med*. 2011 Aug;57(4):190-6.
 130. Toulis KA, Iliadou PK, Venetis CA, Tsametis C, Tarlatzis BC, Papadimas I, Goulis DG. Inhibin B and anti-Müllerian hormone as markers of persistent spermatogenesis in men with non-obstructive azoospermia: a meta-analysis of diagnostic accuracy studies. *Hum Reprod Update*. 2010 Nov-Dec;16(6):713-24.
 131. Bonaparte E, Moretti M, Colpi GM, Nerva F, Contalbi G, Vaccalluzzo L, Tabano S, Grati FR, Gazzano G, Sirchia SM, Simoni G, Gallina A, Miozzo M. ESX1 gene expression as a robust marker of residual spermatogenesis in azoospermic men. *Hum Reprod*. 2010 Jun;25(6):1398-403.
 132. Kalsi JS, Shah P, Thum Y, Muneer A, Ralph DJ, Minhas S. Salvage micro-dissection testicular sperm extraction; Outcome in men with Non obstructive azoospermia with previous failed sperm retrievals. *BJU Int*. 2014 Sep 15.

-
133. Modarresi T, Sabbaghian M, Shahverdi A, Hosseinfar H, Akhlaghi AA, Sadighi Gilani MA. Enzymatic digestion improves testicular sperm retrieval in non-obstructive azoospermic patients. *Iran J Reprod Med*. 2013 Jun;11(6):447-52.
 134. Shin DH, Turek PJ. Sperm retrieval techniques. *Nat Rev Urol*. 2013 Dec;10(12):723-30.
 135. Shinjo E, Shiraishi K, Matsuyama H. The effect of human chorionic gonadotropin-based hormonal therapy on intratesticular testosterone levels and spermatogonial DNA synthesis in men with non-obstructive azoospermia. *Andrology*. 2013 Nov;1(6):929-35.
 136. Hussein A, Ozgok Y, Ross L, Rao P, Niederberger C. Optimization of spermatogenesis-regulating hormones in patients with non-obstructive azoospermia and its impact on sperm retrieval: a multicentre study. *BJU Int*. 2013 Mar;111(3 Pt B):E110-4.
 137. Shiraishi K, Ohmi C, Shimabukuro T, Matsuyama H. Human chorionic gonadotrophin treatment prior to microdissection testicular sperm extraction in non-obstructive azoospermia. *Hum Reprod*. 2012 Feb;27(2):331-9.
 138. Cavallini G, Beretta G, Biagiotti G. Preliminary study of letrozole use for improving spermatogenesis in non-obstructive azoospermia patients with normal serum FSH. *Asian J Androl*. 2011 Nov;13(6):895-7.
 139. Kumar R. Medical management of non-obstructive azoospermia. *Clinics (Sao Paulo)*. 2013;68 Suppl 1:75-9.
 140. Berget SM, Moore C, Sharp PA. Spliced segments at the 5' terminus of adenovirus 2 late mRNA. *Proc Natl Acad Sci U S A*. 1977 Aug;74(8):3171-5.
 141. Chow LT, Gelinas RE, Broker TR, Roberts RJ. An amazing sequence arrangement at the 5' ends of adenovirus 2 messenger RNA. *Cell*. 1977 Sep;12(1):1-8.
 142. Clancy, S. (2008) RNA splicing: introns, exons and spliceosome. *Nature Education* 1(1):31.
 143. UCSC table browser: <http://genome.ucsc.edu/cgi-bin/hgTables>
 144. Cooper GM, Sunderland (MA). *The Cell: A Molecular Approach*. 2nd edition. Sinauer Associates; 2000. Chapter 6, <http://www.ncbi.nlm.nih.gov/books/NBK9864/>
 145. Black DL. Mechanisms of alternative pre-messenger RNA splicing. *Annu Rev Biochem*. 2003;72:291-336.
 146. Alberts B, Johnson A, Lewis J, et al. *Molecular Biology of the Cell*. 4th edition. New York: Garland Science; 2002. Chapter 6, <http://www.ncbi.nlm.nih.gov/books/NBK26887/>.
 147. Steitz JA, Dreyfuss G, Krainer AR, Lamond AI, Matera AG, Padgett RA. Where in the cell is the minor spliceosome? *Proc Natl Acad Sci U S A*. 2008 Jun 24;105(25):8485-6.]
 148. Early P, Rogers J, Davis M, Calame K, Bond M, Wall R, Hood L. Two mRNAs can be produced from a single immunoglobulin mu gene by alternative RNA processing pathways. *Cell*. 1980 Jun;20(2):313-9.
 149. Pan Q, Shai O, Lee LJ, Frey BJ, Blencowe BJ: Deep surveying of alternative splicing complexity in the human transcriptome by high-throughput sequencing. *Nat Genet* 2008, 40:1413-1415.
 150. Celotto AM, Graveley BR. Alternative splicing of the *Drosophila* Dscam pre-mRNA is both temporally and spatially regulated. *Genetics*. 2001 Oct;159(2):599-608.

-
151. Schmucker D, Clemens JC, Shu H, Worby CA, Xiao J, Muda M, Dixon JE, Zipursky SL. *Drosophila Dscam* is an axon guidance receptor exhibiting extraordinary molecular diversity. *Cell*. 2000 Jun 9;101(6):671-84.
 152. Kim E, Magen A, Ast G. Different levels of alternative splicing among eukaryotes. *Nucleic Acids Res*. 2007;35(1):125-31.
 153. Florea L, Song L, Salzberg SL. Thousands of exon skipping events differentiate among splicing patterns in sixteen human tissues. Version 2. *F1000Res*. 2013 Sep 16 [revised 2013 Nov 21];2:188.
 154. Yeo G, Holste D, Kreiman G, Burge CB. Variation in alternative splicing across human tissues. *Genome Biol*. 2004;5(10):R74.
 155. Hertel KJ. Combinatorial control of exon recognition. *J Biol Chem*. 2008 Jan 18;283(3):1211-5.
 156. Mandal M, Breaker RR. Gene regulation by riboswitches. *Nat Rev Mol Cell Biol*. 2004 Jun;5(6):451-63.
 157. Serganov A, Nudler E. A decade of riboswitches. *Cell*. 2013 Jan 17;152(1-2):17-24.
 158. Chen M, Manley JL. Mechanisms of alternative splicing regulation: insights from molecular and genomics approaches. *Nat Rev Mol Cell Biol*. 2009 Nov;10(11):741-54.
 159. Wang Z, Burge CB. Splicing regulation: from a parts list of regulatory elements to an integrated splicing code. *RNA*. 2008 May;14(5):802-13.
 160. Kelemen O, Convertini P, Zhang Z, Wen Y, Shen M, Falaleeva M, Stamm S. Function of alternative splicing. *Gene*. 2013 Feb 1;514(1):1-30.
 161. Tazi J, Bakkour N, Stamm S. Alternative splicing and disease. *Biochim Biophys Acta*. 2009 Jan;1792(1):14-26.
 162. He C, Zuo Z, Chen H, Zhang L, Zhou F, Cheng H, Zhou R: Genome-wide detection of testis- and testicular cancer-specific alternative splicing. *Carcinogenesis* 2007, 28:2484-2490.
 163. Blair CA, Zi X: Potential molecular targeting of splice variants for cancer treatment. *Indian J Exp Biol* 2011, 49:836-9.
 164. Garcia-Blanco MA, Baraniak AP, Lasda EL: Alternative splicing in disease and therapy. *Nat Biotechnol* 2004, 22:535-46.
 165. Venables JP: Aberrant and alternative splicing in cancer. *Cancer Res* 2004, 64:7647-54.
 166. Garcia-Blanco MA: Alternative splicing: therapeutic target and tool. *Prog Mol Subcell Biol* 2006, 44:47-64.
 167. Gaur RK: RNA interference: a potential therapeutic tool for silencing splice isoforms linked to human diseases. *Biotechniques* 2006, (Suppl):15-22.
 168. Accelerating the Development of Biomarkers for Drug Safety: Workshop Summary. Institute of Medicine (US) Forum on Drug Discovery, Development, and Translation. Washington (DC): National Academies Press (US); 2009. <http://www.ncbi.nlm.nih.gov/books/NBK32710/#ch1.r1>
 169. Richard Mayeux, Biomarkers: Potential Uses and Limitations. *NeuroRx*. 2004 April; 1(2): 182–188.
 170. M. S. Kumar: Biomarkers of disease in medicine. *Current Trends in Science*, pp. 403–417, 2009.

Bibliography for chapter 2

1. Su AI, Cooke MP, Ching KA, Hakak Y, Walker JR, Wiltshire T, Orth AP, Vega RG, Sapinoso LM, Moqrich A, Patapoutian A, Hampton GM, Schultz PG, Hogenesch JB: Large-scale analysis of the human and mouse transcriptomes. *Proc Natl Acad Sci U S A* 2002, 99:4465-4470. (<http://biogps.org/>)
2. RefExA: http://157.82.78.238/refexa/main_search.jsp
3. Tanino M, Debily MA, Tamura T, Hishiki T, Ogasawara O, Murakawa K, Kawamoto S, Itoh K, Watanabe S, de Souza SJ, Imbeaud S, Graudens E, Eveno E, Hilton P, Sudo Y, Kelso J, Ikeo K, Imanishi T, Gojobori T, Auffray C, Hide W, Okubo K. The Human Anatomic Gene Expression Library (H-ANGEL), the H-Inv integrative display of human gene expression across disparate technologies and platforms. *Nucleic Acids Res.* 2005 Jan 1;33(Database issue):D567-72. (<http://www.h-invitational.jp/hinv/h-angel/>)
4. Lardenois A, Gattiker A, Collin O, Chalmel F, Primig M. GermOnline 4.0 is a genomics gateway for germline development, meiosis and the mitotic cell cycle. *Database (Oxford)*. 2010 Dec 11;2010:baq030. (<http://www.germonline.org/index.html>)
5. Lee TL, Cheung HH, Claus J, Sastry C, Singh S, Vu L, Rennert O, Chan WY: GermSAGE: a comprehensive SAGE database for transcript discovery on male germ cell development. *Nucleic Acids Res* 2009, 37:D891-D897. (<http://germsage.nichd.nih.gov/germsage/home.html>)
6. SAGE Genie: <http://cgap.nci.nih.gov/SAGE/AnatomicViewer>
7. Boguski MS, Schuler GD. ESTablishing a human transcript map. *Nat Genet* 1995, 10:369-371. (<http://www.ncbi.nlm.nih.gov/unigene>)
8. Liu X, Yu X, Zack DJ, Zhu H, Qian J. TiGER: a database for tissue-specific gene expression and regulation. *BMC Bioinformatics*. 2008 Jun 9;9:271. (<http://bioinfo.wilmer.jhu.edu/tiger/>)
9. Ogasawara O, Otsuji M, Watanabe K, Iizuka T, Tamura T, Hishiki T, Kawamoto S, Okubo K. BodyMap-Xs: anatomical breakdown of 17 million animal ESTs for cross-species comparison of gene expression. *Nucleic Acids Res.* 2006 Jan 1;34(Database issue):D628-31. (<http://bodymap.genes.nig.ac.jp/>)
10. Bianchi V, Colantoni A, Calderone A, Ausiello G, Ferrè F, Helmer-Citterich M. DBATE: database of alternative transcripts expression. *Database (Oxford)*. 2013 Jul 9;2013:bat050. (<http://bioinformatica.uniroma2.it/DBATE/>)
11. Krupp M, Marquardt JU, Sahin U, Galle PR, Castle J, Teufel A. RNA-Seq Atlas--a reference database for gene expression profiling in normal tissue by next-generation sequencing. *Bioinformatics*. 2012 Apr 15;28(8):1184-5. (http://medicalgenomics.org/rna_seq_atlas)
12. BlotBase: <http://www.medicalgenomics.org/Databases/BlotBase>
13. Keshava Prasad TS, Goel R, Kandasamy K, Keerthikumar S, Kumar S, Mathivanan S, Telikicherla D, Raju R, Shafreen B, Venugopal A, Balakrishnan L, Marimuthu A, Banerjee S, Somanathan DS, Sebastian A, Rani S, Ray S, Harrys Kishore CJ, Kanth S, Ahmed M, Kashyap MK, Mohmood R, Ramachandra YL, Krishna V, Rahiman BA, Mohan S, Ranganathan P, Ramabadran S, Chaerkady R, Pandey A: Human Protein Reference Database--2009 update. *Nucleic Acids Res* 2009, 37:D767-D772. (<http://www.hprd.org/>)
14. Culhane AC, Schröder MS, Sultana R, Picard SC, Martinelli EN, Kelly C, Haibe-Kains B, Kapushesky M, St Pierre AA, Flahive W, Picard KC, Gusenleitner D, Papenhausen G, O'Connor N, Correll M, Quackenbush J. GeneSigDB: a manually curated database and resource for analysis of gene expression signatures. *Nucleic Acids Res.* 2012, (<http://compbio.dfci.harvard.edu/genesigdb/>)

15. <http://www.uniprot.org/uniprot/?query=reviewed%3Ayes>
16. Barrett T, Troup DB, Wilhite SE, Ledoux P, Evangelista C, Kim IF, Tomashevsky M, Marshall KA, Phillippy KH, Sherman PM, Muetter RN, Holko M, Ayanbule O, Yefanov A, Soboleva A. NCBI GEO: archive for functional genomics data sets--10 years on. *Nucleic Acids Res.* 2011, 39(Database issue):D1005-1010. (<http://www.ncbi.nlm.nih.gov/geo/>)
17. Parkinson H, Kapushesky M, Kolesnikov N, Rustici G, Shojatalab M, Abeygunawardena N, Berube H, Dylag M, Emam I, Farne A, Holloway E, Lukk M, Malone J, Mani R, Pilicheva E, Rayner TF, Rezwan F, Sharma A, Williams E, Bradley XZ, Adamusiak T, Brandizi M, Burdett T, Coulson R, Krestyaninova M, Kurnosov P, Maguire E, Neogi SG, Rocca-Serra P, Sansone SA, Sklyar N, Zhao M, Sarkans U, Brazma A. ArrayExpress update--from an archive of functional genomics experiments to the atlas of gene expression. *Nucleic Acids Res.* 2009, 37(Database issue):D868-872. (<https://www.ebi.ac.uk/arrayexpress/>)
18. Startbioinfo portal: <http://www.startbioinfo.com/gene-expression>
19. Skrabanek L, Campagne F. TissueInfo: high-throughput identification of tissue expression profiles and specificity. *Nucleic Acids Res.* 2001 Nov 1;29(21):E102-2. (<http://pbttest.med.cornell.edu/services/tissueinfo/query>)
20. CleanEx: <http://cleanex.vital-it.ch/>
21. Ochsner SA, Steffen DL, Stoeckert CJ Jr, McKenna NJ. Much room for improvement in deposition rates of expression microarray datasets. *Nat Methods.* 2008 Dec;5(12):991.
22. Irizarry RA, Warren D, Spencer F, Kim IF, Biswal S, Frank BC, Gabrielson E, Garcia JG, Geoghegan J, Germino G, Griffin C, Hilmer SC, Hoffman E, Jedlicka AE, Kawasaki E, Martínez-Murillo F, Morsberger L, Lee H, Petersen D, Quackenbush J, Scott A, Wilson M, Yang Y, Ye SQ, Yu W. Multiple-laboratory comparison of microarray platforms. *Nat Methods* 2005, 2:345-350.
23. Shi L, Tong W, Fang H, Scherf U, Han J, Puri RK, Frueh FW, Goodsaid FM, Guo L, Su Z, Han T, Fuscoe JC, Xu ZA, Patterson TA, Hong H, Xie Q, Perkins RG, Chen JJ, Casciano DA. Cross-platform comparability of microarray technology: intra-platform consistency and appropriate data analysis procedures are essential. *BMC Bioinformatics* 2005, 6 (Suppl 2):S12.
24. Cahan P, Rovegno F, Mooney D, Newman JC, St Laurent G 3rd, McCaffrey TA. Meta-analysis of microarray results: challenges, opportunities, and recommendations for standardization. *Gene* 2007, 401:12-18.
25. Wilkes T, Laux H, Foy CA. Microarray data quality - review of current developments. *OMICS* 2007, 11:1-13.
26. Wilder SP, Kaisaki PJ, Argoud K, Salhan A, Ragoussis J, Bihoreau MT, Gauguier D. Comparative analysis of methods for gene transcription profiling data derived from different microarray technologies in rat and mouse models of diabetes. *BMC Genomics* 2009, 10:63.
27. Brazma A, Hingamp P, Quackenbush J, Sherlock G, Spellman P, Stoeckert C, Aach J, Ansorge W, Ball CA, Causton HC, Gaasterland T, Glenisson P, Holstege FC, Kim IF, Markowitz V, Matese JC, Parkinson H, Robinson A, Sarkans U, Schulze-Kremer S, Stewart J, Taylor R, Vilo J, Vingron M. Minimum information about a microarray experiment (MIAME)-toward standards for microarray data. *Nat Genet* 2001, 29:365-371.
28. Spellman PT, Miller M, Stewart J, Troup C, Sarkans U, Chervitz S, Bernhart D, Sherlock G, Ball C, Lepage M, Swiatek M, Marks WL, Goncalves J, Markel S, Jordan D, Shojatalab M, Pizarro A, White J, Hubley R, Deutsch E, Senger M, Aronow BJ, Robinson A, Bassett D, Stoeckert CJ, Jr., Brazma A. Design and implementation of microarray gene expression markup language (MAGE-ML). *Genome Biol* 2002, 3:0046.1-0046.9.

-
29. Subramanian A, Tamayo P, Mootha VK, Mukherjee S, Ebert BL, Gillette MA, Paulovich A, Pomeroy SL, Golub TR, Lander ES, Mesirov JP. Gene set enrichment analysis: a knowledge-based approach for interpreting genome-wide expression profiles. *Proc Natl Acad Sci U S A*. 2005 Oct 25;102(43):15545-50.
 30. Choi H, Shen R, Chinnaiyan AM, Ghosh D. A latent variable approach for meta-analysis of gene expression data from multiple microarray experiments. *BMC Bioinformatics*. 2007 Sep 27;8:364.
 31. Pihur V, Datta S, Datta S. Finding common genes in multiple cancer types through meta-analysis of microarray experiments: a rank aggregation approach. *Genomics*. 2008 Dec;92(6):400-3.
 32. Krallinger M, Valencia A, Hirschman L. Linking genes to literature: text mining, information extraction, and retrieval applications for biology. *Genome Biol*. 2008;9 Suppl 2:S8.
 33. Burge S, Attwood TK, Bateman A, Berardini TZ, Cherry M, O'Donovan C, Xenaris L, Gaudet P. Biocurators and biocuration: surveying the 21st century challenges. *Database (Oxford)*. 2012 Mar 20;2012:bar059
 34. Petrusz P, Jeyaraj DA, Grossman G. Microarray analysis of androgen-regulated gene expression in testis: the use of the androgen-binding protein (ABP)-transgenic mouse as a model. *Reprod Biol Endocrinol* 2005, 3:70.
 35. Sadate-Ngatchou PI, Pouchnik DJ, Griswold MD. Follicle-stimulating hormone induced changes in gene expression of murine testis. *Mol Endocrinol*. 2004 Nov;18(11):2805-16.
 36. Almstrup K, Hoei-Hansen CE, Nielsen JE, Wirkner U, Ansorge W, Skakkebaek NE, Rajpert-De Meyts E, Leffers H. Genome-wide gene expression profiling of testicular carcinoma in situ progression into overt tumours. *Br J Cancer* 2005, 92:1934-1941.
 37. Rhodes DR, Yu J, Shanker K, Deshpande N, Varambally R, Ghosh D, Barrette T, Pandey A, Chinnaiyan AM. ONCOMINE: a cancer microarray database and integrated data-mining platform. *Neoplasia* 2004, 6:1-6. (<https://www.oncomine.org/resource/login.html>)
 38. Howe D, Costanzo M, Fey P, Gojobori T, Hannick L, Hide W, Hill DP, Kania R, Schaeffer M, St Pierre S, Twigger S, White O, Rhee SY. Big data: The future of biocuration. *Nature*. 2008 Sep 4;455(7209):47-50.
 39. Schlatt S, Meinhardt A, Nieschlag E. Paracrine regulation of cellular interactions in the testis: factors in search of a function. *Eur J Endocrinol*. 1997 Aug;137(2):107-17.
 40. Soumillon M, Necsulea A, Weier M, Brawand D, Zhang X, Gu H, Barthès P, Kokkinaki M, Nef S, Gnirke A, Dym M, de Massy B, Mikkelsen TS, Kaessmann H. Cellular source and mechanisms of high transcriptome complexity in the mammalian testis. *Cell Rep*. 2013 Jun 27;3(6):2179-90.
 41. PubMed: <http://www.ncbi.nlm.nih.gov/pubmed>
 42. Yan N, Lu Y, Sun H, Qiu W, Tao D, Liu Y, Chen H, Yang Y, Zhang S, Li X, Ma Y. Microarray profiling of microRNAs expressed in testis tissues of developing primates. *J Assist Reprod Genet*. 2009 Apr;26(4):179-86.
 43. Antonangeli F, Giampietri C, Petrungaro S, Filippini A, Ziparo E. Expression profile of a 400-bp *Stra8* promoter region during spermatogenesis. *Microsc Res Tech*. 2009 Nov;72(11):816-22.
 44. Maugars G, Schmitz M. Gene expression profiling during spermatogenesis in early maturing male Atlantic salmon parr testes. *Gen Comp Endocrinol*. 2008 Nov-Dec;159(2-3):178-87.
 45. Shen XY, Cui JZ, Gong QL, Liu YJ, Nagahama Y. Transcript expression profiles of *Takifugu rubripes* spermatozoa and eggs by expressed sequence tag analysis. *Fish Physiol Biochem*. 2008 Sep;34(3):235-43.

-
46. Nie D, Yang X, Yankai Z. Molecular cloning and expression profile analysis of a novel mouse testis-specific expression gene mtlQ1. *Mol Biol Rep*. 2009 May;36(5):1203-9.
 47. Domae S, Nakamura Y, Nakamura Y, Uenaka A, Wada H, Nakata M, Oka M, Kishimoto K, Tsukamoto G, Yoshihama Y, Matsuoka J, Gochi A, Kohno S, Saika T, Sasaki A, Nakayama E, Ono T. Identification of CCDC62-2 as a novel cancer/testis antigen and its immunogenicity. *Int J Cancer*. 2009 May 15;124(10):2347-52.
 48. Leman ES, Magheli A, Yong KM, Netto G, Hinz S, Getzenberg RH. Identification of nuclear structural protein alterations associated with seminomas. *J Cell Biochem*. 2009 Dec 15;108(6):1274-9.
 49. Richie JP. Gene expression profiling of early- and late-relapse nonseminomatous germ cell tumor and primitive neuroectodermal tumor of the testis. *J Urol*. 2005 Nov;174(5):1826-7.
 50. Zendman AJ, Zschocke J, van Kraats AA, de Wit NJ, Kurpysz M, Weidle UH, Ruitter DJ, Weiss EH, van Muijen GN. The human SPANX multigene family: genomic organization, alignment and expression in male germ cells and tumor cell lines. *Gene*. 2003 May 8;309(2):125-33.
 51. Demeter J, Beauheim C, Gollub J, Hernandez-Boussard T, Jin H, Maier D, Matese JC, Nitzberg M, Wymore F, Zachariah ZK, Brown PO, Sherlock G, Ball CA. The Stanford Microarray Database: implementation of new analysis tools and open source release of software. *Nucleic Acids Res* 2007, 35:D766-D770. (<http://smd.princeton.edu/>)
 52. Ikeo K, Ishi-i J, Tamura T, Gojobori T, Tateno Y. CIBEX: center for information biology gene expression database. *C R Biol* 2003, 326:1079-1082. (<http://cibex.nig.ac.jp/data/index.html>)
 53. ATCC: <http://www.atcc.org/Products/Cells%20and%20Microorganisms/Cell%20Lines/Human.aspx>
 54. Mouse atlas project: <http://genex.hgu.mrc.ac.uk/Databases/Anatomy/MAstaging.shtml>
 55. Okada H, Tajima A, Shichiri K, Tanaka A, Tanaka K, Inoue I. Genome-wide expression of azoospermia testes demonstrates a specific profile and implicates ART3 in genetic susceptibility. *PLoS Genet*. 2008 Feb;4(2):e26.
 56. Acharya KK, Chandrashekar DS, Chitturi N, Shah H, Malhotra V, Sreelakshmi KS, Deepti H, Bajpai A, Davuluri S, Bora P, Rao L. A novel tissue-specific meta-analysis approach for gene expression predictions, initiated with a mammalian gene expression testis database. *BMC Genomics* 2010, 11:467.
 57. NCBI gene database: <http://www.ncbi.nlm.nih.gov/gene>
 58. Sunitha Kogenaru, Coral del Val, Agnes Hotz-Wagenblatt, Karl-Heinz Glatting. TissueDistributionDBs: a repository of organism-specific tissue-distribution profiles. March 2010, Volume 125, Issue 3-6, pp 651-658. (http://genius.embnet.dkfz-heidelberg.de/menu/tissue_db/)
 59. Bgee database: <http://bgee.unil.ch/bgee/bgee>
 60. Finger JH, Smith CM, Hayamizu TF, McCright IJ, Eppig JT, Kadin JA, Richardson JE, Ringwald M. The mouse Gene Expression Database (GXD): 2011 update. *Nucleic Acids Res*. 2011 Jan;39(Database issue):D835-41. (<http://www.informatics.jax.org/mgihome/GXD/aboutGXD.shtml>)
 61. MRG: <http://mrgd.org/>
 62. Haudry Y, Berube H, Letunic I, Weeber PD, Gagneur J, Girardot C, Kapushesky M, Arendt D, Bork P, Brazma A, Furlong EE, Wittbrodt J, Henrich T. 4DXpress: a database for cross-species expression pattern comparisons. *Nucleic Acids Res*. 2008 Jan;36(Database issue):D847-53. (<http://ani.embl.de/4DXpress>)

-
63. Dadoune JP, Pawlak A, Alfonsi MF, Siffroi JP. Identification of transcripts by macroarrays, RT-PCR and in situ hybridization in human ejaculate spermatozoa. *Mol Hum Reprod*. 2005 Feb;11(2):133-40.
 64. Lopez F, Textoris J, Bergon A, Didier G, Remy E, Granjeaud S, Imbert J, Nguyen C, Puthier D. TranscriptomeBrowser: a powerful and flexible toolbox to explore productively the transcriptional landscape of the Gene Expression Omnibus database. *PLoS One* 2008, 3:e4001. (<http://tagc.univ-mrs.fr/tbrowser/>)
 65. Hruz T, Laule O, Szabo G, Wessendorp F, Bleuler S, Oertle L, Widmayer P, Grissem W and P Zimmermann. Genevestigator V3: a reference expression database for the meta-analysis of transcriptomes. *Advances in Bioinformatics* 2008, 2008: 420747. (<https://genevestigator.com/>)
 66. Obayashi T, Hayashi S, Shibaoka M, Saeki M, Ohta H, Kinoshita K. COXPRESdb: a database of coexpressed gene networks in mammals. *Nucleic Acids Res* 2008, 36:D77-D82. (<http://coxpresdb.jp/>)
 67. Ioannidis JP, Allison DB, Ball CA, Coulibaly I, Cui X, Culhane AC, Falchi M, Furlanello C, Game L, Jurman G, Mangion J, Mehta T, Nitzberg M, Page GP, Petretto E, van Noort V. Repeatability of published microarray gene expression analyses. *Nat Genet* 2009, 41:149-155.
 68. dbEST: <http://www.ncbi.nlm.nih.gov/dbEST/>
 69. Claussen S, Prokosch HU, Nieschlag E, Gromoll J. TestisBank: an internet-based gene sequence database of the testis. *Int J Androl*. 2002 Jun;25(3):175-9. (<http://medweb.uni-muenster.de/TestisBank/>)
 70. dbTest: <http://www.bch.msu.edu/~zacharet/dbtest/>
 71. Gardiner-Garden M, Littlejohn TG. A comparison of microarray databases. *Brief Bioinform*. 2001 May;2(2):143-58. Erratum in: *Brief Bioinform* 2001 Sep;2(3):220.

Bibliography for chapter 3

1. Pan Q, Shai O, Lee LJ, Frey BJ, Blencowe BJ. Deep surveying of alternative splicing complexity in the human transcriptome by high-throughput sequencing. *Nat Genet.* 2008 Dec;40(12):1413-5.
2. Keren H, Lev-Maor G, Ast G. Alternative splicing and evolution: diversification, exon definition and function. *Nat Rev Genet.* 2010 May;11(5):345-55.
3. Breitbart RE, Andreadis A, Nadal-Ginard B. Alternative splicing: a ubiquitous mechanism for the generation of multiple protein isoforms from single genes. *Annu Rev Biochem.* 1987;56:467-95.
4. Kim E, Magen A, Ast G. Different levels of alternative splicing among eukaryotes. *Nucleic Acids Res.* 2007;35(1):125-31.
5. Florea L, Song L, Salzberg SL. Thousands of exon skipping events differentiate among splicing patterns in sixteen human tissues. Version 2. *F1000Res.* 2013 Sep 16 [revised 2013 Nov 21];2:188.
6. Yeo G, Holste D, Kreiman G, Burge CB. Variation in alternative splicing across human tissues. *Genome Biol.* 2004;5(10):R74.
7. Hertel KJ. Combinatorial control of exon recognition. *J Biol Chem.* 2008 Jan 18;283(3):1211-5.
8. Mabon SA, Misteli T. Differential recruitment of pre-mRNA splicing factors to alternatively spliced transcripts in vivo. *PLoS Biol.* 2005 Nov;3(11):e374.
9. Grosso AR, Gomes AQ, Barbosa-Morais NL, Caldeira S, Thorne NP, Grech G, von Lindern M, Carmo-Fonseca M. Tissue-specific splicing factor gene expression signatures. *Nucleic Acids Res.* 2008 Sep;36(15):4823-32.
10. Mandal M, Breaker RR. Gene regulation by riboswitches. *Nat Rev Mol Cell Biol.* 2004 Jun;5(6):451-63.
11. Serganov A, Nudler E. A decade of riboswitches. *Cell.* 2013 Jan 17;152(1-2):17-24.
12. Long JC, Caceres JF. The SR protein family of splicing factors: master regulators of gene expression. *Biochem J.* 2009 Jan 1;417(1):15-27.
13. Han SP, Tang YH, Smith R. Functional diversity of the hnRNPs: past, present and perspectives. *Biochem J.* 2010 Sep 15;430(3):379-92.
14. Black DL. Mechanisms of alternative pre-messenger RNA splicing. *Annu Rev Biochem.* 2003;72:291-336.
15. Nilsen TW, Graveley BR. Expansion of the eukaryotic proteome by alternative splicing. *Nature.* 2010 Jan 28;463(7280):457-63.
16. Kamma H, Portman DS, Dreyfuss G. Cell type-specific expression of hnRNP proteins. *Exp Cell Res.* 1995 Nov;221(1):187-96.
17. Cao H, Wu J, Lam S, Duan R, Newnham C, Molday RS, Graziotto JJ, Pierce EA, Hu J. Temporal and tissue specific regulation of RP-associated splicing factor genes PRPF3, PRPF31 and PRPC8--implications in the pathogenesis of RP. *PLoS One.* 2011 Jan 19;6(1):e15860.
18. Elliott DJ, Grellscheid SN. Alternative RNA splicing regulation in the testis. *Reproduction.* 2006 Dec;132(6):811-9.
19. de la Grange P, Gratadou L, Delord M, Dutertre M, Auboeuf D. Splicing factor and exon profiling across human tissues. *Nucleic Acids Res.* 2010 May;38(9):2825-38.

20. Yang J, Hung LH, Licht T, Kostin S, Looso M, Khrameeva E, Bindereif A, Schneider A, Braun T. RBM24 Is a Major Regulator of Muscle-Specific Alternative Splicing. *Dev Cell*. 2014 Oct 13;31(1):87-99.
21. Holste D, Ohler U. Strategies for identifying RNA splicing regulatory motifs and predicting alternative splicing events. *PLoS Comput Biol*. 2008 Jan;4(1):e21.
22. Pedrotti S, Busà R, Compagnucci C, Sette C. The RNA recognition motif protein RBM11 is a novel tissue-specific splicing regulator. *Nucleic Acids Res*. 2012 Feb;40(3):1021-32.
23. Buckanovich RJ, Posner JB, Darnell RB. Nova, the paraneoplastic Ri antigen, is homologous to an RNA-binding protein and is specifically expressed in the developing motor system. *Neuron*. 1993 Oct;11(4):657-72.
24. Polydorides AD, Okano HJ, Yang YY, Stefani G, Darnell RB. A brain-enriched polypyrimidine tract-binding protein antagonizes the ability of Nova to regulate neuron-specific alternative splicing. *Proc Natl Acad Sci U S A*. 2000 Jun 6;97(12):6350-5.
25. Warzecha CC, Sato TK, Nabet B, Hogenesch JB, Carstens RP. ESRP1 and ESRP2 are epithelial cell-type-specific regulators of FGFR2 splicing. *Mol Cell*. 2009 Mar 13;33(5):591-601.
26. Calarco JA, Superina S, O'Hanlon D, Gabut M, Raj B, Pan Q, Skalska U, Clarke L, Gelinas D, van der Kooy D, Zhen M, Ciruna B, Blencowe BJ. Regulation of vertebrate nervous system alternative splicing and development by an SR-related protein. *Cell*. 2009 Sep 4;138(5):898-910.
27. Akerman M, David-Eden H, Pinter RY, Mandel-Gutfreund Y. A computational approach for genome-wide mapping of splicing factor binding sites. *Genome Biol*. 2009;10(3):R30.
28. Wang XY, Zheng ZZ, Song HS, Xu YZ. Conserved RNA cis-elements regulate alternative splicing of Lepidopteran doublesex. *Insect Biochem Mol Biol*. 2014 Jan;44:1-11.
29. Thomas J, Palusa SG, Prasad KV, Ali GS, Surabhi GK, Ben-Hur A, Abdel-Ghany SE, Reddy AS. Identification of an intronic splicing regulatory element involved in auto-regulation of alternative splicing of SCL33 pre-mRNA. *Plant J*. 2012 Aug 22.
30. Lee J, Zhou J, Zheng X, Cho S, Moon H, Loh TJ, Jo K, Shen H. Identification of a novel cis-element that regulates alternative splicing of Bcl-x pre-mRNA. *Biochem Biophys Res Commun*. 2012 Apr 6;420(2):467-72.
31. Seth P, Walker LC, Yeowell HN. Identification of exonic cis-elements regulating the alternative splicing of scleroderma-associated lysyl hydroxylase 2 mRNA. *J Invest Dermatol*. 2011 Feb;131(2):537-9.
32. Streuli M, Saito H. Regulation of tissue-specific alternative splicing: exon-specific cis-elements govern the splicing of leukocyte common antigen pre-mRNA. *EMBO J*. 1989 Mar;8(3):787-96.
33. Cereda M, Pozzoli U, Rot G, Juvan P, Schweitzer A, Clark T, Ule J. RNAmotifs: prediction of multivalent RNA motifs that control alternative splicing. *Genome Biol*. 2014 Jan 31;15(1):R20.
34. Fairbrother WG, Yeh RF, Sharp PA, Burge CB. Predictive identification of exonic splicing enhancers in human genes. *Science*. 2002 Aug 9;297(5583):1007-13.
35. Cartegni L, Wang J, Zhu Z, Zhang MQ, Krainer AR. ESEfinder: A web resource to identify exonic splicing enhancers. *Nucleic Acids Res*. 2003 Jul 1;31(13):3568-71.
36. Paz I, Akerman M, Dror I, Kosti I, Mandel-Gutfreund Y. SFmap: a web server for motif analysis and prediction of splicing factor binding sites. *Nucleic Acids Res*. 2010 Jul;38(Web Server issue):W281-5.

-
37. Erkelenz S, Theiss S, Otte M, Widera M, Peter JO, Schaal H. Genomic HEXploring allows landscaping of novel potential splicing regulatory elements. *Nucleic Acids Res.* 2014;42(16):10681-97.
 38. Badr E, Heath LS. Identifying splicing regulatory elements with de bruijn graphs. *J Comput Biol.* 2014 Dec;21(12):880-97.
 39. Li W, Dai C, Kang S, Zhou XJ. Integrative analysis of many RNA-seq datasets to study alternative splicing. *Methods.* 2014 Jun 1;67(3):313-24.
 40. Zheng CL, Fu XD, Gribskov M. Characteristics and regulatory elements defining constitutive splicing and different modes of alternative splicing in human and mouse. *RNA.* 2005 Dec;11(12):1777-87.
 41. Voelker RB, Berglund JA. A comprehensive computational characterization of conserved mammalian intronic sequences reveals conserved motifs associated with constitutive and alternative splicing. *Genome Res.* 2007 Jul;17(7):1023-33.
 42. Aznarez I, Barash Y, Shai O, He D, Zielenski J, Tsui LC, Parkinson J, Frey BJ, Rommens JM, Blencowe BJ. A systematic analysis of intronic sequences downstream of 5' splice sites reveals a widespread role for U-rich motifs and TIA1/TIAL1 proteins in alternative splicing regulation. *Genome Res.* 2008 Aug;18(8):1247-58.
 43. Suyama M, Harrington ED, Vinokourova S, von Knebel Doeberitz M, Ohara O, Bork P. A network of conserved co-occurring motifs for the regulation of alternative splicing. *Nucleic Acids Res.* 2010 Dec;38(22):7916-26.
 44. Ke S, Chasin LA. Intronic motif pairs cooperate across exons to promote pre-mRNA splicing. *Genome Biol.* 2010;11(8):R84.
 45. Wen J, Chen Z, Cai X. A biophysical model for identifying splicing regulatory elements and their interactions. *PLoS One.* 2013;8(1):e54885.
 46. Brudno M, Gelfand MS, Spengler S, Zorn M, Dubchak I, Conboy JG. Computational analysis of candidate intron regulatory elements for tissue-specific alternative pre-mRNA splicing. *Nucleic Acids Res.* 2001 Jun 1;29(11):2338-48.
 47. Das D, Clark TA, Schweitzer A, Yamamoto M, Marr H, Arribere J, Minovitsky S, Poliakov A, Dubchak I, Blume JE, Conboy JG. A correlation with exon expression approach to identify cis-regulatory elements for tissue-specific alternative splicing. *Nucleic Acids Res.* 2007;35(14):4845-57.
 48. Castle JC, Zhang C, Shah JK, Kulkarni AV, Kalsotra A, Cooper TA, Johnson JM. Expression of 24,426 human alternative splicing events and predicted cis regulation in 48 tissues and cell lines. *Nat Genet.* 2008 Dec;40(12):1416-25.
 49. Wen J, Chiba A, Cai X. Computational identification of tissue-specific alternative splicing elements in mouse genes from RNA-Seq. *Nucleic Acids Res.* 2010 Dec;38(22):7895-907.
 50. Fagnani M, Barash Y, Ip JY, Misquitta C, Pan Q, Saltzman AL, Shai O, Lee L, Rozenhek A, Mohammad N, Willaime-Morawek S, Babak T, Zhang W, Hughes TR, van der Kooy D, Frey BJ, Blencowe BJ. Functional coordination of alternative splicing in the mammalian central nervous system. *Genome Biol.* 2007;8(6):R108.
 51. Wang X, Wang K, Radovich M, Wang Y, Wang G, Feng W, Sanford JR, Liu Y. Genome-wide prediction of cis-acting RNA elements regulating tissue-specific pre-mRNA alternative splicing. *BMC Genomics.* 2009 Jul 7;10 Suppl 1:S4.
 52. Llorian M, Schwartz S, Clark TA, Hollander D, Tan LY, Spellman R, Gordon A, Schweitzer AC, de la Grange P, Ast G, Smith CW. Position-dependent alternative splicing activity revealed by global

-
- profiling of alternative splicing events regulated by PTB. *Nat Struct Mol Biol.* 2010 Sep;17(9):1114-23.
53. Wang ET, Sandberg R, Luo S, Khrebtkova I, Zhang L, Mayr C, Kingsmore SF, Schroth GP, Burge CB. Alternative isoform regulation in human tissue transcriptomes. *Nature.* 2008 Nov 27;456(7221):470-6.
54. Kalsotra A, Xiao X, Ward AJ, Castle JC, Johnson JM, Burge CB, Cooper TA. A postnatal switch of CELF and MBNL proteins reprograms alternative splicing in the developing heart. *Proc Natl Acad Sci U S A.* 2008 Dec 23;105(51):20333-8.
55. Chen RL, Guo W, Shi Y, Wu H, Wang J, Sun G. Computational identification of specific splicing regulatory elements from RNA-seq in lung cancer. *Eur Rev Med Pharmacol Sci.* 2013 Jul;17(13):1716-21.
56. Helfman DM, Ricci WM. Branch point selection in alternative splicing of tropomyosin pre-mRNAs. *Nucleic Acids Res.* 1989 Jul 25;17(14):5633-50.
57. Gao K, Masuda A, Matsuura T, Ohno K. Human branch point consensus sequence is yUnAy. *Nucleic Acids Res.* 2008 Apr;36(7):2257-67.
58. Corvelo A, Hallegger M, Smith CW, Eyraas E. Genome-wide association between branch point properties and alternative splicing. *PLoS Comput Biol.* 2010 Nov 24;6(11):e1001016.
59. Fontana W, Konings DA, Stadler PF, Schuster P. Statistics of RNA secondary structures. *Biopolymers.* 1993 Sep;33(9):1389-404.
60. Hiller M, Zhang Z, Backofen R, Stamm S. Pre-mRNA secondary structures influence exon recognition. *PLoS Genet.* 2007 Nov;3(11):e204.
61. Eperon LP, Graham IR, Griffiths AD, Eperon IC. Effects of RNA secondary structure on alternative splicing of pre-mRNA: is folding limited to a region behind the transcribing RNA polymerase? *Cell.* 1988 Jul 29;54(3):393-401.
62. Muro AF, Caputi M, Pariyarath R, Pagani F, Buratti E, Baralle FE. Regulation of fibronectin EDA exon alternative splicing: possible role of RNA secondary structure for enhancer display. *Mol Cell Biol.* 1999 Apr;19(4):2657-71.
63. Jacquenet S, Ropers D, Bilodeau PS, Damier L, Mougins A, Stoltzfus CM, Branlant C. Conserved stem-loop structures in the HIV-1 RNA region containing the A3 3' splice site and its cis-regulatory element: possible involvement in RNA splicing. *Nucleic Acids Res.* 2001 Jan 15;29(2):464-78.
64. Franchina M, Hooper J, Kay PH. Five novel alternatively spliced transcripts of DNA (cytosine-5) methyltransferase 2 in human peripheral blood leukocytes. *Int J Biochem Cell Biol.* 2001 Nov;33(11):1104-15.
65. Warf MB, Berglund JA. Role of RNA structure in regulating pre-mRNA splicing. *Trends Biochem Sci.* 2010 Mar;35(3):169-78.
66. Buratti E, Baralle FE. Influence of RNA secondary structure on the pre-mRNA splicing process. *Mol Cell Biol.* 2004 Dec;24(24):10505-14.
67. Shepard PJ, Hertel KJ. Conserved RNA secondary structures promote alternative splicing. *RNA.* 2008 Aug;14(8):1463-9.
68. Zhang J, Kuo CC, Chen L. GC content around splice sites affects splicing through pre-mRNA secondary structures. *BMC Genomics.* 2011 Jan 31;12:90.

-
69. Vu MM, Jameson NE, Masuda SJ, Lin D, Larralde-Ridaura R, Lupták A. Convergent evolution of adenosine aptamers spanning bacterial, human, and random sequences revealed by structure-based bioinformatics and genomic SELEX. *Chem Biol.* 2012 Oct 26;19(10):1247-54.
 70. Bugała K, Zywicki M, Wyszko E, Barciszewska MZ, Barciszewski J. [Riboswitches]. *Postepy Biochem.* 2005;51(2):111-9.
 71. Kaempfer R. RNA sensors: novel regulators of gene expression. *EMBO Rep.* 2003 Nov;4(11):1043-7.
 72. Ray PS, Jia J, Yao P, Majumder M, Hatzoglou M, Fox PL. A stress-responsive RNA switch regulates VEGFA expression. *Nature.* 2009 Feb 12;457(7231):915-9.
 73. Arcondéguy T, Lacazette E, Millevoi S, Prats H, Touriol C. VEGF-A mRNA processing, stability and translation: a paradigm for intricate regulation of gene expression at the post-transcriptional level. *Nucleic Acids Res.* 2013 Sep;41(17):7997-8010.
 74. Chang TH, Huang HY, Hsu JB, Weng SL, Horng JT, Huang HD. An enhanced computational platform for investigating the roles of regulatory RNA and for identifying functional RNA motifs. *BMC Bioinformatics.* 2013;14 Suppl 2:S4.
 75. Chang TH, Huang HD, Wu LC, Yeh CT, Liu BJ, Horng JT. Computational identification of riboswitches based on RNA conserved functional sequences and conformations. *RNA.* 2009 Jul;15(7):1426-30.
 76. Abreu-Goodger C, Merino E. RibEx: a web server for locating riboswitches and other conserved bacterial regulatory elements. *Nucleic Acids Res.* 2005 Jul 1;33(Web Server issue):W690-2.
 77. Bengert P, Dandekar T. Riboswitch finder--a tool for identification of riboswitch RNAs. *Nucleic Acids Res.* 2004 Jul 1;32(Web Server issue):W154-9.
 78. PubMed: <http://www.ncbi.nlm.nih.gov/pubmed/>
 79. Iborra S, Hirschfeld M, Jaeger M, Zur Hausen A, Braicu I, Sehouli J, Gitsch G, Stickeler E. Alterations in expression pattern of splicing factors in epithelial ovarian cancer and its clinical impact. *Int J Gynecol Cancer.* 2013 Jul;23(6):990-6.
 80. Wong J. Altered expression of RNA splicing proteins in Alzheimer's disease patients: evidence from two microarray studies. *Dement Geriatr Cogn Dis Extra.* 2013 Mar 12;3(1):74-85.
 81. Giulietti M, Piva F, D'Antonio M, D'Onorio De Meo P, Paoletti D, Castrignanò T, D'Erchia AM, Picardi E, Zambelli F, Principato G, Pavesi G, Pesole G. SpliceAid-F: a database of human splicing factors and their RNA-binding sites. *Nucleic Acids Res.* 2013 Jan;41(Database issue):D125-31.
 82. Liu J, Huang B, Xiao Y, Xiong HM, Li J, Feng DQ, Chen XM, Zhang HB, Wang XZ. Aberrant expression of splicing factors in newly diagnosed acute myeloid leukemia. *Onkologie.* 2012;35(6):335-40.
 83. Sveen A, Agesen TH, Nesbakken A, Rognum TO, Lothe RA, Skotheim RI. Transcriptome instability in colorectal cancer identified by exon microarray analyses: Associations with splicing factor expression levels and patient survival. *Genome Med.* 2011 May 27;3(5):32.
 84. Piekielko-Witkowska A, Wiszomirska H, Wojcicka A, Poplawski P, Boguslawska J, Tanski Z, Nauman A. Disturbed expression of splicing factors in renal cancer affects alternative splicing of apoptosis regulators, oncogenes, and tumor suppressors. *PLoS One.* 2010 Oct 27;5(10):e13690.
 85. Grosso AR, Martins S, Carmo-Fonseca M. The emerging role of splicing factors in cancer. *EMBO Rep.* 2008 Nov;9(11):1087-93.

-
86. Barbosa-Morais NL, Carmo-Fonseca M, Aparicio S. Systematic genome-wide annotation of spliceosomal proteins reveals differential gene family expansion. *Genome Res.* 2006 Jan;16(1):66-77.
 87. SplicFac database: <http://rulai.cshl.edu/cgi-bin/SpliceFac/spliceFac.cgi?process=home>
 88. AMIGO GOOSE web-server: <http://www.berkeleybop.org/goose/>
 89. Acharya KK, Chandrashekar DS, Chitturi N, Shah H, Malhotra V, Sreelakshmi KS, Deepti H, Bajpai A, Davuluri S, Bora P, Rao L: A novel tissue-specific meta-analysis approach for gene expression predictions, initiated with a mammalian gene expression testis database. *BMC Genomics* 2010, 11:467.
 90. Huang DW, Sherman BT, Lempicki RA. Systematic and integrative analysis of large gene lists using DAVID Bioinformatics Resources. *Nature Protoc.* 2009;4(1):44-57, Huang DW, Sherman BT, Lempicki RA. Bioinformatics enrichment tools: paths toward the comprehensive functional analysis of large gene lists. *Nucleic Acids Res.* 2009;37(1):1-13.
 91. UCSC browser [<http://genome.ucsc.edu/>]
 92. Galaxy server [<https://usegalaxy.org/>]
 93. Bailey TL, Boden M, Buske FA, Frith M, Grant CE, Clementi L, Ren J, Li WW, Noble WS. MEME SUITE: tools for motif discovery and searching. *Nucleic Acids Res.* 2009 Jul;37(Web Server issue):W202-8.
 94. MEME suite: <http://ebi.edu.au/ftp/software/MEME/index.html>
 95. TOMTOM [<http://meme.nbcr.net/meme/cgi-bin/tomtom.cgi>]
 96. UCSC table browser [<http://genome.ucsc.edu/cgi-bin/hgTables>]
 97. Zuker M. Mfold web server for nucleic acid folding and hybridization prediction. *Nucleic Acids Res.* 2003 Jul 1;31(13):3406-15.
 98. Sang Won Lee, Liang Zhao, Arthur Pardi, and Tianbing Xia¹, Ultrafast Dynamics Show That the Theophylline and 3-Methylxanthine Aptamers Employ a Conformational Capture Mechanism for Binding Their Ligands. *Biochemistry.* 2010 April 6; 49(13): 2943–2951.
 99. blastn [<http://blast.ncbi.nlm.nih.gov/Blast.cgi>]
 100. Keshava Prasad TS, Goel R, Kandasamy K, Keerthikumar S, Kumar S, Mathivanan S, Telikicherla D, Raju R, Shafreen B, Venugopal A, Balakrishnan L, Marimuthu A, Banerjee S, Somanathan DS, Sebastian A, Rani S, Ray S, Harrys Kishore CJ, Kanth S, Ahmed M, Kashyap MK, Mohmood R, Ramachandra YL, Krishna V, Rahiman BA, Mohan S, Ranganathan P, Ramabadran S, Chaerkady R, Pandey A: Human Protein Reference Database--2009 update. *Nucleic Acids Res* 2009, 37:D767-D772.
 101. SwissProt [<http://www.uniprot.org/uniprot/?query=reviewed%3Ayes>]
 102. Russell R. RNA misfolding and the action of chaperones. *Front Biosci.* 2008 Jan 1;13:1-20.

Bibliography for chapter 4

1. Barrett T, Troup DB, Wilhite SE, Ledoux P, Evangelista C, Kim IF, Tomashevsky M, Marshall KA, Phillippy KH, Sherman PM, Muetter RN, Holko M, Ayanbule O, Yefanov A, Soboleva A. NCBI GEO: archive for functional genomics data sets--10 years on. *Nucleic Acids Res.* 2011, 39(Database issue):D1005-1010.
2. Parkinson H, Kapushesky M, Kolesnikov N, Rustici G, Shojatalab M, Abeygunawardena N, Berube H, Dylag M, Emam I, Farne A, Holloway E, Lukk M, Malone J, Mani R, Pilicheva E, Rayner TF, Rezwan F, Sharma A, Williams E, Bradley XZ, Adamusiak T, Brandizi M, Burdett T, Coulson R, Krestyaninova M, Kurnosov P, Maguire E, Neogi SG, Rocca-Serra P, Sansone SA, Sklyar N, Zhao M, Sarkans U, Brazma A. ArrayExpress update--from an archive of functional genomics experiments to the atlas of gene expression. *Nucleic Acids Res.* 2009, 37(Database issue):D868-872.
3. Bajpai AK, Davuluri S, Chandrashekar DS, Ilakya S, Dinakaran M, Acharya KK. MGEx-Udb: a mammalian uterus database for expression-based cataloguing of genes across conditions, including endometriosis and cervical cancer. *PLoS One* 2012, 7:e36776.
4. Pan Q, Shai O, Lee LJ, Frey BJ, Blencowe BJ. Deep surveying of alternative splicing complexity in the human transcriptome by high-throughput sequencing. *Nat Genet* 2008, 40:1413-1417.
5. He C, Zuo Z, Chen H, Zhang L, Zhou F, Cheng H, Zhou R. Genome-wide detection of testis- and testicular cancer-specific alternative splicing. *Carcinogenesis* 2007, 28:2484-2490.
6. Blair CA, Zi X. Potential molecular targeting of splice variants for cancer treatment. *Indian J Exp Biol* 2011, 49:836-9.
7. Garcia-Blanco MA, Baraniak AP, Lasda EL. Alternative splicing in disease and therapy. *Nat Biotechnol* 2004, 22:535-46.
8. Venables JP. Aberrant and alternative splicing in cancer. *Cancer Res* 2004, 64:7647-54.
9. Garcia-Blanco MA. Alternative splicing: therapeutic target and tool. *Prog Mol Subcell Biol* 2006, 44:47-64.
10. Gaur RK. RNA interference: a potential therapeutic tool for silencing splice isoforms linked to human diseases. *Biotechniques* 2006, (Suppl):15-22.
11. Lapuk A, Marr H, Jakkula L, Pedro H, Bhattacharya S, Purdom E, Hu Z, Simpson K, Pachter L, Durinck S, Wang N, Parvin B, Fontenay G, Speed T, Garbe J, Stampfer M, Bayandorian H, Dorton S, Clark TA, Schweitzer A, Wyrobek A, Feiler H, Spellman P, Conboy J, Gray JW. Exon-level microarray analyses identify alternative splicing programs in breast cancer. *Mol Cancer Res* 2010, 8:961-74.
12. Bemmo A, Benovoy D, Kwan T, Gaffney DJ, Jensen RV, Majewski J. Gene expression and isoform variation analysis using Affymetrix Exon Arrays. *BMC Genomics* 2008, 9:529.
13. Turro E, Lewin A, Rose A, Dallman MJ, Richardson S. MMBGX: a method for estimating expression at the isoform level and detecting differential splicing using whole-transcript Affymetrix arrays. *Nucleic Acids Res* 2010, 38(1):e4.
14. Bryant DW Jr, Priest HD, Mockler TC. Detection and quantification of alternative splicing variants using RNA-seq. *Methods Mol Biol.* 2012, 883:97-110.
15. Sultan M, Schulz MH, Richard H, Magen A, Klingenhoff A, Scherf M, Seifert M, Borodina T, Soldatov A, Parkhomchuk D, Schmidt D, O'Keefe S, Haas S, Vingron M, Lehrach H, Yaspo ML. A global view of gene activity and alternative splicing by deep sequencing of the human transcriptome. *Science* 2008, 321(5891):956-60.

-
16. Verdugo RA, Medrano JF. Comparison of gene coverage of mouse oligonucleotide microarray platforms. *BMC Genomics* 2006, 21:7:58.
 17. Acharya KK, Chandrashekar DS, Chitturi N, Shah H, Malhotra V, Sreelakshmi KS, Deepti H, Bajpai A, Davuluri S, Bora P, Rao L. A novel tissue-specific meta-analysis approach for gene expression predictions, initiated with a mammalian gene expression testis database. *BMC Genomics* 2010, 11:467.
 18. Moll AG, Lindenmeyer MT, Kretzler M, Nelson PJ, Zimmer R, Cohen CD. Transcript-specific expression profiles derived from sequence-based analysis of standard microarrays. *PLoS One* 2009, 4(3):e4702.
 19. Salisbury J, Hutchison KW, Wigglesworth K, Eppig JJ, Graber JH. Probe-level analysis of expression microarrays characterizes isoform-specific degradation during mouse oocyte maturation. *PLoS One* 2009, 4(10):e7479.
 20. Lee JC, Stiles D, Lu J, Cam MC. A detailed transcript-level probe annotation reveals alternative splicing based microarray platform differences. *BMC Genomics* 2007, 8:284.
 21. Yu H, Wang F, Tu K, Xie L, Li YY, Li YX. Transcript-level annotation of Affymetrix probesets improves the interpretation of gene expression data. *BMC Bioinformatics* 2007, 8:194.
 22. Lu J, Lee JC, Salit ML, Cam MC. Transcript-based redefinition of grouped oligonucleotide probe sets using AceView: high-resolution annotation for microarrays. *BMC Bioinformatics* 2007, 8:108.
 23. Hu GK, Madore SJ, Moldover B, Jatcoe T, Balaban D, Thomas J, Wang Y. Predicting splice variant from DNA chip expression data. *Genome Res* 2001, 11(7):1237-45.
 24. Robinson MD, Speed TP. Differential splicing using whole-transcript microarrays. *BMC Bioinformatics* 2009, 10:156.
 25. Shi L, Jones WD, Jensen RV, Harris SC, Perkins RG, Goodsaid FM, Guo L, Croner LJ, Boysen C, Fang H, Qian F, Amur S, Bao W, Barbacioru CC, Bertholet V, Cao XM, Chu TM, Collins PJ, Fan XH, Frueh FW, Fuscoe JC, Guo X, Han J, Herman D, Hong H, Kawasaki ES, Li QZ, Luo Y, Ma Y, Mei N, Peterson RL, Puri RK, Shippey R, Su Z, Sun YA, Sun H, Thorn B, Turpaz Y, Wang C, Wang SJ, Warrington JA, Willey JC, Wu J, Xie Q, Zhang L, Zhang L, Zhong S, Wolfinger RD, Tong W. The balance of reproducibility, sensitivity, and specificity of lists of differentially expressed genes in microarray studies. *BMC Bioinformatics*. 2008 Aug 12;9 Suppl 9:S10.
 26. Draghici S, Khatri P, Eklund AC, Szallasi Z. Reliability and reproducibility issues in DNA microarray measurements. *Trends Genet.* 2006 Feb;22(2):101-9.
 27. Harbig J, Sprinkle R, Enkemann SA. A sequence-based identification of the genes detected by probesets on the Affymetrix U133 plus 2.0 array. *Nucleic Acids Res* 2005, 33(3):e31.
 28. Mecham BH, Wetmore DZ, Szallasi Z, Sadovsky Y, Kohane I, Mariani TJ. Increased measurement accuracy for sequence-verified microarray probes. *Physiol Genomics* 2004, 18(3):308-15.
 29. Dai M, Wang P, Jakupovic E, Watson SJ, Meng F. Web-based GeneChip analysis system for large-scale collaborative projects. *Bioinformatics*. 2007 Aug 15;23(16):2185-7.
 30. Human RefSeq transcript sequences:
ftp://ftp.ncbi.nlm.nih.gov/refseq/H_sapiens/mRNA_Prot/human.rna.fna.gz
 31. Mouse RefSeq transcript sequences:
ftp://ftp.ncbi.nlm.nih.gov/refseq/M_musculus/mRNA_Prot/mouse.rna.fna.gz
 32. Rat RefSeq transcript sequences:
ftp://ftp.ncbi.nlm.nih.gov/refseq/R_norvegicus/mRNA_Prot/rat.rna.fna.gz

-
33. Affymetrix support: <http://www.affymetrix.com/support/technical/byproduct.affx?cat=exparrays>
 34. GEO FTP site: <ftp://ftp.ncbi.nlm.nih.gov/geo/>
 35. GEO HTTP site: <http://www.ncbi.nlm.nih.gov/geo/>
 36. NCBI BLAST: <http://mirrors.vbi.vt.edu/mirrors/ftp.ncbi.nih.gov/blast/executables/release/2.2.25/>
 37. Bioconductor makecdf package:
<http://www.bioconductor.org/packages/2.2/bioc/html/makecdfenv.html>
 38. Bioconductor affy package: <http://www.bioconductor.org/packages/2.2/bioc/html/affy.html>
 39. Bioconductor simple affy package:
<http://www.bioconductor.org/packages/2.12/bioc/html/simpleaffy.html>
 40. Bioconductor makecdf manual:
<http://www.bioconductor.org/packages/release/bioc/manuals/makecdfenv/man/makecdfenv.pdf>,
 41. Bioconductor affy manual:
<http://www.bioconductor.org/packages/release/bioc/manuals/affy/man/affy.pdf>
 42. Bioconductor simpleaffy manual:
<http://www.bioconductor.org/packages/release/bioc/manuals/simpleaffy/man/simpleaffy.pdf>
 43. Statistics Ttest package: <http://search.cpan.org/~yunfang/Statistics-TTest-1.1.0/TTest.pm>
 44. Statistics distribution package: <http://search.cpan.org/~mikek/Statistics-Distributions-1.02/Distributions.pm>
 45. NCBI database: <http://www.ncbi.nlm.nih.gov/>
 46. Magrane M, Consortium U. UniProt Knowledgebase: a hub of integrated protein data. Database (Oxford) 2011, 2011:bar009.
 47. Piva F, Giulietti M, Burini AB, Principato G. SpliceAid 2: a database of human splicing factors expression data and RNA target motifs. Hum Mutat 2012, 33(1):81-5.
 48. Bengert P, Dandekar T. A software tool-box for analysis of regulatory RNA elements. Nucleic Acids Res. 2003, 31(13):3441-5.
 49. Fairbrother WG, Yeo GW, Yeh R, Goldstein P, Mawson M, Sharp PA, Burge CB. RESCUE-ESE identifies candidate exonic splicing enhancers in vertebrate exons. Nucleic Acids Res. 2004, 32(Web Server issue):W187-90.
 50. ESRsearch: <http://esrsearch.tau.ac.il/>
 51. ACESCAN2: <http://genes.mit.edu/acescan2/index.html>
 52. Crooks GE, Hon G, Chandonia JM, Brenner SE. WebLogo: a sequence logo generator. Genome Res. 2004, 14(6):1188-90.
 53. Huang HD, Horng JT, Lee CC, Liu BJ. ProSplicer: a database of putative alternative splicing information derived from protein, mRNA and expressed sequence tag sequence data. Genome Biol. 2003, 4(4):R29
 54. Thierry-Mieg D, Thierry-Mieg J: AceView. a comprehensive cDNA-supported gene and transcripts annotation. Genome Biol. 2006, 7(Suppl 1):S12.
 55. de la Grange P, Dutertre M, Martin N, Auboeuf D. FAST DB: a website resource for the study of the expression regulation of human gene products. Nucleic Acids Res. 2005 Jul 28;33(13):4276-84. <http://www.fast-db.com/fastdb2/frame.html>

-
56. Leipzig J, Pevzner P, Heber S. The Alternative Splicing Gallery (ASG): bridging the gap between genome and transcriptome. *Nucleic Acids Res.* 2004, 32(13):3977-83.
 57. Kahn AB, Ryan MC, Liu H, Zeeberg BR, Jamison DC, Weinstein JN. SpliceMiner: a high-throughput database implementation of the NCBI Evidence Viewer for microarray splice variant analysis. *BMC Bioinformatics.* 2007, 8:75.
 58. ASTRA: http://www.cbrc.jp/cbrc/news/astra_eng.html
 59. PubMed: <http://www.ncbi.nlm.nih.gov/pubmed/>
 60. Startbioinfo portal: <http://www.startbioinfo.com/>
 61. Zhang Z, Pal S, Bi Y, Tchou J, Davuluri RV. Isoform level expression profiles provide better cancer signatures than gene level expression profiles. *Genome Med.* 2013 Apr 17;5(4):33.
 62. Wang Z, Gerstein M, Snyder M. RNA-Seq: a revolutionary tool for transcriptomics. *Nat Rev Genet.* 2009 Jan;10(1):57-63.
 63. Consiglio A, Carella M, De Caro G, Delle Foglie G, Giovannelli C, Grillo G, Ianigro M, Licciulli F, Palumbo O, Piepoli A, Ranieri E, Liuni S. BEAT: Bioinformatics Exon Array Tool to store, analyze and visualize Affymetrix GeneChip Human Exon Array data from disease experiments. *BMC Bioinformatics.* 2012 Mar 28;13 Suppl 4:S21.
 64. Robinson TJ, Dinan MA, Dewhirst M, Garcia-Blanco MA, Pearson JL. SplicerAV: a tool for mining microarray expression data for changes in RNA processing. *BMC Bioinformatics.* 2010 Feb 25;11:108.
 65. Gellert P, Jenniches K, Braun T, Uchida S. C-It: a knowledge database for tissue-enriched genes. *Bioinformatics.* 2010 Sep 15;26(18):2328-33.
 66. Hu GK, Madore SJ, Moldover B, Jatko T, Balaban D, Thomas J, Wang Y. Predicting splice variant from DNA chip expression data. *Genome Res.* 2001 Jul;11(7):1237-45.
 67. Risueño A, Fontanillo C, Dinger ME, De Las Rivas J. GATEExplorer: genomic and transcriptomic explorer; mapping expression probes to gene loci, transcripts, exons and ncRNAs. *BMC Bioinformatics.* 2010 Apr 29;11:221.
 68. Kim N, Alekseyenko AV, Roy M, Lee C. The ASAP II database: analysis and comparative genomics of alternative splicing in 15 animal species. *Nucleic Acids Res.* 2007 Jan;35(Database issue):D93-8.
 69. Nurdinov RN, Neverov AD, Mal'ko DB, Kosmodem'ianskiĭ IA, Ermakova EO, Ramenskiĭ VE, Mironov AA, Gel'fand MS. [EDAS, databases of alternatively spliced human genes]. *Biofizika.* 2006 Jul-Aug;51(4):589-92.
 70. Skrabanek L, Campagne F. TissueInfo: high-throughput identification of tissue expression profiles and specificity. *Nucleic Acids Res.* 2001 Nov 1;29(21):E102-2.
 71. Griffith M, Griffith OL, Mwenifumbo J, Goya R, Morrissy AS, Morin RD, Corbett R, Tang MJ, Hou YC, Pugh TJ, Robertson G, Chittaranjan S, Ally A, Asano JK, Chan SY, Li HI, McDonald H, Teague K, Zhao Y, Zeng T, Delaney A, Hirst M, Morin GB, Jones SJ, Tai IT, Marra MA. Alternative expression analysis by RNA sequencing. *Nat Methods.* 2010 Oct;7(10):843-7.
 72. Alternative splicing resource comparison: <http://www.startbioinfo.com/AltSpliceCompare/>

Bibliography for chapter 5

1. Chitturi N, Balagannavar G, Chandrashekar DS, Abinaya S, Srini VS, Acharya KK. TIPMaP: a web server to establish transcript isoform profiles from reliable microarray probes. *BMC Genomics*. 2013 Dec 27;14:922.
2. Acharya KK, Chandrashekar DS, Chitturi N, Shah H, Malhotra V, Sreelakshmi KS, Deepti H, Bajpai A, Davuluri S, Bora P, Rao L. A novel tissue-specific meta-analysis approach for gene expression predictions, initiated with a mammalian gene expression testis database. *BMC Genomics* 2010, 11:467.
3. NOA gene expression data source (E-TABM-234): <http://www.ebi.ac.uk/arrayexpress/experiments/E-TABM-234>
4. Parkinson H, Kapushesky M, Kolesnikov N, Rustici G, Shojatalab M, Abeygunawardena N, Berube H, Dylag M, Emam I, Farne A, Holloway E, Lukk M, Malone J, Mani R, Pilicheva E, Rayner TF, Rezwan F, Sharma A, Williams E, Bradley XZ, Adamusiak T, Brandizi M, Burdett T, Coulson R, Krestyaninova M, Kurnosov P, Maguire E, Neogi SG, Rocca-Serra P, Sansone SA, Sklyar N, Zhao M, Sarkans U, Brazma A. ArrayExpress update--from an archive of functional genomics experiments to the atlas of gene expression. *Nucleic Acids Res*. 2009, 37(Database issue):D868-872.
5. Johnsen SG. Testicular biopsy score count--a method for registration of spermatogenesis in human testes: normal values and results in 335 hypogonadal males. *Hormones*. 1970;1(1):2-25.
6. Barrett T, Troup DB, Wilhite SE, Ledoux P, Evangelista C, Kim IF, Tomashevsky M, Marshall KA, Phillippy KH, Sherman PM, Muetter RN, Holko M, Ayanbule O, Yefanov A, Soboleva A. NCBI GEO: archive for functional genomics data sets--10 years on. *Nucleic Acids Res*. 2011, 39(Database issue):D1005-1010.
7. Korkola JE, Houldsworth J, Chadalavada RS, Olshen AB, Dobrzynski D, Reuter VE, Bosl GJ, Chaganti RS. Down-regulation of stem cell genes, including those in a 200-kb gene cluster at 12p13.31, is associated with in vivo differentiation of human male germ cell tumors. *Cancer Res*. 2006 Jan 15;66(2):820-7.
8. Su AI, Wiltshire T, Batalov S, Lapp H, Ching KA, Block D, Zhang J, Soden R, Hayakawa M, Kreiman G, Cooke MP, Walker JR, Hogenesch JB. A gene atlas of the mouse and human protein-encoding transcriptomes. *Proc Natl Acad Sci U S A*. 2004 Apr 20;101(16):6062-7.
9. Gashaw I, Grümmer R, Klein-Hitpass L, Dushaj O, Bergmann M, Brehm R, Grobholz R, Kliesch S, Neuvians TP, Schmid KW, von Ostau C, Winterhager E. Gene signatures of testicular seminoma with emphasis on expression of ets variant gene 4. *Cell Mol Life Sci*. 2005 Oct;62(19-20):2359-68.
10. Normal testis gene expression data source (GSE7307): <http://www.ncbi.nlm.nih.gov/geo/query/acc.cgi?acc=GSE7307>
11. Roth RB, Hevezi P, Lee J, Willhite D, Lechner SM, Foster AC, Zlotnik A. Gene expression analyses reveal molecular relationships among 20 regions of the human CNS. *Neurogenetics*. 2006 May;7(2):67-80.
12. Huang DW, Sherman BT, Lempicki RA. Systematic and integrative analysis of large gene lists using DAVID Bioinformatics Resources. *Nature Protoc*. 2009;4(1):44-57, Huang DW, Sherman BT, Lempicki RA. Bioinformatics enrichment tools: paths toward the comprehensive functional analysis of large gene lists. *Nucleic Acids Res*. 2009;37(1):1-13.
13. PubMed: <http://www.ncbi.nlm.nih.gov/pubmed/>
14. RiboPure kit: <http://www.lifetechnologies.com/order/catalog/product/AM1924>
15. RNA isolation procedure: https://tools.lifetechnologies.com/content/sfs/manuals/fm_1924.pdf

-
16. Verso cDNA kit: <http://www.thermoscientificbio.com/reverse-transcription-rtqpcr-verso-cdna-synthesis-kit/>
 17. cDNA synthesis procedure: <http://www.thermoscientificbio.com/uploadedFiles/Resources/prodinfo-AB-1453-B-Verso-cDNA-Synthesis-Kit.pdf>
 18. Ye J, Coulouris G, Zaretskaya I, Cutcutache I, Rozen S, Madden TL. Primer-BLAST: a tool to design target-specific primers for polymerase chain reaction. *BMC Bioinformatics*. 2012 Jun 18;13:134.
 19. OligoAnalyzer: <https://eu.idtdna.com/calc/analyzer>
 20. DyNAzyme II DNA Polymerase kit: <http://www.thermoscientificbio.com/pcr-enzymes-and-master-mixes/dynazyme-I-and-II-polymerases/>
 21. PCR procedure: <http://www.thermoscientificbio.com/uploadedFiles/Resources/tech-manual-f-501-dynazyme-ii-dna-polymerase.pdf>
 22. Rondanino C, Ouchchane L, Chauffour C, Marceau G, Déchelotte P, Sion B, Pons-Rejraji H, Janny L, Volle DH, Lobaccaro JM, Brugnon F. Levels of liver X receptors in testicular biopsies of patients with azoospermia. *Fertil Steril*. 2014 Aug;102(2):361-371.e5.
 23. Malcher A, Rozwadowska N, Stokowy T, Kolanowski T, Jedrzejczak P, Zietkowiak W, Kurpisz M. Potential biomarkers of nonobstructive azoospermia identified in microarray gene expression analysis. *Fertil Steril*. 2013 Dec;100(6):1686-94.e1-7.
 24. Dorosh A, Tepla O, Zatecka E, Ded L, Koci K, Peknicova J. Expression analysis of MND1/GAJ, SPATA22, GAPDHS and ACR genes in testicular biopsies from non-obstructive azoospermia (NOA) patients. *Reprod Biol Endocrinol*. 2013 May 15;11:42.
 25. Rolland AD, Lavigne R, Dauly C, Calvel P, Kervarrec C, Freour T, Evrard B, Rioux-Leclercq N, Auger J, Pineau C. Identification of genital tract markers in the human seminal plasma using an integrative genomics approach. *Hum Reprod*. 2013 Jan;28(1):199-209.
 26. O'Bryan MK, Grealy A, Stahl PJ, Schlegel PN, McLachlan RI, Jamsai D. Genetic variants in the ETV5 gene in fertile and infertile men with nonobstructive azoospermia associated with Sertoli cell-only syndrome. *Fertil Steril*. 2012 Oct;98(4):827-35.e1-3.
 27. Laurentino S, Gonçalves J, Cavaco JE, Oliveira PF, Alves MG, de Sousa M, Barros A, Socorro S. Apoptosis-inhibitor Aven is downregulated in defective spermatogenesis and a novel estrogen target gene in mammalian testis. *Fertil Steril*. 2011 Sep;96(3):745-50.
 28. Aslani F, Modarresi MH, Soltanghoraee H, Akhondi MM, Shabani A, Lakpour N, Sadeghi MR. Seminal molecular markers as a non-invasive diagnostic tool for the evaluation of spermatogenesis in non-obstructive azoospermia. *Syst Biol Reprod Med*. 2011 Aug;57(4):190-6.
 29. Abdou AG, Hammam MA, Farag AG, Farouk S, Fawzy M. Immunohistochemical expression of cyclin A in testicular biopsies of fertile and infertile men: correlation with the morphometry of seminiferous tubules. *Andrologia*. 2011 Feb;43(1):57-64.
 30. Ramasamy R, Ridgeway A, Lipshultz LI, Lamb DJ. Integrative DNA methylation and gene expression analysis identifies discoidin domain receptor 1 association with idiopathic nonobstructive azoospermia. *Fertil Steril*. 2014 Oct;102(4):968-973.e3.
 31. Malcher A, Rozwadowska N, Stokowy T, Jedrzejczak P, Zietkowiak W, Kurpisz M. The gene expression analysis of paracrine/autocrine factors in patients with spermatogenetic failure compared with normal spermatogenesis. *Am J Reprod Immunol*. 2013 Dec;70(6):522-8.

Bibliography for chapter 6

1. Jarvi K, Lo K, Fischer A, Grantmyre J, Zini A, Chow V, Mak V. CUA Guideline: The workup of azoospermic males. *Can Urol Assoc J*. 2010 Jun;4(3):163-7.
2. Oates R. Evaluation of the azoospermic male. *Asian J Androl*. 2012 Jan;14(1):82-7.
3. Gallego A, Rogel R, Luján S, Plaza B, Delgado F, Boronat F. AZF gene microdeletions: Case series and literature review. *Actas Urol Esp*. 2014 Jun 18. pii: S0210-4806(14)00146-6.
4. Alhalabi M, Kenj M, Monem F, Mahayri Z, Abou Alchamat G, Madania A. High prevalence of genetic abnormalities in Middle Eastern patients with idiopathic non-obstructive azoospermia. *J Assist Reprod Genet*. 2013 Jun;30(6):799-805.
5. Zhang Y, He XJ, Song B, Ye L, Xie XS, Ruan J, Zhou FS, Zuo XB, Cao YX, Du WD. Association of single nucleotide polymorphisms in the USF1, GTF2A1L and OR2W3 genes with non-obstructive azoospermia in the Chinese population. *J Assist Reprod Genet*. 2014 Nov 6.
6. Abu-Halima M, Hammadeh M, Backes C, Fischer U, Leidinger P, Lubbad AM, Keller A, Meese E. Panel of five microRNAs as potential biomarkers for the diagnosis and assessment of male infertility. *Fertil Steril*. 2014 Oct;102(4):989-997.e1.
7. Tewes AC, Ledig S, Tüttelmann F, Kliesch S, Wieacker P. DMRT1 mutations are rarely associated with male infertility. *Fertil Steril*. 2014 Sep;102(3):816-820.e3., Hojati Z, Heidari S, Motovali-Bashi M. Exon 10 CFTR gene mutation in male infertility. *Iran J Reprod Med*. 2012 Jul;10(4):315-20.
8. Lin YH, Lin YM, Teng YN, Hsieh TY, Lin YS, Kuo PL. Identification of ten novel genes involved in human spermatogenesis by microarray analysis of testicular tissue. *Fertil Steril*. 2006 Dec;86(6):1650-8.
9. Gatta V, Raicu F, Ferlin A, Antonucci I, Scioletti AP, Garolla A, Palka G, Foresta C, Stuppia L. Testis transcriptome analysis in male infertility: new insight on the pathogenesis of oligo-azoospermia in cases with and without AZFc microdeletion. *BMC Genomics*. 2010 Jun 24;11:401.
10. Spiess AN, Feig C, Schulze W, Chalmel F, Cappallo-Obermann H, Primig M, Kirchhoff C. Cross-platform gene expression signature of human spermatogenic failure reveals inflammatory-like response. *Hum Reprod*. 2007 Nov;22(11):2936-46.
11. Feig C, Kirchhoff C, Ivell R, Naether O, Schulze W, Spiess AN. A new paradigm for profiling testicular gene expression during normal and disturbed human spermatogenesis. *Mol Hum Reprod*. 2007 Jan;13(1):33-43.
12. Ellis PJ, Furlong RA, Conner SJ, Kirkman-Brown J, Afnan M, Barratt C, Griffin DK, Affara NA. Coordinated transcriptional regulation patterns associated with infertility phenotypes in men. *J Med Genet*. 2007 Aug;44(8):498-508.
13. Malcher A, Rozwadowska N, Stokowy T, Kolanowski T, Jedrzejczak P, Zietkowiak W, Kurpisz M. Potential biomarkers of nonobstructive azoospermia identified in microarray gene expression analysis. *Fertil Steril*. 2013 Dec;100(6):1686-94.e1-7.
14. Malcher A, Rozwadowska N, Stokowy T, Jedrzejczak P, Zietkowiak W, Kurpisz M. The gene expression analysis of paracrine/autocrine factors in patients with spermatogenic failure compared with normal spermatogenesis. *Am J Reprod Immunol*. 2013 Dec;70(6):522-8.
15. Okada H, Tajima A, Shichiri K, Tanaka A, Tanaka K, Inoue I. Genome-wide expression of azoospermia testes demonstrates a specific profile and implicates ART3 in genetic susceptibility. *PLoS Genet*. 2008 Feb;4(2):e26.

16. Yang B, Yuan JL, Gao XK, Liu HL, Qin WJ, Shao C, Liu F, Kang FX. [Expressions of cadherin molecules CDH18 and PCDH17 in human azoospermic testes]. *Zhonghua Nan Ke Xue*. 2009 Dec;15(12):1081-4.
17. Yang B, Yuan JL, Gao XK, Wang H, Shao C, Liu HL, Chen BQ, Qin RL, Shao GX, Kang FX. [Expression of COX10 in human non-obstructive azoospermia testes]. *Zhonghua Nan Ke Xue*. 2009 Jul;15(7):599-603.
18. Drabovich AP, Dimitromanolakis A, Saraon P, Soosaipillai A, Batruch I, Mullen B, Jarvi K, Diamandis EP. Differential diagnosis of azoospermia with proteomic biomarkers ECM1 and TEX101 quantified in seminal plasma. *Sci Transl Med*. 2013 Nov 20;5(212):212ra160.
19. Batruch I, Smith CR, Mullen BJ, Grober E, Lo KC, Diamandis EP, Jarvi KA. Analysis of seminal plasma from patients with non-obstructive azoospermia and identification of candidate biomarkers of male infertility. *J Proteome Res*. 2012 Mar 2;11(3):1503-11.
20. Drabovich AP, Jarvi K, Diamandis EP. Verification of male infertility biomarkers in seminal plasma by multiplex selected reaction monitoring assay. *Mol Cell Proteomics*. 2011 Dec;10(12):M110.004127.
21. Légaré C, Cloutier F, Makosso-Kallyth S, Laflamme N, Jarvi K, Tremblay RR, Sullivan R. Cysteine-rich secretory protein 1 in seminal plasma: potential biomarker for the distinction between obstructive and nonobstructive azoospermia. *Fertil Steril*. 2013 Nov;100(5):1253-60.
22. Dorosh A, Tepla O, Zatecka E, Ded L, Koci K, Peknicova J. Expression analysis of MND1/GAJ, SPATA22, GAPDHS and ACR genes in testicular biopsies from non-obstructive azoospermia (NOA) patients. *Reprod Biol Endocrinol*. 2013 May 15;11:42.
23. Pan Q, Shai O, Lee LJ, Frey BJ, Blencowe BJ. Deep surveying of alternative splicing complexity in the human transcriptome by high-throughput sequencing. *Nat Genet* 2008, 40:1413-1417.
24. He C, Zuo Z, Chen H, Zhang L, Zhou F, Cheng H, Zhou R. Genome-wide detection of testis- and testicular cancer-specific alternative splicing. *Carcinogenesis* 2007, 28:2484-2490.
25. Blair CA, Zi X. Potential molecular targeting of splice variants for cancer treatment. *Indian J Exp Biol* 2011, 49:836-9.
26. Garcia-Blanco MA, Baraniak AP, Lasda EL. Alternative splicing in disease and therapy. *Nat Biotechnol* 2004, 22:535-46.
27. Venables JP. Aberrant and alternative splicing in cancer. *Cancer Res* 2004, 64:7647-54.
28. Garcia-Blanco MA: Alternative splicing: therapeutic target and tool. *Prog Mol Subcell Biol* 2006, 44:47-64.
29. Gaur RK: RNA interference: a potential therapeutic tool for silencing splice isoforms linked to human diseases. *Biotechniques* 2006, (Suppl):15-22.
30. Wang Z, Gerstein M, Snyder M. RNA-Seq: a revolutionary tool for transcriptomics. *Nat Rev Genet*. 2009 Jan;10(1):57-63.
31. Barrett T, Troup DB, Wilhite SE, Ledoux P, Evangelista C, Kim IF, Tomashevsky M, Marshall KA, Phillippy KH, Sherman PM, Muetter RN, Holko M, Ayanbule O, Yefanov A, Soboleva A. NCBI GEO: archive for functional genomics data sets--10 years on. *Nucleic Acids Res*. 2011, 39(Database issue):D1005-1010.
32. Chu P, Liu H, Yang Q, Wang Y, Yan G, Guan R. An RNA-seq transcriptome analysis of floral buds of an interspecific Brassica hybrid between *B. carinata* and *B. napus*. *Plant Reprod*. 2014 Nov 15.
33. Sharma D, Kim MS, D'Mello SR. Transcriptome profiling of expression changes during neuronal death by RNA-Seq. *Exp Biol Med (Maywood)*. 2014 Sep 25. pii: 1535370214551688.

34. Han SS, Kim WJ, Hong Y, Hong SH, Lee SJ, Ryu DR, Lee W, Cho YH, Lee S, Ryu YJ, Won JY, Rhee H, Park JH, Jang SJ, Lee JS, Choi CM, Lee JC, Lee SD, Oh YM. RNA sequencing identifies novel markers of non-small cell lung cancer. *Lung Cancer*. 2014 Jun;84(3):229-35.
35. Wang L, Zhan C, Zhang Y, Ma J, Xi J, Jiang W, Shi Y, Wang Q. Quantifying the expression of tumor marker genes in lung squamous cell cancer with RNA sequencing. *J Thorac Dis*. 2014 Oct;6(10):1380-7.
36. Lindskog C, Fagerberg L, Hallström B, Edlund K, Hellwig B, Rahnenführer J, Kampf C, Uhlén M, Pontén F, Micke P. The lung-specific proteome defined by integration of transcriptomics and antibody-based profiling. *FASEB J*. 2014 Aug 28.
37. González-Porta M, Frankish A, Rung J, Harrow J, Brazma A. Transcriptome analysis of human tissues and cell lines reveals one dominant transcript per gene. *Genome Biol*. 2013 Jul 1;14(7):R70.
38. RiboPure kit: <http://www.lifetechnologies.com/order/catalog/product/AM1924>
39. RNA isolation procedure: https://tools.lifetechnologies.com/content/sfs/manuals/fm_1924.pdf
40. Bioanalyzer: http://www.genomics.agilent.com/en/product.jsp?cid=AG-PT-106&_requestid=94544
41. Illumina TruSeq RNA kit: http://support.illumina.com/content/dam/illumina-support/documents/documentation/chemistry_documentation/samplepreps_truseq/truseqrna/truseq-rna-sample-prep-v2-guide-15026495-f.pdf
42. Brawand D, Soumillon M, Necsulea A, Julien P, Csárdi G, Harrigan P, Weier M, Liechti A, Aximu-Petri A, Kircher M, Albert FW, Zeller U, Khaitovich P, Grützner F, Bergmann S, Nielsen R, Pääbo S, Kaessmann H. The evolution of gene expression levels in mammalian organs. *Nature*. 2011 Oct 19;478(7369):343-8.
43. SRA: <http://www.ncbi.nlm.nih.gov/sra/>
44. FASTQC: <http://www.bioinformatics.babraham.ac.uk/projects/fastqc/>
45. UCSC browser: <https://genome.ucsc.edu/>
46. Bowtie: <http://bowtie-bio.sourceforge.net/index.shtml>
47. TopHat: <http://ccb.jhu.edu/software/tophat/index.shtml>
48. Cufflinks: <http://cufflinks.cbcb.umd.edu/>
49. UCSC human genome 19: <http://hgdownload.soe.ucsc.edu/goldenPath/hg19/bigZips/hg19.2bit>
50. Huang DW, Sherman BT, Lempicki RA. Systematic and integrative analysis of large gene lists using DAVID Bioinformatics Resources. *Nature Protoc*. 2009;4(1):44-57, Huang DW, Sherman BT, Lempicki RA. Bioinformatics enrichment tools: paths toward the comprehensive functional analysis of large gene lists. *Nucleic Acids Res*. 2009;37(1):1-13.
51. R package, RColorBrewer: <http://cran.r-project.org/web/packages/RColorBrewer/index.html>
52. R package, gplots: <http://cran.r-project.org/web/packages/gplots/index.html>
53. Amigo Gene Ontology server: Genes associated with spermatogenesis: <http://amigo.geneontology.org/amigo/search/bioentity?q=spermatogenesis>
54. PubMed: <http://www.ncbi.nlm.nih.gov/pubmed/>
55. Chitturi N, Balagannavar G, Chandrashekar DS, Abinaya S, Srini VS, Acharya KK. TIPMaP: a web server to establish transcript isoform profiles from reliable microarray probes. *BMC Genomics*. 2013 Dec 27;14:922.

-
56. Amigo Gene Ontology server: <http://amigo.geneontology.org/amigo>
 57. KEGG pathway database: <http://www.genome.jp/kegg/>
 58. Parmentier G, Bastian FB, Robinson-Rechavi M. Homolonto: generating homology relationships by pairwise alignment of ontologies and application to vertebrate anatomy. *Bioinformatics*. 2010 Jul 15;26(14):1766-71.
 59. Pansa A, Sirchia SM, Melis S, Giacchetta D, Castiglioni M, Colapietro P, Fiori S, Falcone R, Paganini L, Bonaparte E, Colpi G, Miozzo M, Tabano S. ESX1 mRNA expression in seminal fluid is an indicator of residual spermatogenesis in non-obstructive azoospermic men. *Hum Reprod*. 2014 Dec;29(12):2620-7.
 60. Wang C, Gong B, Bushel PR, Thierry-Mieg J, Thierry-Mieg D, Xu J, Fang H, Hong H, Shen J, Su Z, Meehan J, Li X, Yang L, Li H, Łabaj PP, Kreil DP, Megherbi D, Gaj S, Caiment F, van Delft J, Kleinjans J, Scherer A, Devanarayan V, Wang J, Yang Y, Qian HR, Lancashire LJ, Bessarabova M, Nikolsky Y, Furlanello C, Chierici M, Albanese D, Jurman G, Riccadonna S, Filosi M, Visintainer R, Zhang KK, Li J, Hsieh JH, Svoboda DL, Fuscoe JC, Deng Y, Shi L, Paules RS, Auerbach SS, Tong W. The concordance between RNA-seq and microarray data depends on chemical treatment and transcript abundance. *Nat Biotechnol*. 2014 Sep;32(9):926-32.
 61. Zhao S, Fung-Leung WP, Bittner A, Ngo K, Liu X. Comparison of RNA-Seq and microarray in transcriptome profiling of activated T cells. *PLoS One*. 2014 Jan 16;9(1):e78644.
 62. Hicks C, Miele L, Koganti T, Young-Gaylor L, Rogers D, Vijayakumar V, Megason G. Analysis of Patterns of Gene Expression Variation within and between Ethnic Populations in Pediatric B-ALL. *Cancer Inform*. 2013 Aug 27;12:155-73.

Bibliography for chapter 7

1. Acharya KK, Chandrashekar DS, Chitturi N, Shah H, Malhotra V, Sreelakshmi KS, Deepti H, Bajpai A, Davuluri S, Bora P, Rao L. A novel tissue-specific meta-analysis approach for gene expression predictions, initiated with a mammalian gene expression testis database. *BMC Genomics* 2010, 11:467.
2. Chitturi N, Balagannavar G, Chandrashekar DS, Abinaya S, Srini VS, Acharya KK. TIPMaP: a web server to establish transcript isoform profiles from reliable microarray probes. *BMC Genomics*. 2013 Dec 27;14:922.