

REFERENCES

1. Ahmadian, A., Gharizadeh, B., Gustafsson, A.C., Sterky, F., Nyren, P., Uhlen, M. and Lundeberg, J. (2000) Single-nucleotide polymorphism analysis by pyrosequencing. *Anal. Biochem.*, 280, 103-10.
2. Akey, J.M., Sosnoski, D., Parra, E., Dios, S., Hiester, K., Su, B., Bonilla, C., Jin, L. and Shriver, M.D. (2001) Melting curve analysis of SNPs (McSNP): a gel-free and inexpensive approach for SNP genotyping. *Biotechniques*, 30, 358-62, 364, 366-7.
3. Alderborn, A., Kristofferson, A. and Hammerling, U. (2000) Determination of single-nucleotide polymorphisms by realtime pyrophosphate DNA sequencing. *Genome Res.*, 10, 1249-58.
4. Alonso-Blanco, C. and Koornneef, M. (2000) Naturally occurring variation in Arabidopsis: An underexploited resource for plant genetics. *Trends Plant. Sci.*, 5, 22-9.
5. Alsmadi, O.A., Bornarth, C.J., Song, W., Wisniewski, M., Du, J., Brockman, J.P., Faruqi, A.F., Hosono, S., Sun, Z., Du, Y., Wu, X., Egholm, M., Abarzua, P., Lasken, R.S. and Driscoll, M.D. (2003) High accuracy genotyping directly from genomic DNA using a rolling circle amplification based assay. *BMC Genomics.*, 4, 21.
6. An, P., Nelson, G.W., Wang, L., Donfield, S., Goedert, J.J., Phair, J., Vlahov, D., Buchbinder, S., Farrar, W.L., Modi, W., O'Brien, S.J. and Winkler, C.A. (2002) Modulating influence on HIV/AIDS by interacting RANTES gene variants. *Proc. Natl Acad. Sci. USA*, 99, 10002-7.
7. Antson, D.O., Isaksson, A., Landegren, U. and Nilsson, M. (2000) PCRgenerated padlock probes detect single nucleotide variation in genomic DNA. *Nucleic Acids Res.*, 28, e58.
8. Baner, J., Isaksson, A., Waldenstrom, E., Jarvius, J., Landegren, U. and Nilsson, M. (2003) Parallel gene analysis with allele-specific padlock probes and tag microarrays. *Nucleic Acids Res.*, 31, e103.
9. Baner, J., Nilsson, M., Mendel-Hartvig, M. and Landegren, U. (1998) Signal amplification of padlock probes by rolling circle replication. *Nucleic Acids Res.*, 26, 5073-8.
10. Barany F. (1991) Genetic disease detection and DNA amplification using cloned thermostable ligase. *Proc. Natl Acad. Sci. USA*, 88, 189-93.

11. Barany, F., Lubin, M. and Belgrader, P. (2001) Detection of nucleic sequence differences using coupled ligase detection and polymerase chain reaction. United States Patent 6,268,148.
12. Beaudet, L., Bedard, J., Breton, B., Mercuri, R. J. and Budarf, M. L. (2001) Homogeneous assays for single-nucleotide polymorphism typing using AlphaScreen. *Genome Res.*, 11, 600–8.
13. Borodina, T.A., Lehrach, H. and Soldatov, A.V. (2003) Ligation-based synthesis of oligonucleotides with block structure. *Anal. Biochem.*, 318, 309-13.
14. Borodina, T.A., Lehrach, H. and Soldatov, A.V. (2003a) DNA purification on homemade silica spin-columns. *Anal. Biochem.*, 321, 135-7.
15. Borodina, T.A., Lehrach, H. and Soldatov, A.V. (2004) Ligation detection reaction-TaqMan procedure for single nucleotide polymorphism detection on genomic DNA. *Anal. Biochem.*, 333, 309-19.
16. Braun, A., Little, D.P. and Koster, H. (1997) Detecting CFTR gene mutations by using primer oligo base extension and mass spectrometry. *Clin. Chem.*, 43, 1151-8.
17. Breen, G. (2002) Novel and alternate SNP and genetic technologies. *Psychiatr. Genet.*, 12, 83-8.
18. Brookes, A.J. (1999) The essence of SNP. *Gene*, 234, 177-86.
19. Cai, H., White, P.S., Torney, D., Deshpande, A., Wang, Z., Keller, R.A., Marrone, B. and Nolan, J.P. (2000) Flow cytometry-based minisequencing: a new platform for high-throughput single-nucleotide polymorphism scoring. *Genomics*, 66, 135–43.
20. Chen, D.C., Saarela, J., Nuotio, I., Jokiahho, A., Peltonen, L. and Palotie, A. (2003) Comparison of GenFlex Tag array and Pyrosequencing in SNP genotyping. *J. Mol. Diagn.*, 5, 243-9.
21. Chen, J., Iannone, M.A., Li, M.S., Taylor, J.D., Rivers, P., Nelsen, A.J., Slentz-Kesler, K.A., Roses, A. and Weiner, M.P. (2000) A microsphere-based assay for multiplexed single nucleotide polymorphism analysis using single base chain extension. *Genome Res.*, 10, 549–57.
22. Chen, X. and Kwok, P.-Y. (1997) Templatedirected dye-terminator incorporation (TDI) assay: a homogeneous DNA diagnostic method based on fluorescence energy transfer. *Nucleic Acids Res.*, 25, 347–53.
23. Chen, X., Levine, L. and Kwok, P.-Y. (1999) Fluorescence polarization in homogeneous nucleic acid analysis. *Genome Res.*, 9, 492-8.

24. Chen, X., Livak, K. and Kwok, P.-Y. (1998) A homogeneous, ligase-mediated DNA diagnostic test. *Genome Res.*, 8, 549–56.
25. Chiang, K.P., Gerber, A.L., Sipe, J.C. and Cravatt, B.F. (2004) Reduced cellular expression and activity of the P129T mutant of human fatty acid amide hydrolase: evidence for a link between defects in the endocannabinoid system and problem drug use. *Hum. Mol. Genet.*, 13, 2113-9.
26. Cho, R.J., Mindrinos, M., Richards, D.R., Sapolsky, R.J., Anderson, M., Drenkard, E., Dewdney, J., Reuber, T.L., Stammers, M., Federspiel, N. et al. (1999) Genome-wide mapping with biallelic markers in *Arabidopsis thaliana*. *Nat. Genet.*, 23, 203-7.
27. Cohen, V., Panet-Raymond, V., Sabbaghian, N., Morin, I., Batist, G. and Rozen, R. (2003) Methylenetetrahydrofolate reductase polymorphism in advanced colorectal cancer: a novel genomic predictor of clinical response to fluoropyrimidine-based chemotherapy. *Clin. Cancer. Res.*, 9, 1611-5.
28. Consolandi, C., Frosini, A., Pera, C., Ferrara, G.B., Bordoni, R., Castiglioni, B., Rizzi, E., Mezzelani, A., Bernardi, L.R., De Bellis, G. and Battaglia, C. (2004) Polymorphism analysis within the HLA-A locus by universal oligonucleotide array. *Hum. Mutat.*, 24, 428-34.
29. Cronin, M.T., Fucini, F.V., Kim, S.M., Masino, R.S., Wepsi, R.M. and Miyada, C.G. (1996) Cystic fibrosis mutation detection by hybridisation to light generated DNA probe arrays. *Hum. Mutat.*, 7, 244-55.
30. Cronin, M.T., Pho, M. and Brennan, T.M. (1998) Applying an array hybridisation strategy to high throughput genotyping for drug metabolising enzymes. *ISSX Proc.*, 13, 19-20.
31. Crow, J.F. (1995) Spontaneous mutation as a risk factor. *Exp. Clin. Immunogenet.*, 12, 121-8.
32. Daniliuk, N.K., Iastrebov, S.I., Artamonova, T.P. and Popov, S.G. (1986) A simplified variant of the Maxam-Gilbert method for determining the primary structure of oligonucleotides and DNA fragments, *Bioorg. Khim. (rus)*, 12, 1185-1188.
33. Davignon, J., Gregg, R.E. and Sing, C.F. (1988) Apolipoprotein E polymorphism and atherosclerosis. *Arteriosclerosis*, 8, 1–21.
34. Dean, M., Carrington, M., Winkler, C., Huttley, G.A., Smith, M.W., Allikmets, R., Goedert, J.J., Buchbinder, S.P., Vittinghoff, E., Gomperts, E., Donfield, S. et al.

- (1996) Genetic resistance of HIV infection and progression to AIDS by deletion of the CKR5 structural gene. *Science*, 273, 1856–62.
35. Ehn, M., Nourizad, N., Bergstrom, K., Ahmadian, A., Nyren, P., Lundeberg, J. and Hober, S. (2004) Toward pyrosequencing on surface-attached genetic material by use of DNA-binding luciferase fusion proteins. *Anal. Biochem.*, 329, 11-20.
 36. Enattah, N.S., Sahi, T., Savilahti, E., Terwilliger, J.D., Peltonen, L. and Jarvela, I. (2002) Identification of a variant associated with adult-type hypolactasia. *Nat. Genet.*, 30, 233-7.
 37. Engelke, D.R., Krikos, A., Bruck, M.E. and Ginsburg, D. (1990) Purification of *Thermus aquaticus* DNA polymerase expressed in *Escherichia coli*. *Anal. Biochem.*, 191, 396-400.
 38. Eriksson, J., Gharizadeh, B., Nordstrom, T. and Nyren, P. (2004) Pyrosequencing trade mark technology at elevated temperature. *Electrophoresis*, 25, 20-7.
 39. Fan, J.B., Chen, X., Halushka, M.K., Berno, A., Huang, X., Ryder, T., Lipshutz, R.J., Lockhart, D.J. and Chakravarti, A. (2000) Parallel genotyping of human SNPs using generic high-density oligonucleotide tag arrays. *Genome Res.*, 10, 853–60.
 40. Faruqi, A.F., Hosono, S., Driscoll, M.D., Dean, F.B., Alsmadi, O., Bandaru, R., Kumar, G., Grimwade, B., Zong, Q., Sun, Z., Du, Y., Kingsmore, S., Knott, T. and Lasken, R.S. (2001) High-throughput genotyping of single nucleotide polymorphisms with rolling circle amplification. *BMC Genomics*, 2, 4.
 41. Favis, R., Day, J.P., Gerry, N.P., Phelan, C., Narod, S., and Barany, F. (2000) Universal DNA array detection of small insertions and deletions in BRCA1 and BRCA2. *Nat. Biotechnol.*, 18, 561-4.
 42. Fei, Z., Ono, T. and Smith, L.M. (1998) MALDI-TOF mass spectrometric typing of single nucleotide polymorphisms with mass-tagged ddNTPs. *Nucleic Acids Res.*, 26, 2827-8.
 43. Foster, M.W. and Sharp, R.R. (2002) Race, ethnicity, and genomics: social classifications as proxies of biological heterogeneity. *Genome Res.*, 12, 844-50.
 44. Fredman, D., Munns, G., Rios, D., Sjolholm, F., Siegfried, M., Lenhard, B., Lehvaslaiho, H. and Brookes, A. J. (2004) HGVbase: a curated resource describing human DNA variation and phenotype relationships. *Nucleic Acids Res.*, 32, D516-9.
 45. Frosst, P., Blom, H. J., Milos, R., Goyette, P., Sheppard, C. A., Matthews, R.G., Boers, G.J., den Heijer, M., Kluijtmans, L.A., van den Heuvel, L. P. and Rozen, R.

- (1995) A candidate genetic risk factor for vascular disease: a common mutation in methylenetetrahydrofolate reductase. *Nat. Genet.*, 10, 111-3.
46. Gardemann, A., Stricker, J., Humme, J., Nguyen, Q.D., Katz, N., Philipp, M., Tillmanns, H., Hehrlein, F.W. and Haberbosch, W. (1999) Angiotensinogen T174M and M235T gene polymorphisms are associated with the extent of coronary atherosclerosis. *Atherosclerosis*, 145, 309-14.
 47. Gaylord, B.S., Massie, M.R., Feinstein, S.C. and Bazan, G.C. (2005) SNP detection using peptide nucleic acid probes and conjugated polymers: Applications in neurodegenerative disease identification. *Proc Natl Acad Sci USA*, 102, 34-9.
 48. Germer, S. and Higuchi, R. (1999) Single-tube genotyping without oligonucleotide probes. *Genome Res.*, 9, 72-8.
 49. Gerry, N.P., Witowski, N.E., Day, J., Hammer, R.P., Barany, G. and Barany, F. (1999) Universal DNA microarray method for multiplex detection of low abundance point mutations. *J. Mol. Biol.*, 292, 251-62.
 50. Gharizadeh, B., Eriksson, J., Nourizad, N., Nordstrom, T. and Nyren, P. (2004) Improvements in Pyrosequencing technology by employing Sequenase polymerase. *Anal. Biochem.*, 330, 272-80.
 51. Ghiselli, G., Gregg, R.E., Zech, L.A., Schaefer, E.J. and Brewer, H.B. (1982) Phenotype study of apolipoprotein E isoforms in hyperlipoproteinaemic patients. *Lancet*, 2, 405-7.
 52. Griffin, T.J., Hall, J.G., Prudent, J.R. and Smith, L.M. (1999) Direct genetic analysis by matrix-assisted laser desorption/ionization mass spectrometry. *Proc. Natl Acad. Sci. USA*, 96, 6301-6.
 53. Griffin, T.J., Tang, W. and Smith, L.M. (1997) Genetic analysis by peptide nucleic acid affinity MALDI-TOF mass spectrometry. *Nat. Biotechnol.*, 15, 1368-72.
 54. Grossman, P.D., Bloch, W., Brinson, E., Chang, C.C., Eggerding, F.A., Fung, S., Iovannisci, D.M., Woo, S. and Winn-Deen, E.S. (1994) High-density multiplex detection of nucleic acid sequences: oligonucleotide ligation assay and sequence-coded separation. *Nucleic Acids Res.*, 22, 4527-34.
 55. Guo, D.C., Qi, Y., He, R, Gupta, P. and Milewicz, D.M. (2003) High throughput detection of small genomic insertions or deletions by Pyrosequencing. *Biotechnol. Lett.*, 25, 1703-7.
 56. Guo, Z., Guilfoyle, R.A., Thiel, A.J., Wang, R. and Smith, L.M. (1994) Direct fluorescence analysis of genetic polymorphisms by hybridisation with oligonucleotide arrays on glass supports. *Nucleic Acids Res.*, 22, 5456-65.

57. Hacia, J.G., Sun, B., Hunt, N., Edgemon, K., Mosbrook, D., Robbins, C., Fodor, S.P., Tagle, D.A. and Collins, F.S. (1998) Strategies for mutational analysis of the large multiexon ATM gene using high-density oligonucleotide arrays. *Genome Res.*, 8, 1245-58.
58. Haff, L.A. and Smirnov, I.P. (1997) Single-nucleotide polymorphism identification assays using a thermostable DNA polymerase and delayed extraction MALDI-TOF mass spectrometry. *Genome Res.*, 7, 378-88.
59. Hall, J.G., Eis, P.S., Law, S.M., Reynaldo, L.P., Prudent, J.R., Marshall, D.J., Allawi, H.T., Mast, A.L., Dahlberg, J.E., Kwiatkowski, R.W., de Arruda, M., Neri, B.P. and Lyamichev, V.I. (2000) Sensitive detection of DNA polymorphisms by the serial invasive signal amplification reaction. *Proc. Natl Acad. Sci. USA*, 97, 8272-7.
60. Hardenbol, P., Baner, J., Maneesh, J., Nilsson, M., Namsaraev, E.A., Karlin-Neumann, G.A., Fakhrai-Rad, H., Ronaghi, M., Willis, T.D., Landegren, U. and Davis, R.W. (2003) Multiplexed genotyping with sequence-tagged molecular inversion probes. *Nat. Biotechnol.*, 21, 673-8.
61. Hawkins, J.R., Khripin, Y., Valdes, M. and Weaver, T.A. (2002) Miniaturized sealed-tube allele-specific PCR. *Hum. Mutat.*, 19, 543-53.
62. Hayney, M.S., Dimanlig, P., Lipsky, J.J. and Poland, G.A. (1995) Utility of a "swish and spit" technique for the collection of buccal cells for TAP haplotype determination. *Mayo Clin. Proc.*, 70, 951-4.
63. Heaton, R.J., Peterson, A.W. and Georgiadis, R.M. (2001) Electrostatic surface plasmon resonance: direct electric field-induced hybridization and denaturation in monolayer nucleic acid films and label-free discrimination of base mismatches. *Proc. Natl Acad. Sci. USA*, 98, 3701-4.
64. Hirschhorn, J.N., Sklar, P., Lindblad-Toh, K., Lim, Y.M., Ruiz-Gutierrez, M., Bolk, S., Langhorst, B., Schaffner, S., Winchester, E. and Lander, E.S. (2000) SBE-TAGS: an array-based method for efficient single-nucleotide polymorphism genotyping. *Proc. Natl Acad. Sci. USA*, 97, 12164-9.
65. Holliday, R. and Grigg, G.W. (1993) DNA methylation and mutation. *Mutation Res.*, 285, 61-7.
66. Horikawa, Y., Oda, N., Cox, N.J., Li, X., Orho-Melander, M., Hara, M., Hinokio, Y., Lindner, T.H., Mashima, H., Schwarz, P.E., del Bosque-Plata, L., Horikawa, Y., Oda, Y., Yoshiuchi, I., Colilla, S., Polonsky, K.S., Wei, S., Concannon, P., Iwasaki, N., Schulze, J., Baier, L.J., Bogardus, C., Groop, L., Boerwinkle, E.,

- Hanis, C.L. and Bell, G.I. (2000) Genetic variation in the gene encoding calpain-10 is associated with type 2 diabetes mellitus. *Nat. Genet.*, 26, 163-75.
67. Howell, W.M., Jobs, M., Gyllensten, U. and Brookes, A.J. (1999) Dynamic allele-specific hybridization. A new method for scoring single nucleotide polymorphisms. *Nat. Biotechnol.*, 17, 87-8.
 68. Huang, Y., Paxton, W.A. and Wolinsky, S.M. (1996) The role of a mutant CCR5 allele in HIV-1 transmission and disease progression. *Nature Med.*, 2, 1240-3.
 69. Iannone, M.A., Taylor, J.D., Chen, J., Li, M.S., Rivers, P., Slentz-Kesler, K.A. and Weiner, M.P. (2000) Multiplexed single nucleotide polymorphism genotyping by oligonucleotide ligation and flow cytometry. *Cytometry*, 39, 131-40.
 70. Ivanov, I., Loffert, D., Kang, J., Ribbe, J. and Steinert, K. (2001) Method for reversible modification of thermostable enzymes. United States Patent 6,183,998.
 71. Jacobsen, N., Bentzen, J., Meldgaard, M., Jakobsen, M.H., Fenger, M., Kauppinen, S. and Skouv, J. (2002) LNA-enhanced detection of single nucleotide polymorphisms in the apolipoprotein E. *Nucleic Acids Res.*, 30, e100.
 72. Khanna, M., Park, P., Zirvi, M., Cao, W., Picon, A., Day, J., Paty, P., and Barany, F. (1999) Multiplex PCR/LDR for detection of K-ras mutations in primary colon tumors. *Oncogene*, 18, 27-38.
 73. Kirk, B.W., Feinsod, M., Favis, R., Kliman, R.M. and Barany, F. (2002) Single nucleotide polymorphism seeking long term association with complex disease. *Nucleic Acids Res.*, 30, 3295-311.
 74. Kostrikis, L.G., Tyagi, S., Mhlanga, M.M., Ho, D.D. and Kramer, F.R. (1998) Spectral genotyping of human alleles. *Science*, 279, 1228-9.
 75. Kozhekbaeva, J.M., Borodina, T.A., Borinskajya, S.A., Gusar, V.A., Feshchenko, S.P., Akhmetova, V.L., Khusainova, R.I., Gupalo, E.Yu., Spitsyn, V.A., Grechanina, E.Ya., Khusnutdinova, E.K. and Yankovsky, N.K. (2004) Distribution of HIV-protective alleles (CCR5delta32, CCR2-64I and SDF1 3'A) in samples of russians, ucrainians and belorussians. *Genetika (rus)*, 40, 1-8.
 76. Kristensen, V.N., Kelefiotis, D., Kristensen, T. and Borresen-Dale, A.L. (2001) High-throughput methods for detection of genetic variation. *Biotechniques*, 30, 318-22, 324, 326.
 77. Kuppaswamy, M.N., Hoffmann, J.W., Kasper, C.K., Spitzer, S.G., Groce, S.L. and Bajaj, S.P. (1991) Single nucleotide primer extension to detect genetic diseases: experimental application to hemophilia B (factor IX) and cystic fibrosis genes. *Proc. Natl Acad. Sci. USA*, 88, 1143-7.

78. Kurg, A., Tonisson, N., Georgiou, I., Shumaker, J., Tollett, J. and Metspalu, A. (2000) Arrayed primer extension: solid-phase four-color DNA resequencing and mutation detection technology. *Genet. Test.*, 4, 1-7.
79. Kwok, P.-Y. and Chen, X. (2003) Detection of single nucleotide polymorphisms. *Curr. Issues Mol. Biol.*, 5, 43-60.
80. Kwok, P.-Y. (2001) Methods for genotyping single nucleotide polymorphisms. *Annu. Rev. Genomics Hum. Genet.*, 2, 235-58.
81. Lai, E. (2001) Application of SNP technologies in medicine: lessons learned and future challenges. *Genome Res.*, 11, 927-9.
82. Landegren, U., Kaiser, R., Sanders, J. and Hood, L. (1988) A ligasemediated gene detection technique. *Science*, 241, 1077-80.
83. Lee, L.G., Livak, K.J., Mullah, B., Graham, R.J., Vinayak, R.S. and Woudenberg, T.M. (1999) Seven-color, homogeneous detection of six PCR products. *Biotechniques*, 27, 342-9.
84. Li, J., Butler, J.M., Tan, Y., Lin, H., Royer, S., Ohler, L., Shaler, T.A., Hunter, J.M., Pollart, D.J., Monforte, J.A. and Becker, C.H. (1999) Single nucleotide polymorphism determination using primer extension and time-of-flight mass spectrometry. *Electrophoresis*, 20, 1258-65.
85. Li, W.H., Ellsworth, D.L., Krushkal, J., Chang, B.H. and Hewett-Emmet, D. (1996) Rates of nucleotide substitution in primates and rodents and the generation-time effect hypothesis. *Mol. Phylogenet. Evol.*, 5, 182-7.
86. Lindblad-Toh, K., Winchester, E., Daly, M.J., Wang, D.G., Hirschhorn, J.N., Lavolette, J.P., Ardlie, K., Reich, D.E., Robinson, E., Sklar, P., Shah, N., Thomas, D., Fan, J.B., Gingeras, T., Warrington, J., Patil, N., Hudson, T.J. and Lander, E.S. (2000) Large-scale discovery and genotyping of single-nucleotide polymorphisms in the mouse. *Nat. Genet.*, 24, 381-6.
87. Lindroos, K., Liljedahl, U., Raitio, M. and Syvanen, A. C. (2001) Minisequencing on oligonucleotide microarrays: comparison of immobilisation chemistries. *Nucleic Acids Res.*, 29, e69.
88. Little, D.P., Braun, A., Darnhofer-Demar, B., Frilling, A., Li, Y., McIver, R.T. Jr, and Koster, H. (1997) Detection of RET proto-oncogene codon 634 mutations using mass spectrometry. *J. Mol. Med.*, 75, 745-50.
89. Little, D.P., Braun, A., Darnhofer-Demar, B. and Koster H. (1997) Identification of apolipoprotein E polymorphisms using temperature cycled primer oligo base extension and mass spectrometry. *Eur. J. Clin. Chem. Clin. Biochem.*, 35, 545-8.

90. Liu, R., Paxton, W.A., Choe, S., Ceradini, D., Martin, S.R., Horuk, R., MacDonald, M.E., Stuhlmann, H., Koup, R.A. and Landau, N.R. (1996) Homozygous defect in HIV co-receptor accounts for resistance of some multiply-exposed individuals to HIV-1 infection. *Cell*, 86, 367-77.
91. Livak, K. J. (1999) Allelic discrimination using fluorogenic probes and the 5' nuclease assay. *Genet. Anal.*, 14, 143-9.
92. Lizardi, P.M., Huang, X., Zhu, Z., Bray-Ward, P., Thomas, D.C. and Ward, D.C. (1998) Mutation detection and single-molecule counting using isothermal rolling-circle amplification. *Nat Genet.*, 19, 225-32.
93. Lyamichev, V., Mast, A.L., Hall, J.G., Prudent, J.R., Kaiser, M.W., Takova, T., Kwiatkowski, R.W., Sander, T.J., de Arruda, M., Arco, D.A., Neri, B.P. and Brow, M.A. (1999) Polymorphism identification and quantitative detection of genomic DNA by invasive cleavage of oligonucleotide probes. *Nat. Biotechnol.*, 17, 292-6.
94. Lysov, Iu.P., Florent'ev, V.L., Khorlin, A.A., Khrapko, K.R. and Shik, V.V. (1988) Determination of the nucleotide sequence of DNA using hybridization with oligonucleotides. A new method. *Dokl. Akad. Nauk. SSSR (rus)*, 303, 1508-11.
95. Martin, E.R., Lai, E.H., Gilbert, J.R., Rogala, A.R., Afshari, A.J., Riley, J., Finch, K.L., Stevens, J.F., Livak, K.J., Slotterbeck, B.D., Slifer, S.H., Warren, L.L., Conneally, P.M., Schmechel, D.E., Purvis, I., Pericak-Vance, M.A., Roses, A.D. and Vance, J.M. (2000) SNPping away at complex diseases: analysis of single-nucleotide polymorphisms around APOE in Alzheimer disease. *Am. J. Hum. Genet.*, 67, 383-94.
96. Matisse, T.C., Sachidanandam, R., Clark, A.G., Kruglyak, L., Wijisman, E., Kakol, J., Buyske, S., Chui, B., Cohen, P., Toma, Cd.C., Ehm, M., Glanowski, S., He, C., Heil, J., Markianos, K., McMullen, I., Pericak-Vance, M.A., Silbergleit, A., Stein, L., Wagner, M., Wilson, A.F., Winick, J.D., Winn-Deen, E.S., Yamashiro, C.T., Cann, H.M., Lai, E. and Holden, A.L. (2003). A 3.9-centimorgan-resolution human single-nucleotide polymorphism linkage map and screening set. *Am. J. Hum. Genet.*, 73, 271-84.
97. McCarthy, L.C., Hosford, D.A., Riley, J.H., Bird, M.I., White, N.J., Hewett, D.R., Peroutka, S.J., Griffiths, L.R., Boyd, P.R., Lea, R.A., Bhatti, S.M., Hosking, L.K., Hood, C.M., Jones, K.W., Handley, A.R., Rallan, R., Lewis, K.F. et al. (2002) Single-nucleotide polymorphism alleles in the insulin receptor gene are associated with typical migraine. *Genomics*, 78, 135-49.

98. Medintz, I., Wong, W.W., Berti, L., Shioh, L., Tom, J., Scherer, J., Sensabaugh, G. and Mathies, R.A. (2001) High-performance multiplex SNP analysis of three hemochromatosis-related mutations with capillary array electrophoresis microplates. *Genome Res.*, 11, 413-21.
99. Meinke, D.W., Cherry, J.M., Dean, C., Rounsley, S.D. and Koornneef, M. (1998) *Arabidopsis thaliana*: A model plant for genome analysis. *Science*, 282, 662–82.
100. Mengel-Jorgensen, J., Sanchez, J.J., Borsting, C., Kirpekar, F. and Morling, N. (2004) MALDI-TOF mass spectrometric detection of multiplex single base extended primers. A study of 17 y-chromosome single-nucleotide polymorphisms. *Anal. Chem.*, 76, 6039-45.
101. Mhlanga, M.M. and Malmberg, L. (2001) Using molecular beacons to detect single-nucleotide polymorphisms with real-time PCR. *Methods*, 25, 463-71.
102. Mir, K. U. and Southern, E. M. (1999) Determining the influence of structure on hybridization using oligonucleotide arrays. *Nature Biotechnol.*, 17, 788–92.
103. Myakishev, M.V., Khripin, Y., Hu, S. and Hamer, D.H. (2001) High-throughput SNP genotyping by allele-specific PCR with universal energy-transfer-labeled primers, *Genome Res.*, 11, 163-169.
104. Myer, S.E. and Day, D.J. (2001) Synthesis and application of circularizable ligation probes. *Biotechniques*, 30, 584–93.
105. Newton, C.R., Graham, A., Heptinstall, L.E., Powell, S.J., Summers, C., Kalsheker, N., Smith, J.C. and Markham, A.F. (1989) Analysis of any point mutation in DNA. The amplification refractory mutation system (ARMS). *Nucleic Acids Res.*, 17, 2503–16.
106. Nickerson, D.A., Kaiser, R., Lappin, S., Stewart, J., Hood, L. and Landegren, U. (1990) Automated DNA diagnostics using an ELISA-based oligonucleotide ligation assay. *Proc. Natl Acad. Sci. USA*, 87, 8923-7.
107. Nikiforov, T.T., Rendle, R.B., Goelet, P., Rogers, Y.H., Kotewicz, M.L., Anderson, S., Trainor, G.L. and Knapp, M.R. (1994) Genetic bit analysis: a solid phase method for typing single nucleotide polymorphisms. *Nucleic Acids Res.*, 22, 4167-75.
108. Nilsson, M., Baner, J., Mendel-Hartvig, M., Dahl, F., Antson, D.-O., Gullberg, M. and Landegren, U. (2002) Making ends meet in genetic analysis using padlock probes. *Hum. Mutat.*, 19, 410-5.
109. Nilsson, M., Krejci, K., Koch, J., Kwiatkowski, M., Gustavsson, P. and Landegren, U. (1997) Padlock probes reveal single-nucleotide differences, parent

- of origin and in situ distribution of centromeric sequences in human chromosomes 13 and 21. *Nature Genet.*, 16, 252-5.
110. Nilsson, M., Malmgren, H., Samiotaki, M., Kwiatkowski, M., Chowdhary, B.P. and Landegren, U. (1994) Padlock probes: circularizing oligonucleotides for localized DNA detection. *Science*, 265, 2085-8.
111. Nordborg, M. and Tavaré, S. (2002) Linkage disequilibrium: what history has to tell us. *Trends Genet.*, 18, 83-90.
112. O'Connell, C.D., Atha, D.H., Oldenburg, M.C., Tian, J., Siebert, M., Handrow, R., Grooms, K., Heisler, L. and de Arruda, M. (1999) Detection of p53 gene mutations: analysis by single-strand conformation polymorphism and Cleavase fragment length polymorphism. *Electrophoresis*, 20, 1211-23.
113. Oldenburg, M.C. and Siebert, M. (2000) New Cleavase Fragment Length Polymorphism method improves the mutation detection assay. *Biotechniques*, 28, 351-7.
114. Orum, H., Jakobsen, M.H., Koch, T., Vuust, J. and Borre, M.B. (1999) Detection of the factor V Leiden mutation by direct allele-specific hybridization of PCR amplicons to photoimmobilized locked nucleic acids. *Clin. Chem.*, 45, 1898-905.
115. Pastinen, T., Kurg, A., Metspalu, A., Peltonen, L. and Syvanen, A.C. (1997). Minisequencing: a specific tool for DNA analysis and diagnostics on oligonucleotide arrays. *Genome Res.*, 7, 606-14.
116. Pastinen, T., Raitio, M., Lindroos, K., Tainola, P., Peltonen, L. and Syvanen, A.C. (2000) A system for specific, high-throughput genotyping by allele-specific primer extension on microarrays. *Genome Res.*, 10, 1031-42.
117. Patil, N., Berno, A.J., Hinds, D.A., Barrett, W.A., Doshi, J.M., Hacker, C.R., Kautzer, C.R., Lee, D.H., Marjoribanks, C., McDonough, D.P., Nguyen, B.T., Norris, M.C. et al. (2001) Blocks of limited haplotype diversity revealed by high-resolution scanning of human chromosome 21. *Science*, 294, 1719-23.
118. Pease, A.C., Solas, D., Sullivan, E.J., Cronin, M.T., Holmes, C.P. and Fodor, S.P. (1994) Light-generated oligonucleotide arrays for rapid DNA sequence analysis. *Proc. Natl Acad. Sci. USA*, 91, 5022-6
119. Pickering, J., Bamford, A., Godbole, V., Briggs, J., Scozzafava, G., Roe, P., Wheeler, C., Ghouze, F., and Cuss, S. (2002) Integration of DNA ligation and rolling circle amplification for the homogeneous, end point detection of single nucleotide polymorphisms. *Nucleic Acids Res.*, 30, e60.

120. Prince, J.A., Feuk, L., Howell, W.M., Jobs, M., Emahazion, T., Blennow, K. and Brookes, A.J. (2001) Robust and accurate single nucleotide polymorphism genotyping by dynamic allele-specific hybridization (DASH): design criteria and assay validation. *Genome Res.*, 11, 152-62.
121. Raitio, M., Lindroos, K., Laukkanen, M., Pastinen, T., Sistonen, P., Sajantila, A. and Syvanen, A.C. (2001) Y-chromosomal SNPs in Finno-Ugricspeaking populations analyzed by minisequencing on microarrays. *Genome Res.*, 11, 471-82.
122. Rao, K.V., Stevens, P.W., Hall, J.G., Lyamichev, V., Neri, B.P. and Kelso, D.M. (2003) Genotyping single nucleotide polymorphisms directly from genomic DNA by invasive cleavage reaction on microspheres. *Nucleic Acids Res.*, 31, e66.
123. Ren, B., Zhou, J.M. and Komiyama, M. (2004) Straightforward detection of SNPs in double-stranded DNA by using exonuclease III/nuclease S1/PNA system. *Nucleic Acids Res.*, 32, e42.
124. Rickert, A.M., Premstaller, A., Gebhardt, C. and Oefner, P.J. (2002) Genotyping of Snps in a polyploid genome by pyrosequencing. *Biotechniques*, 32, 592-3, 596-8.
125. Rickert, A.M., Borodina, T.A., Kuhn, E.J., Lehrach, H. and Sperling, S. (2004) Refinement of single-nucleotide polymorphism genotyping methods on human genomic DNA: amplifluor allele-specific polymerase chain reaction versus ligation detection reaction-TaqMan. *Anal Biochem.*, 330, 288-97.
126. Robinson, M. and Williams, S.M. (2004) Role of two angiotensinogen polymorphisms in blood pressure variation. *J. Hum. Hypertens.*, 18, 865-9.
127. Rogers, S.O. and Bendich, A.J. (1985) Extraction of DNA from milligram amounts of fresh, herbarium and mummified plant tissues. *Plant Molecular Biology*, 5, 69-76.
128. Ronaghi, M. (2001) Pyrosequencing sheds light on DNA sequencing. *Genome Res.*, 11, 3-11.
129. Ronaghi, M., Karamohamed, S., Pettersson, B., Uhlen, M. and Nyren, P. (1996) Real-time DNA sequencing using detection of pyrophosphate release. *Anal. Biochem.*, 242, 84-9.
130. Ross, P., Hall, L., Smirnov, I. and Haff, L. (1998) High level multiplex genotyping by MALDI-TOF mass spectrometry. *Nat. Biotechnol.*, 16, 1347-51.

131. Ross, P.L., Lee, K. and Belgrader, P. (1997) Discrimination of single nucleotide polymorphisms in human DNA using peptide nucleic acid probes detected by MALDI-TOF mass spectrometry. *Anal. Chem.*, 69, 4197–202.
132. Rossi, R., Montecucco, A., Ciarrocchi, G. and Biamonti, G. (1997) Functional characterization of the T4 DNA ligase: a new insight into the mechanism of action. *Nucleic Acids Res.*, 25, 2106-13.
133. Rotimi, C., Cooper, R., Ogunbiyi, O., Morrison, L., Ladipo, M., Tewksbury, D. and Ward, R. (1997) Hypertension, serum angiotensinogen, and molecular variants of the angiotensinogen gene among Nigerians. *Circulation*, 95, 2348-50.
134. Sachidanandam, R., Weissman, D., Schmidt, S., Kakol, J., Stein, L., Marth, G., Sherry, S., Mullikin, J., Mortimore, B., Willey, D., Hunt, S., Cole, C., Coghill, P., Rice, C. et al. The International SNP Map Working Group (2001) A map of human genome sequence variation containing 1.42 million single nucleotide polymorphisms. *Nature*, 409, 928–33.
135. Saiki, R.K., Bugawan, T.L., Horn, G.T., Mullis, K.B. and Erlich, H.A. (1986) Analysis of enzymatically amplified beta-globin and HLA-DQ alpha DNA with allele-specific oligonucleotide probes. *Nature*, 324, 163-6.
136. Sambrook J. and Russell, D.W. (2001) Molecular cloning: a laboratory manual. NY, Cold Spring Harbor Laboratory press.
137. Samiotaki, M., Kwiatkowski, M., Parik, J. and Landegren, U. (1994) Dual-color detection of DNA sequence variants by ligase-mediated analysis. *Genomics*, 20, 238-42.
138. Sander, T., Olson, S., Hall, J., Siebert, M., Grooms, K., Heisler, L., de Arruda, M. and Neri, B. (1999) Comparison of detection platforms and post-polymerase chain reaction DNA purification methods for use in conjunction with Cleavase fragment length polymorphism analysis. *Electrophoresis*, 20, 1131-40.
139. Sapolsky, R.J., Hsie, L., Berno, A., Ghandour, G., Mittmann, M. and Fan, J.B. (1999) High-throughput polymorphism screening and genotyping with high-density oligonucleotide arrays. *Genet. Anal.*, 14, 187-92.
140. Sasvari-Szekely, M., Gerstner, A., Ronai, Z., Staub, M. and Guttman, A. (2000) Rapid genotyping of factor V Leiden mutation using single-tube bidirectional allele-specific amplification and automated ultrathin-layer agarose gel electrophoresis. *Electrophoresis*, 21, 816-21.

141. Sauer, S., Lechner, D., Berlin, K., Lehrach, H., Escary, J.L., Fox, N. and Gut, I.G. (2000) A novel procedure for efficient genotyping of single nucleotide polymorphisms. *Nucleic Acids Res.*, 28, e13.
142. Schmid, K.J., Sorensen, T.R., Stracke, R., Torjek, O., Altmann, T., Mitchell-Olds, T. and Weisshaar, B. (2003) Large-scale identification and analysis of genome-wide single-nucleotide polymorphisms for mapping in *Arabidopsis thaliana*. *Genome Res.*, 13, 1250-7.
143. Schork, N.J., Cardon, L.R. and Xu, X. (1998) The future of genetic epidemiology. *Trends Genet.*, 14, 266-72.
144. Schouten, J.P., McElgunn, C.J., Waaijer, R., Zwijnenburg, D., Diepvens, F. and Pals, G. (2002) Relative quantification of 40 nucleic acid sequences by multiplex ligation-dependent probe amplification. *Nucleic Acids Res.*, 30, e57.
145. Schumm, J.W., Knowlton, R.G., Braman, J.C., Barker, D.F., Botstein, D., Akots, G., Brown, V.A., Gravius, T.C., Helms, C., Hsiao, K. et al. (1988) Identification of more than 500 RFLPs by screening random genomic clones. *Am. J. Hum. Genet.*, 42, 143-159.
146. Sherry, S.T., Ward, M.-H., Kholodov, M., Baker, J., Phan, L., Smigielski, E. M. and Sirotkin, K. (2001) dbSNP: the NCBI database of genetic variation. *Nucleic Acids Res.*, 29, 308-11.
147. Shi, M.M. (2001) Enabling large-scale pharmacogenetic studies by high-throughput mutation detection and genotyping technologies. *Clin Chem.*, 47, 164-72.
148. Shumaker, J.M., Metspalu, A. and Caskey, C.T. (1996) Mutation detection by solid phase primer extension. *Hum. Mutat.*, 7, 346-54.
149. Sipe J.C., Chiang, K., Gerber, A.L., Beutler, E. and Cravatt, B.F. (2002) A missense mutation in human fatty acid amide hydrolase associated with problem drug use. *Proc. Natl Acad. Sci. USA*, 99, 8394-9.
150. Soldatov, A.V., Borodina, T.A., and Lehrach, H. Ligation-based method of analysis of single nucleotide polymorphisms on genomic DNA. European patent EP03019521.
151. Soldatov, A.V., Borodina, T.A., and Lehrach, H. Ligation-based synthesis of oligonucleotides with block structure. European patent PCT/EP 2004/003921
152. Sosnowski, R.G., Tu, E., Butler, W.F., O'Connell, J.P. and Heller, M.J. (1997) Rapid determination of single base mismatch mutations in DNA hybrids by direct electric field control. *Proc. Natl Acad. Sci. USA*, 94, 1119-23.

153. Southern, E.M., Maskos, U. and Elder, J.K. (1992) Analyzing and comparing nucleic acid sequences by hybridization to arrays of oligonucleotides: evaluation using experimental models. *Genomics*, 13, 1008-17.
154. Steemers, F.J., Ferguson, J.A. and Walt, D.R. (2000) Screening unlabeled DNA targets with randomly ordered fiber-optic gene arrays. *Nature Biotechnol.*, 18, 91-4.
155. Steinmetz, L., Mindrinos, M., and Oefner, P. (2000) Combining genome sequences and new technologies for dissecting the genetics of complex phenotypes. *Trends Plant Sci.*, 5, 397-401.
156. Sun, X., Ding, H., Hung, K., Guo, B. (2000) A new MALDI-TOF based mini-sequencing assay for genotyping of SNPS. *Nucleic Acids Res.*, 8, e68.
157. Syvanen, A.C. (2001) Accessing genetic variation: genotyping single nucleotide polymorphisms. *Nat Rev Genet.*, 2, 930-42.
158. Taylor, J.D., Briley, D., Nguyen, Q., Long, K., Iannone, M.A., Li, M.S., Ye, F., Afshari, A., Lai, E., Wagner, M., Chen, J. and Weiner, M.P. (2001) Flow cytometric platform for highthroughput single nucleotide polymorphism analysis. *Biotechniques*, 30, 661-6, 668-9.
159. The Arabidopsis Genome Initiative. (2000) Analysis of the genome sequence of the flowering plant *Arabidopsis thaliana*. *Nature*, 408, 796-815.
160. The International HapMap Consortium. (2004) Integrating ethics and science in the International HapMap Project. *Nat. Rev. Genet.*, 5, 467-75.
161. Thorisson, G.A. and Stein, L.D. (2003) The SNP Consortium website: past, present and future. *Nucleic Acids Res.*, 31, 124-27.
162. Todd, A.V., Fuery, C.J., Impey, H.L., Applegate, T.L. and Haughton, M.A. (2000) DzyNAPCR: use of DNAzymes to detect and quantify nucleic acid sequences in a real-time fluorescent format. *Clin. Chem.*, 46, 625-30.
163. Tonisson, N., Zernant, J., Kurg, A., Pavel, H., Slavin, G., Roomere, H., Meiel, A., Hainaut, P. and Metspalu, A. (2002) Evaluating the arrayed primer extension resequencing assay of TP53 tumor suppressor gene. *Proc. Natl Acad. Sci. USA*, 99, 5503-8.
164. Torjek, O., Berger, D., Meyer, R.C., Mussig, C., Schmid, K.J., Rosleff Sorensen, T., Weisshaar, B., Mitchell-Olds, T. and Altmann, T. (2003) Establishment of a high-efficiency SNP-based framework marker set for Arabidopsis. *Plant J.*, 36, 122-40.

165. Twyman, R.M. (2004) SNP discovery and typing technologies for pharmacogenomics. *Curr. Top Med. Chem.*, 4, 1421-9.
166. Twyman, R.M. and Primrose, S.B. (2003) Techniques patents for SNP genotyping. *Pharmacogenomics*, 4, 67-79.
167. Tyagi, S., Bratu, D.P. and Kramer, F.R. (1998) Multicolor molecular beacons for allele discrimination. *Nature Biotechnol.*, 16, 49–53.
168. Tyagi, S., Marras, S.A. and Kramer, F.R. (2000) Wavelength-shifting molecular beacons. *Nature Biotechnol.*, 18, 1191–96.
169. Utting, M., Hampe, J., Platzer, M. and Huse, K. (2004) Locking of 3'-ends of single-stranded DNA templates for improved Pyrosequencing performance. *Biotechniques*, 37, 66-7, 70-3.
170. Vallone, P.M., Fahr, K. and Kostrzewa, M. (2004) Genotyping SNPs Using a UV-Photocleavable Oligonucleotide in MALDI-TOF MS. *Methods Mol. Biol.*, 297, 169-78.
171. Vos, P., Hogers, R., Bleeker, M., Reijans, M., van de Lee, T., Hornes, M., Frijters, A., Pot, J., Peleman, J., Kuiper M. et al., (1995) AFLP: a new technique for DNA fingerprinting. *Nucleic Acids Res.*, 23, 4407-14.
172. Wang, D.G., Fan, J.B., Siao, C.J., Berno, A., Young, P., Sapolsky, R., Ghandour, G., Perkins, N., Winchester, E., Spencer, J., Kruglyak, L., Stein, L., Hsie, L. et al. (1998) Large-scale identification, mapping, and genotyping of single- nucleotide polymorphisms in the human genome. *Science*, 280, 1077–82.
173. Wheeler, D.L., Church, D.M., Edgar, R., Federhen, S., Helmberg, W., Madden, T.L., Pontius, J.U., Schuler, G.D., Schriml, L.M., Sequeira, E., Suzek, T.O., Tatusova, T.A. and Wagner, L. (2004). Database resources of the National Center for Biotechnology Information: update. *Nucleic Acids Res.*, 32, D35-40.
174. Winkler, C., Modi, W., Smith, M.W., Nelson, G.W., Wu, X., Carrington, M., Dean, M., Honjo, T., Tashiro, K., Yabe, D., Buchbinder, S., Vittinghoff, E., Goedert, J.J. et al. (1998) Genetic restriction of AIDS pathogenesis by an SDF-1 chemokine gene variant. *Science*, 279, 389–93.
175. Wu, D.Y. and Wallace, R.B. (1989) Specificity of the nick-closing activity of bacteriophage T4 DNA ligase. *Gene*, 76, 245-54.
176. Xu, Y. and Kool, E.T. (1999). High sequence fidelity in a non-enzymatic DNA autoligation reaction. *Nucleic Acids Res.*, 27, 875-81.

177. Ye, S., Dhillon, S., Ke, X., Collins, A.R. and Day, I.N. (2001) An efficient procedure for genotyping single nucleotide polymorphisms. *Nucleic Acids Res.*, 29, e88.
178. Zhang, D.Y., Brandwein, M., Hsuih, T.C. and Li, H. (1998) Amplification of target-specific, ligation-dependent circular probe. *Gene*, 211, 277-85.
179. Zhang, J. and Li, K. (2003) Single-base discrimination mediated by proofreading 3'-phosphorothioate-modified primers. *Mol Biotechnol.*, 25, 223-8.
180. Zhu, K.Y. and Clark, J.M. (1996) Addition of a competitive primer can dramatically improve the specificity of PCR amplification of specific alleles. *Biotechniques*, 21, 586-90.