

Jurg Ott, PhD

Updated 26 October 2014

BOOKS

- Ott J (1999) *Analysis of Human Genetic Linkage*, 3rd edition. Johns Hopkins University Press, Baltimore, 382 pages. Japanese translation of first edition: Soft Science, Tokyo, 1987
Terwilliger JD, Ott J (1994) *Handbook of Human Genetic Linkage*. Johns Hopkins University Press, Baltimore

ORIGINAL ARTICLES, INVITED REVIEWS

- 1) **Ott J** (1968) Natural reproductive isolation between *Sorex gemellus* sp.n. and *Sorex araneus* Linnaeus 1758 in Switzerland (Mammalia, Insectivora) (in German with English summary). *Rev suisse Zool* 75, 53-75
- 2) Heusser H, **Ott J** (1968) Wanderzeit und populationsspezifische Sollzeit der Laichwanderung bei der Erdkröte, *Bufo bufo* (L.). *Rev suisse Zool* 75, 1005-1022
- 3) **Ott J**, Olert J (1970) Color differences between *Sorex araneus* Linnaeus 1758 and *Sorex gemellus* Ott 1968 (Mammalia, Insectivora) (in German with English summary). *Rev suisse Zool* 77, 283-291
- 4) Wehner R, **Ott J** (1972) Capability of honey bees to discriminate certain patterns (in German with English summary). *Rev suisse Zool* 79, 843-860
- 5) **Ott J** (1974) Estimation of the recombination fraction in human pedigrees: efficient computation of the likelihood for human linkage studies. *Am J Hum Genet* 26, 588-597
- 6) **Ott J**, Schrott HG, Goldstein JL, Hazzard WR, Allen FH Jr, Falk CT, Motulsky AG (1974) Linkage studies in a large kindred with familial hypercholesterolemia. *Am J Hum Genet* 26, 598-603
- 7) Bremner WJ, **Ott J**, Moore DJ, Paulsen CA (1974) Reifenschein's syndrome: Investigation of linkage to X-chromosomal loci. *Clin Genet* 6, 216-220
- 8) **Ott J**, Goldstein JL, Harrod MJ (1975) Linkage investigation of a large family with Reifenschein's syndrome. *Clin Genet* 7, 342-344
- 9) Weitkamp LR, Lovrien EW, Olaisen B, Fenger K, Gedde-Dahl Jr T, Sorensen SA, Conneally PM, Bias WB, **Ott J** (1975) Linkage relations of the loci for the MN blood group and red cell acid phosphatase. *Cytogenet Cell Genet* 14, 446-450
- 10) **Ott J**, Kronmal RA (1976) Some classification procedures for multivariate binary data using orthogonal functions. *J Am Statist Assoc* 71, 391-399
- 11) **Ott J** (1976) Note on the prior probability of autosomal linkage. *Ann Hum Genet* 39, 433-434
- 12) **Ott J**, Hecht F, Linder D, Lovrien EW, Kaiser-McCaw B (1976) Human centromere mapping using teratoma data. *Cytogenet Cell Genet* 16, 396-398
- 13) **Ott J**, Linder D, Kaiser-McCaw B, Lovrien EW, Hecht F (1976) Estimating distances from the centromere by means of benign ovarian teratomas in man. *Ann Hum Genet* 40, 191-196
- 14) **Ott J** (1976) A computer program for linkage analysis of general human pedigrees. *Am J Hum Genet* 28, 528-529
- 15) **Ott J** (1977) Counting methods (EM algorithm) in human pedigree analysis: linkage and segregation analysis. *Ann Hum Genet* 40, 443-454
- 16) Kukulich MK, Telsey A, **Ott J**, Motulsky AG (1977) Sudden infant death syndrome: normal QT interval on ECG's of relatives. *Pediatrics* 60, 51-54
- 17) **Ott J** (1977) Linkage analysis with misclassification at one locus. *Clin Genet* 12, 110-124
- 18) **Ott J** (1978) Some statistical properties of the lod method and the method of scoring known recombination events in linkage analysis. *Cytogenet Cell Genet* 22, 702-705

- 19) **Ott J** (1978) A simple scheme for the analysis of HLA linkages in pedigrees. *Ann Hum Genet* 42, 255-257
- 20) **Ott J** (1979) Human gene mapping by postreduction and recombination frequencies under complete interference. *Clin Genet* 15, 11-16
- 21) **Ott J** (1979) Maximum likelihood estimation by counting methods under polygenic and mixed models in human pedigrees. *Am J Hum Genet* 31, 161-175
- 22) **Ott J** (1979) Detection of rare major genes in lipid levels. *Hum Genet* 51, 79-91
- 23) Rossen RD, Brewer EJ, Sharp RM, **Ott J**, Templeton JW (1980) Familial rheumatoid arthritis - Linkage of HLA to disease susceptibility locus in four families where proband presented with juvenile rheumatoid arthritis. *J Clin Invest* 65, 629-642
- 24) **Ott J**, Frater-Schröder M (1981) Absence of linkage between transcobalamin II and ABO. *Hum Genet* 59, 164-165
- 25) Bird TD, **Ott J**, Giblett ER (1982) Evidence for linkage of Charcot-Marie-Tooth neuropathy to the Duffy locus on chromosome 1. *Am J Hum Genet* 34, 388-394
- 26) **Ott J**, Falk CT (1982) Epistatic association and linkage analysis in human families. *Hum Genet* 62, 296-300
- 27) **Ott J** (1983) Tag und Stunde der Geburt. *Zürcher Statistische Nachrichten*, pp. 155-162 ["Day and hour of birth in Zurich", in German; a statistical treatment of city census data by means of circular distributions]
- 28) **Ott J** (1983) Linkage analysis and family classification under heterogeneity. *Ann Hum Genet* 47, 311-320
- 29) Dyck PJ, **Ott J**, Moore SB, Swanson CJ, Lambert EH (1983) Linkage evidence for genetic heterogeneity among kinships with hereditary motor and sensory neuropathy, type I. *Mayo Clin Proc* 58, 430-435
- 30) Bird TD, **Ott J**, Giblett ER, Chance PF, Sumi SM, Kraft GH (1983) Genetic linkage evidence for heterogeneity in Charcot-Marie-Tooth neuropathy (HMSN-Type I). *Ann Neurol* 14, 679-684
- 31) Lathrop GM, Lalouel JM, Julier C, **Ott J** (1984) Strategies for multilocus linkage analysis in humans. *Proc Natl Acad Sci USA* 81, 3443-3446
- 32) Mensink EJBM, Schot JDL, Tippett P, **Ott J**, Schuurman RKB (1984) X-linked agammaglobulinemia and the red blood cell determinants Xg and 12E7 are not closely linked. *Hum Genet* 68, 303-309
- 33) Lathrop GM, Lalouel JM, Julier C, **Ott J** (1985) Multilocus linkage analysis in humans: detection of linkage and estimation of recombination. *Am J Hum Genet* 37, 482-498
- 34) **Ott J** (1985) A chi-square test to distinguish allelic association from other causes of phenotypic association between two loci. *Genet Epidemiol* 2, 79-84
- 35) **Ott J** (1985) Genetic Analysis Workshop III: Combining 2-point analyses under the constraints of a linear map and a constant female/male distance ratio. *Genet Epidemiol* 2, 217-218
- 36) **Ott J** (1986) Y-linkage and pseudoautosomal linkage. *Am J Hum Genet* 38, 891-897
- 37) **Ott J** (1986) The number of families required to detect or exclude linkage heterogeneity. *Am J Hum Genet* 39, 159-165
- 38) Gallmann M, Frater-Schröder M, Scheffrahn W, **Ott J**, Schmid B, Bütler E, Biedermann V, Kierat L (1986) Indication against genetic localisation of the human transcobalamin II gene (TC2) on chromosome 16. *Clin Genet* 29, 349-353
- 39) **Ott J**, Mensink EJBM, Thompson A, Schot JDL, Schuurman RKB (1986) Heterogeneity in the map distance between X-linked agammaglobulinemia and a map of nine RFLP loci. *Hum Genet* 74, 280-283
- 40) Beaudet A, Bowcock A, Buchwald M, Cavalli-Sforza LL, Farrall M, King M-C, Klinger K, Lalouel J-M, Lathrop G, Naylor S, **Ott J**, Tsui L-C, Wainwright B, Watkins P, White R, Williamson R (1986) Linkage of Cystic Fibrosis to two

tightly linked markers: joint report from a collaborative study. *Am J Hum Genet* 39, 681-693

- 41) **Ott J** (1986) Linkage probability and its approximate confidence interval under possible heterogeneity. *Genet Epidemiol Suppl* 1, 251-257
- 42) **Ott J**, Lathrop GM (1987) Goodness of fit tests for locus order in three-point mapping. *Genet Epidemiol* 4, 51-57
- 43) Lathrop GM, Chotai J, **Ott J**, Lalouel JM (1987) Tests of gene order from three-locus linkage data. *Ann Hum Genet* 51, 235-249
- 44) Kwan S-P, Sandkuyl LA, Blaese M, Kunkel LM, Bruns G, Parmley R, Skarshaug S, Page DC, **Ott J**, Rosen FS (1988) Genetic mapping of the Wiskott-Aldrich syndrome with two highly-linked polymorphic DNA markers. *Genomics* 3, 39-43
- 45) Sandkuyl LA, **Ott J** (1989) Affective disorders: Evaluation of a three-allele model accounting for clinical heterogeneity. *Genet Epidemiol* 6, 265-269
- 46) Zoghbi HY, Sandkuyl LA, **Ott J**, Daiger SP, Pollack M, O'Brien WE, Beaudet AL (1989) Assignment of autosomal dominant spinocerebellar ataxia (SCA1) centromeric to the HLA region on the short arm of chromosome 6 using multilocus linkage analysis. *Am J Hum Genet* 44, 255-263
- 47) Hsiao K, Baker HF, Crow TJ, Poulter M, Owen F, Terwilliger JD, Westaway D, **Ott J**, Prusiner SB (1989) Linkage of a prion protein missense variant to Gerstmann-Sträussler syndrome. *Nature* 338, 342-345
- 48) Sandkuyl LA, **Ott J** (1989) Determining informativity of marker typing for genetic counseling in a pedigree. *Hum Genet* 82, 159-162
- 49) **Ott J** (1989) Computer-simulation methods in human linkage analysis. *Proc Natl Acad Sci USA* 86, 4175-4178
- 50) Palotie A, Väisänen P, **Ott J**, Ryhänen L, Elima K, Vikkula M, Cheah K, Vuorio E, Peltonen L (1989) Predisposition to familial osteoarthritis is linked to type II collagen gene. *Lancet* i, 924-927 (April 29)
- 51) **Ott J** (1989) Statistical properties of the haplotype relative risk. *Genet Epidemiol* 6, 127-130
- 52) Sankila E-M, Lehner T, Eriksson AW, Forsius H, Kärnä J, Page D, **Ott J**, de la Chapelle A (1989) Haplotype and multipoint linkage analysis in Finnish choroideremia families. *Hum Genet* 84, 66-70
- 53) Weeks DE, **Ott J** (1989) Risk calculations under heterogeneity. *Am J Hum Genet* 45, 819-821
- 54) Weeks DE, **Ott J** (1990) Reply to Dr. Carothers: Support intervals for genetic risks. *Am J Hum Genet* 47, 166
- 55) **Ott J**, Bhattacharya S, Chen JD, Denton MJ, Donald J, Dubay C, Farrar GJ, Fishman GA, Frey D, Gal A, Humphries P, Jay B, Jay M, Litt M, Mächler M, Musarella M, Neugebauer M, Nussbaum RL, Terwilliger JD, Weleber RG, Wirth B, Wong F, Worton RG, Wright AF (1990) Localizing multiple X chromosome-linked retinitis pigmentosa loci using multilocus homogeneity tests. *Proc Natl Acad Sci USA* 87, 701-704
- 56) Ikonen E, Palo J, **Ott J**, Gusella J, Somer H, Karila L, Palotie A, Peltonen L (1990) Huntington disease in Finland: Linkage disequilibrium of chromosome 4 RFLP haplotypes and exclusion of a tight linkage between the disease and D4S43 locus. *Am J Hum Genet* 46, 5-11
- 57) Kwan S-P, Terwilliger J, Parmley R, Raghu G, Sandkuyl LA, **Ott J**, Ochs H, Wedgwood R, Rosen F (1990) Identification of a closely linked DNA marker, DXS178, to further refine the X-linked agammaglobulinemia locus. *Genomics* 6, 238-242
- 58) Shiloh Y, Litvak G, Ziv Y, Lehner T, Sandkuyl L, Hildesheimer M, Buchris V, Cremers FPM, Szabo P, White BN, Holden JJA, **Ott J** (1990) Genetic mapping of X-linked albinism-deafness syndrome (ADFN) to Xq26.3-q27.1. *Am J Hum Genet* 47, 20-27
- 59) Musarella MA, Anson-Cartwright L, Leal SM, Gilbert LD, Worton RG, Fishman GA, **Ott J** (1990) Multipoint linkage analysis and heterogeneity testing in 20 X-linked retinitis pigmentosa families. *Genomics* 8, 286-296

- 60) Brzustowicz LM, Lehner T, Castilla LH, Penchaszadeh GK, Wilhelmsen KC, Daniels R, Davies KE, Leppert M, Ziter F, Wood D, Dubowitz V, Zerres K, Hausmanova-Petrusewicz I, **Ott J**, Munsat TL, Gilliam TC (1990) Genetic mapping of chronic childhood-onset spinal muscular atrophy to chromosome 5q11.2-13.3. *Nature* 344, 540-541
- 61) Gilliam TC, Brzustowicz LM, Castilla LH, Lehner T, Penchaszadeh GK, Daniels RJ, Byth BC, Knowles J, Hislop JE, Shapira Y, Dubowitz V, Munsat TL, **Ott J**, Davies KE (1990) Genetic homogeneity between acute and chronic forms of spinal muscular atrophy. *Nature* 345, 823-825
- 62) Weeks DE, Lehner T, Squires-Wheeler E, Kaufmann C, **Ott J** (1990) Measuring the inflation of the lod score due to its maximization over model parameter values in human linkage analysis. *Genet Epidemiol* 7, 237-243
- 63) Superti-Furga A, Steinmann B, Lee B, Ramirez F, Lehner T, **Ott J**, Gaucher A, Morreau P, Werhya G (1990) Autosomal dominant spondyloarthropathy: No linkage to the type II collagen gene. *N Engl J Med* 322, 552-553
- 64) **Ott J**, Caesar J, Mächler M, Schinzel A, Schmid W (1990) Presymptomatic exclusion of myotonic dystrophy in a one-generation pedigree of half-sibs. *Hum Hered* 40, 305-307
- 65) Baron M, Hamburger R, Sandkuyl LA, Risch N, Mandel B, Endicott J, Belmaker RH, **Ott J** (1990) The impact of phenotypic variation on genetic linkage analysis: application to X-linkage in manic-depressive illness. *Acta Psychiatr Scand* 82, 196-203
- 66) Kwan S-P, Lehner T, Hagemann T, Lu B, Blaese M, Ochs H, Wedgwood R, **Ott J**, Craig IW, Rosen FS (1991) Localization of the gene for the Wiskott-Aldrich syndrome between two flanking markers, TIMP and DXS255, on Xp11.22-Xp11.3. *Genomics* 10, 29-33
- 67) Passos-Bueno MR, Terwilliger J, **Ott J**, Vainzof M, Love DR, Davies KE, Zatz M (1991) Linkage analysis in families with autosomal recessive limb-girdle muscular dystrophy (LGMD) and 6q probes flanking the dystrophin-related sequence. *Am J Med Genet* 38, 140-146
- 68) Passos-Bueno MR, Byth B, Love D, Terwilliger J, **Ott J**, Rapaport D, Vainzof M, Zatz M, Davies KE (1991) Exclusion of the gene responsible for facioscapulohumeral muscular dystrophy (FSH) at 6q23-q27. *J Neurol Sci* 102, 206-208
- 69) Vilkki J, **Ott J**, Savontaus M-L, Aula P, Nikoskelainen EK (1991) Optic atrophy in Leber hereditary optic neuroretinopathy is probably determined by an X-chromosomal gene closely linked to DXS7. *Am J Hum Genet* 48, 486-91
- 70) DeLisi LE, Crow TJ, Davies KE, Terwilliger JD, **Ott J**, Ram R, Flint T, Boccio A (1991) No genetic linkage detected for schizophrenia to Xq27-q28. *Brit J Psychiatry* 158, 630-634
- 71) Mérette C, Lehner T, **Ott J** (1991) Interpreting nonsignificant outcomes of heterogeneity tests in gene mapping. *Am J Hum Genet* 49, 1381-1384
- 72) Moser HW, Moser AB, Smith KD, Bergin A, Boral J, Shankroff J, Stine OC, Mérette C, **Ott J**, Krivit W, Shapiro E (1992) Adrenoleukodystrophy: Phenotypic variability; implications for therapy. *J Inher Metab Dis* 15, 645-664
- 73) Daniels RJ, Thomas NH, MacKinnon RN, Lehner T, **Ott J**, Flint TJ, Dubowitz V, Ignatius J, Donner M, Zerres K, Rietschel M, Cookson WOC, Brzustowicz LM, Gilliam TC, Davies KE (1992) Linkage analysis of spinal muscular atrophy. *Genomics* 12, 335-339
- 74) Terwilliger JD, **Ott J** (1992) A multisample bootstrap approach to the estimation of maximized-over-models lod score distributions. *Cytogenet Cell Genet* 59, 142-144
- 75) Vieland V, Greenberg DA, Hodge SE, **Ott J** (1992) Linkage analysis of two-locus diseases under single-locus and two-locus analysis models. *Cytogenet Cell Genet* 59, 145-146
- 76) Ellison KA, Fill CP, Terwilliger J, DeGennaro LJ, Martin-Gallardo A, Anvret M, Percy AK, **Ott J**, Zoghbi H (1992) Examination of X chromosome markers in Rett syndrome: exclusion mapping with a novel variation on

multilocus linkage analysis. *Am J Hum Genet* 50, 278-287

- 77) Weeks DE, Lehner T, **Ott J** (1992) Preliminary ranking procedures for multilocus ordering based on radiation hybrid data. *Cytogenet Cell Genet* 59, 125-127
- 78) Medori R, Tritschler H-J, LeBlanc A, Villare F, Manetto V, Chen HY, Xue R, Leal S, Montagna P, Cortelli P, Tinuper P, Avoni P, Mochi M, Baruzzi A, Hauw JJ, **Ott J**, Lugaresi E, Autilio-Gambetti L, Gambetti P (1992) Fatal familial insomnia: a prion disease with a mutation at codon 178 of the prion protein gene. *New Engl J Med* 326, 444-449
- 79) Arena JF, Schwartz C, Stevenson R, Lawrence L, Carpenter A, Duara R, Ledbetter D, Huang T, Lehner T, **Ott J**, Lubs HA (1992) Spastic paraplegia with iron deposits in the basal ganglia. A new X-linked mental retardation syndrome. *Am J Med Genet* 43, 479-490
- 80) Mérette C, King M-C, **Ott J** (1992) Heterogeneity analysis of breast cancer families by using age at onset as a covariate. *Am J Hum Genet* 50, 515-519
- 81) **Ott J** (1992) Strategies for characterizing highly polymorphic markers in human gene mapping. *Am J Hum Genet* 51, 283-290
- 82) Terwilliger JD, Ding Y, **Ott J** (1992) On the relative importance of marker heterozygosity and intermarker distance in gene mapping. *Genomics* 13, 951-956
- 83) Brzustowicz LM, Kleyn PW, Boyce FM, Lien LL, Monaco AP, Penchaszadeh GK, Das K, Wang CH, Munsat TL, **Ott J**, Kunkel LM, Gilliam TC (1992) Fine-mapping of the spinal muscular atrophy locus to a region flanked by MAP1B and D5S6. *Genomics* 13, 991-998
- 84) Terwilliger J, **Ott J** (1992) A haplotype-based 'haplotype relative risk' approach to detecting allelic associations. *Hum Hered* 42, 337-346
- 85) Petersen RB, Tabaton M, Berg L, Schrank B, Torack RM, Leal S, Julien J, Vital C, Deleplanque B, Pendlebury WW, Drachman D, Smith TW, Martin JJ, Oda M, Montagna P, **Ott J**, Autilio-Gambetti L, Lugaresi E, Gambetti P (1992) Analysis of the prion protein gene in thalamic dementia. *Neurology* 42, 1859-1863
- 86) Baron M, Freimer NF, Risch N, Lerer B, Alexander JR, Straub RE, Asokan S, Das K, Peterson A, Amos J, Endicott, **Ott J**, Gilliam C (1993) Diminished support for linkage between manic depressive illness and X-chromosome markers in three Israeli pedigrees. *Nature Genetics* 3, 49-55
- 87) **Ott J** (1993) Detecting marker inconsistencies in human gene mapping. *Hum Hered* 43, 25-30
- 88) Weeks DE, Lathrop GM, **Ott J** (1993) Multipoint mapping under genetic interference. *Hum Hered* 43, 86-97
- 89) Straub RE, Speer MC, Luo Y, Rojas K, Overhauser J, **Ott J**, Gilliam TC (1993) A microsatellite genetic linkage map of human chromosome 18. *Genomics* 15, 48-56
- 90) Petrukhin KE, Speer MC, Cayanis E, de Fátima Bonaldo M, Tantravahi U, Soares MB, Fischer SG, Warburton D, Gilliam TC, **Ott J** (1993) A microsatellite genetic linkage map of human chromosome 13. *Genomics* 15, 76-85
- 91) Vikkula M, Palotie A, Ritvaniemi P, **Ott J**, Ala-Kokko L, Sievers U, Aho K, Peltonen L (1993) Early-onset osteoarthritis linked to the type II procollagen gene. *Arthritis and Rheumatism* 36, 401-409
- 92) Bunge S, Wedemann H, David D, Terwilliger DJ, Van den Born LI, Auleha-Scholz C, Samanns C, Horn M, **Ott J**, Schwinger E, Schinzel A, Denton MJ, Gal A (1993) Molecular analysis and genetic mapping of the rhodopsin gene in families with autosomal dominant Retinitis Pigmentosa. *Genomics* 17, 230-233
- 93) Terwilliger JD, Speer M, **Ott J** (1993) Chromosome-based method for rapid computer simulation in human genetic linkage analysis. *Genet Epidemiol* 10, 217-224
- 94) Hellsten E, Vesa J, Speer MC, Mäkelä TP, Järvelä I, Alitalo K, **Ott J**, Peltonen L (1993) Refined assignment of the Infantile Neuronal Ceroid Lipofuscinosis (INCL, CLN1) locus at 1p32: incorporation of linkage disequilibrium in multipoint analysis. *Genomics* 16, 720-725

- 95) Brzustowicz LM, Mérette C, Xie X, Townsend L, Gilliam TC, **Ott J** (1993) Molecular and statistical approaches to the detection and correction of errors in genotype databases. *Am J Hum Genet* 53, 1137-1145
- 96) Gabizon R, Rosenman H, Meiner Z, Kahana I, Kahana E, Shugart Y, **Ott J**, Prusiner SB (1993) Mutation and polymorphism of the prion protein gene in Lybian Jews with Creutzfeldt-Jacob disease. *Am J Hum Genet* 53, 828-835
- 97) Mérette C, Lehner T, **Ott J** (1993) Two new approaches toward linkage heterogeneity of FAD: Two-locus models and age of onset as a discriminator. *Genet Epidemiol* 10, 455-459
- 98) Leal SM, **Ott J** (1993) A bootstrap approach to estimating power for linkage heterogeneity. *Genet Epidemiol* 10, 465-470
- 99) Terwilliger JD, **Ott J** (1993) A novel polylocus method for linkage analysis using the lod score or affected sib-pair method. *Genet Epidemiol* 10, 477-482
- 100) Schork NJ, Boehnke M, Terwilliger JD, **Ott J** (1993) Two-trait-locus linkage analysis: a powerful strategy for mapping complex genetic traits. *Am J Hum Genet* 53, 1127-1136
- 101) Brzustowicz LM, Mérette C, Kleyn PW, Lehner T, Castilla LH, Penchaszadeh GK, Das K, Munsat TL, **Ott J**, Gilliam TG (1993) Assessment of nonallelic genetic heterogeneity of chronic (type II and III) spinal muscular atrophy. *Hum Hered* 43, 380-387
- 102) Spence MA, Bishop DT, Boehnke M, Elston RC, Falk C, Hodge SE, **Ott J**, Rice J, Merikangas K, Kupfer D (1993) Methodological issues in linkage analyses for psychiatric disorders: secular trends, assortative mating, bilineal pedigrees. Report of the MacArthur Foundation Network I Task Force on Methodological Issues. *Hum Hered* 43, 166-172
- 103) Petrukhin K, Fischer SG, Pirastu M, Tanzi RE, Chernov I, Devoto M, Brzustowicz LM, Cayanis E, Vitale E, Russo J, Matseoane D, Boukhalter B, Wasco W, Figus AL, Loudianos J, Cao A, Sternlieb I, Evgrafov G, Parano E, Pavone L, Warburton D, **Ott J**, Penchaszadeh G, Scheinberg IH, Gilliam TC (1993) Mapping, cloning, and genetic characterization of the region containing the Wilson disease gene. *Nature Genetics* 5, 338-343
- 104) Weeks DE, **Ott J**, Lathrop GM (1994) Detection of genetic interference: simulation studies and mouse data. *Genetics* 136, 1217-1226
- 105) Leal SM, **Ott J** (1994) A likelihood approach to calculating risk support intervals. *Am J Hum Genet* 54, 913-917
- 106) Crow TJ, Delisi LE, Lofthouse R, Poulter M, Lehner T, Bass N, Shah T, Walsh C, Boccio-Smith A, Shields G, **Ott J** (1994) An examination of linkage to schizophrenia and schizoaffective disorder to the pseudoautosomal region (Xp22.3). *Brit J Psychiat* 164, 159-164
- 107) Tienari PJ, Terwilliger JD, **Ott J**, Palo J, Peltonen L (1994) Two-locus linkage analysis in multiple sclerosis (MS). *Genomics* 19, 320-325
- 108) Baron M, Endicott J, Lerer B, Loth JE, Alexander JR, Simon R, Sharpe L, Gibbon M, Hasin D, Lilliston B, Schacht S, Blumenthal R, Alexander J, Verter A, Tubi N, Fieve RR, Gilliam TC, Lehner T, **Ott J** (1994) A pedigree series for mapping disease genes in bipolar affective disorder: sampling, assessment, and analytic considerations. *Psychiatric Genetics* 4, 43-55
- 109) Mérette C, Brzustowicz LM, Daniels RJ, Davies KE, Gilliam TC, Melki J, Munnich A, Pericak-Vance MA, Siddique T, Voosen B, Wirth B, **Ott J** (1994) An Investigation of Genetic Heterogeneity and Linkage Disequilibrium with 161 families with Spinal Muscular Atrophy. *Genomics* 21, 27-33
- 110) Chatkupt S, Speer MC, Ding Y, Thomas M, Stenroos ES, Dermody JJ, Koenigsberger R, **Ott J**, Johnson WG (1994) Linkage analysis of a candidate locus (HLA) in autosomal dominant sacral defect with anterior meningocele. *Am J Med Genet* 52, 1-4

- 111) Knowles JA, Shugart Y, Banerjee P, Gilliam TC, Lewis CA, Jacobson SG, **Ott J** (1994) Identification of a locus, distinct from RDS-peripherin, for autosomal recessive retinitis pigmentosa on chromosome 6p. *Hum Molec Genet* 3, 1401-1403
- 112) DeLisi LE, Devoto M, Lofthouse R, Poulter M, Smith A, Shields G, Bass N, Chen G, Vita A, Morgati C, **Ott J**, Crow TJ (1994) Search for linkage to schizophrenia on the X and Y chromosomes. *Am J Med Genet (Neuropsych Genet)* 54, 113-121
- 113) Sasse G, Müller H, Chakraborty R, **Ott J** (1994) Estimating the frequency of nonpaternity in Switzerland. *Hum Hered* 44, 337-343
- 114) Straub RE, Lehner T, Luo Y, Loth JE, Shao W, Sharpe L, Alexander JR, Das K, Simon R, Fieve RR, Lerer B, Endicott J, **Ott J**, Gilliam TC, Baron M (1994) A possible vulnerability locus for bipolar affective disorder on chromosome 21q22.3. *Nature Genetics* 8, 291-296
- 115) Gabizon R, Rosenman H, Meiner Z, Kahana I, Kahana E, Shugart Y, **Ott J**, Prusiner SB (1994) Mutation in codon 200 and polymorphism in codon 129 of the prion protein gene in Libyan jews with Creutzfeldt-Jakob disease. *Phil Trans R Soc Lond B* 343, 385-390
- 116) Karayiorgou M, Kasch L, Lasseter VK, Hwang J, Elango R, Bernardini DJ, Kimberland M, Babb R, Francomano CA, Wolyniec PS, Lamacz M, Nestadt G, Meyers D, **Ott J**, Childs B, Antonarakis S, Kazazian HH, Housman DE, Pulver AE (1994) Report from the Maryland epidemiology schizophrenia linkage study: No evidence for linkage between schizophrenia and a number of candidate and other genomic regions using a complex dominant model. *Am J Med Genet (Neuropsychiatric Genetics)* 54, 345-353
- 117) Pulver AE, Karayiorgou M, Wolyniec PS, Lasseter VK, Kasch L, Nestadt G, Antonarakis S, Housman D, Kazazian HH, Meyers D, **Ott J**, Lamacz M, Liang KY, Hanfelt J, Ulrich G, DeMarchi N, Ramu E, McHugh PR, Adler L, Thomas M, Carpenter WT, Manschreck T, Gordon CT, Kimberland M, Babb R, Puck J, Childs B (1994) Sequential strategy to identify a susceptibility gene for schizophrenia: Report of potential linkage on chromosome 22q12-q13.1: Part 1. *Am J Med Genet (Neuropsychiatric Genetics)* 54, 36-43
- 118) Pulver AE, Karayiorgou M, Lasseter VK, Wolyniec P, Kasch L, Antonarakis S, Housman D, Kazazian HH, Meyers D, Nestadt G, **Ott J**, Liang KY, Lamacz M, Thomas M, Childs B, Diehl SR, Wang S, Murphy B, Sun C, O'Neill FA, Nie L, Sham P, Burke J, Duke BW, Duke F, Kipps BR, Bray J, Hunt W, Shinkwin R, Nuallain MN, Su Y, MacLean CJ, Walsh D, Kendler KS, Gill M, Vallada H, Mant R, Asherson P, Collier D, Parfitt E, Roberts E, Nanko S, Walsh C, Daniels J, Murray R, McGuffin P, Owen M, Laurent C, Dumas JB, d'Amato T, Jay M, Martinez M, Campion D, Mallet J (1994) Follow-up of a report of a potential linkage for schizophrenia on chromosome 22q12-q13.1: Part 2. *Am J Med Genet (Neuropsychiatric Genetics)* 54, 44-55
- 119) Pulver AE, Lasseter VK, Kasch L, Wolyniec PS, Nestadt G, Blouin JL, Kimberland M, Babb R, Vourlis S, Chen H, Lalioti M, Morris MA, Karayiorgou M, **Ott J**, Meyers D, Antonarakis S, Housman D, Kazazian HH (1995) Schizophrenia: A genome scan targets chromosomes 3p and 8p as potential sites of susceptibility genes. *Am J Med Genetics (Neuropsychiatric Genetics)* 60, 252-260
- 120) **Ott J** (1995) Linkage analysis with biological markers. *Hum Hered* 45, 169-174
- 121) Lazzarini A, Stenroos ES, Lehner T, McKoy V, Gold B, McCormack MK, Reid CS, **Ott J**, Johnson WG (1995) Short tandem repeat polymorphism linkage studies in a New England family with X-linked mental retardation (MRX20). *Am J Med Genet* 57, 552-557
- 122) Fann CSJ, **Ott J** (1995) Parsimonious estimation of sex-specific map distances by stepwise maximum likelihood regression. *Genomics* 29, 571-575
- 123) Leal SM, **Ott J** (1995) Variability of genotype-specific penetrance probabilities in the calculation of risk support intervals. *Genet Epidemiol* 12, 859-862
- 124) Pauls DL, **Ott J**, Paul SM, Allen CR, Fann CSJ, Carulli JP, Falls KM, Bouthillier CA, Gravius TC, Keith TP, Egeland JA,

- Ginns EI (1995) Linkage analyses of chromosome 18 markers do not identify a major susceptibility locus for bipolar affective disorder in the Old Order Amish. *Am J Hum Genet* 57, 636-643
- 125) Ginns EI, **Ott J**, Egeland JA, Allen CR, Fann CSJ, Pauls DL, Weissenbach J, Carulli JP, Falls KM, Keith TP, Paul SM (1996) A genome-wide search for chromosomal loci linked to bipolar affective disorder in the Old Order Amish. *Nature Genetics* 12, 431-435
- 126) Schuster H, Wienker TF, Bähring S, Bilginturan N, Toka HR, Neitzel H, Jeschke E, Toka O, Gilbert D, Lowe A, **Ott J**, Haller H, Luft FC (1996) Severe autosomal dominant hypertension and brachydactyly in a unique Turkish kindred maps to human chromosome 12. *Nature Genetics* 13, 98-100
- 127) Mérette C, **Ott J** (1996) Estimating parental relationship in linkage analysis of recessive traits. *Am J Med Genet* 63, 386-391
- 128) Spotila LD, Caminis J, Devoto M, Shimoya K, Sereda L, **Ott J**, Whyte MP, Tenenhouse A, Prockop DJ (1996) Osteopenia in 37 members of seven families: Analysis based on a model of dominant inheritance. *Molecular Medicine* 2, 313-324
- 129) Fujita R, Bingham E, Forsythe P, McHenry C, Aita V, Navia BA, Dry K, Segal M, Devoto M, Bruns G, Wright AF, **Ott J**, Sieving PA, Swaroop A (1996) A recombination outside the BB deletion refines the localization of the X-linked retinitis pigmentosa locus RP3. *Am J Hum Genet* 59, 152-158
- 130) Brzustowicz LM, Gardner JP, Hopp L, Jeanclos E, **Ott J**, Yang XY, Fekete Z, Aviv A (1997) Linkage analysis using platelet-activating factor Ca^{2+} response in transformed lymphoblasts. *Hypertension* 29, 158-164
- 131) Müller-Myhsok B, Heiland H-J, Müller CR, Meng G, Grimm T, **Ott J** (1997) Mapping undetected mutations within a gene - evidence for two preferential regions in the DMD gene. *Hum Hered* 47, 61-65
- 132) Karayiorgou M, Altemus M, Galke BL, Goldman D, Murphy DL, **Ott J**, Gogos JA (1997) Genotype determining low catechol-O-methyltransferase activity as a risk factor for obsessive-compulsive disorder. *Proc Natl Acad Sci USA* 94, 4572-4575
- 133) Goring HH, **Ott J** (1997) Relationship estimation in affected sib pair analysis of late-onset diseases. *Eur J Hum Genet* 5, 69-77
- 134) Tsou HC, Teng DH, Ping XL, Brancolini V, Davis T, Hu R, Xie XX, Gruener AC, Schragger CA, Christiano AM, Eng C, Steck P, **Ott J**, Tavtigian SV, Peacocke M (1997) The role of MMAC1 mutations in early-onset breast cancer: causative in association with Cowden syndrome and excluded in BRCA1-negative cases. *Am J Hum Genet* 61, 1036-1043
- 135) Haghghi F, **Ott J** (1997) Estimating recessive disease allele frequency based on genetic maps. *Eur J Hum Genet* 5, 203-205
- 136) McGee TL, Devoto M, **Ott J**, Berson EL, Dryja TP (1997) Evidence that the penetrance of mutations at the RP11 locus causing dominant retinitis pigmentosa is influenced by a gene linked to the homologous RP11 allele. *Am J Hum Genet* 61, 1059-1066
- 137) **Ott J**, Rabinowitz D (1997) The effect of marker heterozygosity on the power to detect linkage disequilibrium. *Genetics* 147, 927-930
- 138) Leal SM and **Ott J** (1997) Analysis of two-locus traits under heterogeneity for recessive versus dominant inheritance. *Genet Epidemiol* 14, 1097-1100
- 139) Lucek PR, **Ott J** (1997) Neural network analysis of complex traits. *Genet Epidemiol* 14, 1101-1106
- 140) Karayiorgou M, Altemus M, Galke BL, Goldman D, Murphy DL, **Ott J**, Gogos JA (1997) Genotype determining low catechol-O-methyltransferase activity as a risk factor for obsessive-compulsive disorder. *Proc Natl Acad Sci USA* 94, 4572-4575

- 141) Banerjee P, Kleyn PW, Knowles JA, Lewis CA, Ross BM, Parano E, Kovats SG, Lee JJ, Penchaszadeh GK, **Ott J**, Jacobson SG, Gilliam TC (1998) TULP1 mutation in two extended Dominican kindreds with autosomal recessive Retinitis pigmentosa. *Nature Genetics* 18, 177-179
- 142) Plášilová M, Feráková E, Kádasi L, Poláková H, Gerinec A, **Ott J**, Ferák V (1998) Linkage of autosomal recessive primary congenital glaucoma to the GLC3A locus in Roms (Gypsies) from Slovakia. *Hum Hered* 48, 30-33
- 143) Banerjee P, Lewis CA, Kleyn PW, Shugart YY, Ross BM, Penchaszadeh GK, **Ott J**, Jacobson SG, Gilliam TC, Knowles JA (1998) Homozygosity and physical mapping of the autosomal recessive retinitis pigmentosa locus (RP14) on chromosome 6p21.3. *Genomics* 48, 171-177
- 144) Knowles JA, Rao PA, Cox-Matise T, Loth JE, de Jesus GM, Levine L, Das K, Penchaszadeh GK, Alexander JR, Lerer B, Endicott, **Ott J**, Gilliam TC, Baron M (1998) No Evidence for Significant Linkage between Bipolar Affective Disorder and Chromosome 18 Pericentromeric Markers in a Large Series of Multiplex Extended Pedigrees. *Am J Hum Genet* 62, 916-924
- 145) Ahmad W, Faiyaz ul Haque M, Brancolini V, Tsou HC, ul Haque S, Lam HM, Aita VM, Owen J, deBlanchiere M, Frank J, Cserhalmi-Friedman PB, Leask A, McGrath JA, Peacocke M, Ahmad M, **Ott J**, Christiano AM (1998) Alopecia universalis associated with a mutation in the human *hairless* gene. *Science* 279, 720-724
- 146) Ahmad W, Brancolini V, Faiyaz ul Haque M, Lam H, ul Haque S, Haider M, Maimon A, Aita VM, Owen J, Brown D, Zegarelli DJ, Ahmad M, **Ott J**, Christiano AM (1998) A Locus for Autosomal Recessive Hypodontia with Associated Dental Anomalies Maps to Chromosome 16q12.1. *Am J Hum Genet* 62, 987-991
- 147) Devoto M, Shimoya K, Caminis J, **Ott J**, Tenenhouse A, Whyte MP, Sereda L, Hall S, Considine E, Williams CJ, Tromp G, Kuivaniemi H, Ala-Kokko L, Prockop DJ, Spotila LD (1998) First-stage autosomal genome screen in extended pedigrees suggests genes predisposing to low bone mineral density on chromosomes 1p, 2p and 4q. *Eur J Hum Genet* 6, 151-157
- 148) Klein ML, Schultz DW, Edwards A, Matise TC, Rust K, Berselli CB, Trzuppek K, Weleber RG, **Ott J**, Wirtz MK, Acott TS (1998) Age-related macular dystrophy degeneration. Clinical features in a large family and linkage to chromosome 1q. *Arch Ophthalmol* 116, 1082-1088
- 149) Li W, Fann CSJ, **Ott J** (1998) Low-order polynomial trends of female-to-male map distance ratios along human chromosomes. *Hum Hered* 48, 266-270
- 150) Lucek P, Hanke J, Reich J, Solla SA, **Ott J** (1998) Multi-locus nonparametric linkage analysis of complex trait loci with neural networks. *Hum Hered* 48, 275-284
- 151) Simonic I, Gericke GS, **Ott J**, Weber JL (1998) Identification of Genetic Markers Associated with Gilles de la Tourette Syndrome in an Afrikaner Population. *Am J Hum Genet* 63, 839-846
- 152) Gieser L, Fujita R, Goring HH, **Ott J**, Hoffman DR, Cideciyan AV, Birch DG, Jacobson SG, Swaroop A (1998) A Novel Locus (RP24) for X-linked Retinitis Pigmentosa Maps to Xq26-27. *Am J Hum Genet* 63, 1439-1447
- 153) Gordon D, Heath SC, **Ott J** (1999) True pedigree errors more frequent than apparent errors for single nucleotide polymorphisms. *Hum Hered* 49, 65-70
- 154) **Ott J**, Rabinowitz D (1999) A principal-components approach based on heritability for combining phenotype information. *Hum Hered* 49, 106-111
- 155) Karayiorgou M, Sobin C, Blundell ML, Galke BL, Malinova L, Goldberg P, **Ott J**, Gogos JA (1999) Family-based association studies support a sexually dimorphic effect of COMT and MAOA on genetic susceptibility to obsessive-compulsive disorder. *Biol Psychiatry* 45, 1178-1189
- 156) Annunen S, Paasilta P, Lohiniva J, Perälä M, Pihlajamaa T, Karppinen J, Tervonen O, Kröger H, Lähde S, Vanharanta H, Ryhänen L, Goring HHH, **Ott J**, Prockop DJ, Ala-Kokko L (1999) An allele of COL9A2 associated with intervertebral disc disease. *Science* 285, 409-412

- 157) Frank J, Pignata C, Panteleyev AA, Prowse DM, Baden H, Weiner L, Gaetaniello L, Ahmad W, Pozzi N, Cserhalmi-Friedman PB, Aita VM, Uyttendaele H, Gordon D, **Ott J**, Brissette JL, Christiano AM (1999) Exposing the human nude phenotype. *Nature* 398, 473-474
- 158) Bhat A, Heath SC, **Ott J** (1999) Heterogeneity for multiple disease loci in linkage analysis. *Hum Hered* 49, 229-231
- 159) Lewis CA, Batlle IR, Batlle KG, Banerjee P, Cideciyan AV, Huang J, Aleman TS, Huang Y, **Ott J**, Gilliam TC, Knowles JA, Jacobson SG (1999) Tubby-like protein 1 homozygous splice-site mutation causes early-onset severe retinal degeneration. *Invest Ophthalmol Vis Sci* 40, 2106-2114
- 160) Le Saux O, Urban Z, Goring HH, Csiszar K, Pope FM, Richards A, Pasquali-Ronchetti I, Terry S, Bercovitch L, Lebowitz MG, Breuning M, van den Berg P, Kornet L, **Ott J**, de Jong PT, Bergen AA, Boyd CD (1999) Pseudoxanthoma elasticum maps to an 820-kb region of the p13.1 region of chromosome 16. *Genomics* 62, 1-10
- 161) Hoh JJ, **Ott J** (2000) Complex inheritance and localizing disease genes. *Hum Hered* 50, 85-89
- 162) Leal SM, **Ott J** (2000) Effects of stratification in the analysis of affected-sib-pair data: benefits and costs. *Am J Hum Genet* 66, 567-575
- 163) Stiburkova B, Majewski J, Sebesta I, Zhang W, **Ott J**, Kmoch S (2000) Familial juvenile hyperuricemic nephropathy: Localization of the gene on chromosome 16p11.2 and evidence for genetic heterogeneity. *Am J Hum Genet* 66, 1989-1994
- 164) Gordon D, Simonic I, **Ott J** (2000) Significant evidence for linkage disequilibrium over a 5-cM region among Afrikaners. *Genomics* 66, 87-92
- 165) Hu FZ, Preston RA, Post JC, White GJ, Kikuchi LW, Wang X, Leal SM, Levenstien MA, **Ott J**, Self TW, Allen G, Stiffler RS, McGraw C, Pulsifer-Anderson EA, Ehrlich GD (2000) Mapping of a gene for severe pediatric gastroesophageal reflux to chromosome 13q14. *JAMA* 284, 325-334
- 166) Hoh J, **Ott J** (2000) Scan statistics to scan markers for susceptibility genes. *Proc Natl Acad Sci USA* 97, 9615-9617
- 167) Mérette C, Brassard A, Potvin A, Bouvier H, Rousseau F, Emond C, Bissonnette L, Roy MA, Maziade M, **Ott J**, Caron C (2000) Significant Linkage for Tourette Syndrome in a Large French Canadian Family. *Am J Hum Genet* 67, 1008-1013
- 168) Hoh J, Wille A, Zee R, Cheng S, Reynolds R, Lindpaintner K, **Ott J** (2000) Selecting SNPs in two-stage analysis of disease association data: a model-free approach. *Ann Hum Genet* 64, 413-417
- 169) Gordon D, Leal SM, Heath SC, **Ott J** (2000) An analytic solution to single nucleotide polymorphism error-detection rates in nuclear families: implications for study design. *Pac Symp Biocomput* 2000, 663-674
- 170) Majewski J, **Ott J** (2000) GT repeats are associated with recombination on human chromosome 22. *Genome Res* 10, 1108-1114
- 171) **Ott J**, Hoh J (2000) Statistical Approaches to Gene Mapping. *Am J Hum Genet* 67, 289-294, Review
- 172) Shmulewitz D, Auerbach SB, Lehner T, Blundell ML, Winick JD, Youngman LD, Skilling V, Heath SC, **Ott J**, Stoffel M, Breslow JL, Friedman JM (2001) Epidemiology and factor analysis of obesity, type II diabetes, hypertension, and dyslipidemia (Syndrome X) on the island of Kosrae, Federated States of Micronesia. *Hum Hered* 51, 8-19
- 173) Hoh J, Heitjan DF, Mérette C, **Ott J** (2001) Ascertainment and anticipation in family studies. *Hum Hered* 51, 23-26
- 174) Simonic I, Nyholt DR, Gericke GS, Matsumoto N, Ledbetter DH, **Ott J**, Weber JL (2001) Further evidence for linkage of Gilles de la Tourette syndrome (GTS) susceptibility loci on chromosomes 2p11, 8q22 and 11q23-24 in South African Afrikaners. *Am J Med Genet (Neuropsychiatric Genetics)* 105, 163-167
- 175) Gordon D, **Ott J** (2001) Assessment and management of single nucleotide polymorphism genotype errors in genetic association analysis. *Pac Symp Biocomput* 2001, 18-29

- 176) **Ott J**, Hoh J (2001) Statistical multilocus methods for disequilibrium analysis in complex traits. *Hum Mutat* 17, 285-288
- 177) Paasilta P, Lohiniva J, Goring HH, Perala M, Raina SS, Karppinen J, Hakala M, Palm T, Kroger H, Kaitila I, Vanharanta H, **Ott J**, Ala-Kokko L (2001) Identification of a novel common genetic risk factor for lumbar disk disease. *JAMA* 285, 1843-1849
- 178) Lee MH, Gordon D, **Ott J**, Lu K, Ose L, Miettinen T, Gylling H, Stalenhoef AF, Pandya A, Hidaka H, Brewer B Jr, Kojima H, Sakuma N, Pegoraro R, Salen G, Patel SB (2001) Fine mapping of a gene responsible for regulating dietary cholesterol absorption; founder effects underlie cases of phytosterolaemia in multiple communities. *Eur J Hum Genet* 9, 375-384
- 179) **Ott J** (2001) Neural networks and disease association studies. *Am J Med Genet* 105, 60-61
- 180) Gordon D, Heath SC, Liu X, **Ott J** (2001) A transmission/disequilibrium test that allows for genotyping errors in the analysis of single-nucleotide polymorphism data. *Am J Hum Genet* 69, 371-380
- 181) Liu J, Nyholt DR, Magnussen P, Parano E, Pavone P, Geschwind D, Lord C, Iversen P, Hoh J, **Ott J**, Gilliam TC (2001) A genomewide screen for autism susceptibility loci. *Am J Hum Genet* 69, 327-340
- 182) Majewski J, Li H, **Ott J** (2001) The Ising model in physics and statistical genetics. *Am J Hum Genet* 69, 853-862
- 183) Hoh J, Wille A, **Ott J** (2001) Trimming, weighting, and grouping SNPs in human case-control association studies. *Genome Res* 11, 2115-2119
- 184) Hoh J, **Ott J** (2001) A train of thoughts on gene mapping. *Theor Popul Biol* 60, 149-153
- 185) Gordon D, Hoh J, Finch SJ, Levenstien MA, Edington J, Li W, Majewski J, **Ott J** (2001) Two approaches for consolidating results from genome scans of complex traits: selection methods and scan statistics. *Genet Epidemiol* 21 Suppl 1, S396-402
- 186) Emala CW, McQuitty CK, Eleff SM, Hopkins-Price P, Lawyer C, Hoh J, **Ott J**, Levine MA, Hirshman CA (2002) Asthma, allergy, and airway hyperresponsiveness are not linked to the beta(2)-Adrenoceptor gene. *Chest* 121, 722-731
- 187) Martinez-Mir A, Liu J, Gordon D, Weiner MS, Ahmad W, Fine JD, **Ott J**, Gilliam TC, Christiano AM (2002) EB Simplex Superficialis resulting from a mutation in the type VII collagen gene. *J Invest Dermatol* 118, 547-549
- 188) Martinez-Mir A, Gordon D, Horev L, Klapholz L, **Ott J**, Christiano AM, Zlotogorski A (2002) Multiple cutaneous and uterine leiomyomas: refinement of the genetic locus for multiple cutaneous and uterine leiomyomas on chromosome 1q42.3-43. *J Invest Dermatol* 118, 876-880
- 189) Wessman M, Kallela M, Kaunisto MA, Marttila P, Sobel E, Hartiala J, Oswell G, Leal SM, Papp JC, Hamalainen E, Broas P, Joslyn G, Hovatta I, Hiekkalinna T, Kaprio J, **Ott J**, Cantor RM, Zwart JA, Ilmavirta M, Havanka H, Farkkila M, Peltonen L, Palotie (2002) A susceptibility locus for migraine with aura, on chromosome 4q24. *Am J Hum Genet* 70, 652-662
- 190) Zee RYL, Hoh J, Cheng S, Reynolds R, Grow MA, Silbergleit A, Walker K, Steiner L, Zangenberg G, Fernandez-Ortiz A, Macaya C, Pintor E, Fernandez-Cruz A, **Ott J**, Lindpaintner K (2002) Multi-locus interactions predict risk for post-PTCA restenosis: an approach to the genetic analysis of common complex disease. *The Pharmacogenomics Journal* 2, 197-201
- 191) Hoh J, Jin S, Parrado T, Edington J, Levine AJ, **Ott J** (2002) The p53MH algorithm and its application in detecting p53-responsive genes. *Proc Natl Acad Sci USA* 99, 8467-8472
- 192) Han Z, Heath SC, Shmulewitz D, Li W, Auerbach SB, Blundell ML, Lehner T, **Ott J**, Stoffel M, Friedman JM, Breslow JL (2002) Candidate genes involved in cardiovascular risk factors by a family-based association study on the island of Kosrae, Federated States of Micronesia. *Am J Med Genet* 110, 234-242

- 193) Jain S, Tang X, Narayanan CS, Agarwal Y, Peterson SM, Brown CD, **Ott J**, Kumar A (2002) Angiotensinogen gene polymorphism at -217 affects basal promoter activity and is associated with hypertension in African-Americans. *J Biol Chem* 277, 36889-36896
- 194) **Ott J**, Hoh J (2002) Association studies for human trait genes. 2002 Proceedings of the American Statistical Association, Statistical Computing Section [CD-ROM], Alexandria, VA: American Statistical Association
- 195) Acuna G, Foernzler D, Leong D, Rabbia M, Smit R, Dorflinger E, Gasser R, Hoh J, **Ott J**, Borroni E, To Z, Thompson A, Li J, Hashimoto L, Lindpaintner K (2002) Pharmacogenetic analysis of adverse drug effect reveals genetic variant for susceptibility to liver toxicity. *Pharmacogenomics J* 2, 327-334
- 196) Gordon D, Finch SJ, Nothnagel M, **Ott J** (2002) Power and sample size calculations for case-control genetic association tests when errors are present: application to single nucleotide polymorphisms. *Hum Hered* 54, 22-33
- 197) Yang Y, **Ott J** (2002) Log-linear models for gene mapping with affected sib pair data. *Hum Hered* 53, 227-236
- 198) Majewski J, **Ott J** (2002) Distribution and characterization of regulatory elements in the human genome. *Genome Res* 12, 1827-1836
- 199) Stiburkova B, Majewski J, Hodanova K, Ondrova L, Jerabkova M, Zikanova M, Vyletal P, Sebesta I, Marinaki A, Simmonds A, Matthijs G, Fryns J-P, Torres R, Puig JG, **Ott J**, Knoch S (2003) Familial juvenile hyperuricaemic nephropathy (FJHN): linkage analysis in 15 families, physical and transcriptional characterisation of the FJHN critical region on chromosome 16p11.2 and the analysis of seven candidate genes. *Eur J Hum Genet* 11, 145-154
- 200) Gordon D, Levenstien MA, Finch SJ, **Ott J** (2003) Errors and linkage disequilibrium interact multiplicatively when computing sample sizes for genetic case-control association studies. *Pac Symp Biocomput* 2003;:490-499
- 201) Knappskog PM, Majewski J, Livneh A, Nilsen PTE, Bringsli JS, **Ott J**, Boman H (2003) Cold-induced sweating syndrome is caused by mutations in the CRLF1 gene. *Am J Hum Genet* 72, 375-383
- 202) Majewski J, **Ott J** (2003) Amino acid substitutions in the human genome: evolutionary implications of single nucleotide polymorphisms. *Gene* 305, 167-173
- 203) Garcia-Barcelo M, Sham MH, Lui VC, Chen BL, **Ott J**, Tam PK (2003) Association study of PHOX2B as a candidate gene for Hirschsprung's disease. *Gut* 52, 563-567
- 204) Gordon D, Corwin MB, Mellersh CS, Ostrander EA, **Ott J** (2003) Establishing appropriate genome-wide significance levels for canine linkage analyses. *J Hered* 94, 1-7
- 205) Liu J, Juo SH, Dewan A, Grunn A, Tong X, Brito M, Park N, Loth JE, Kanyas K, Lerer B, Endicott J, Penchaszadeh G, Knowles JA, **Ott J**, Gilliam TC, Baron M (2003) Evidence for a putative bipolar disorder locus on 2p13-16 and other potential loci on 4q31, 7q34, 8q13, 9q31, 10q21-24, 13q32, 14q21 and 17q11-12. *Mol Psychiatry* 8, 333-342
- 206) Hoh J, Matsuda F, Peng X, Markovic D, Lathrop M, **Ott J** (2003) SNP haplotype tagging from DNA pools of two individuals. *BMC Bioinformatics* 4, 14
- 207) Kljuic A, Bazzi H, Sundberg JP, Martinez-Mir A, O'Shaughnessy R, Mahoney MG, Levy M, Montagutelli X, Ahmad W, Aita VM, Gordon D, Uitto J, Whiting D, **Ott J**, Fischer S, Gilliam TC, Jahoda CA, Morris RJ, Panteleyev AA, Nguyen VT, Christiano AM (2003) Desmoglein 4 in hair follicle differentiation and epidermal adhesion. Evidence from inherited hypotrichosis and acquired pemphigus vulgaris. *Cell* 113, 249-260
- 208) Yang Y, Zhang J, Hoh J, Matsuda F, Xu P, Lathrop M, **Ott J** (2003) Efficiency of single-nucleotide polymorphism haplotype estimation from pooled DNA. *Proc Natl Acad Sci USA* 100, 7225-7230
- 209) Yang Y, Hoh J, Broger C, Neeb M, Edington J, Lindpaintner K, **Ott J** (2003) Statistical methods for analyzing microarray feature data with replications. *J Comput Biol* 10, 157-169
- 210) **Ott J**, Hoh J (2003) Set association analysis of SNP case-control and microarray data. *J Comput Biol* 10, 569-574

- 211) Hoh J, **Ott J** (2003) Mathematical multi-locus approaches to localizing complex human trait genes. *Nat Rev Genet* 4, 701-709. Review
- 212) Martinez-Mir A, Zlotogorski A, **Ott J**, Gordon D, Christiano AM (2003) Genetic linkage studies in Alopecia Areata. *J Investig Dermatol Symp Proc* 8, 199-203
- 213) Helms C, Cao L, Krueger JG, Wijsman EM, Chamian F, Gordon D, Heffernan M, Daw JA, Robarge J, **Ott J**, Kwok PY, Menter A, Bowcock AM (2003) A putative RUNX1 binding site variant between SLC9A3R1 and NAT9 is associated with susceptibility to psoriasis. *Nat Genet* 35, 349-356
- 214) Wille A, Hoh J, **Ott J** (2003) Sum statistics for the joint detection of multiple disease loci in case-control association studies with SNP markers. *Genet Epidemiol* 25, 350-359
- 215) Louie E, **Ott J**, Majewski J (2003) Nucleotide frequency variation across human genes. *Genome Res* 13, 2594-2601
- 216) Majewski J, Schultz DW, Weleber RG, Schain MB, Edwards AO, Matisse TC, Acott TS, **Ott J**, Klein ML (2003) Age-related macular degeneration — a genome scan in extended families. *Am J Hum Genet* 73, 540-550
- 217) Schultz DW, Klein ML, Humpert A, Majewski J, Schain M, Weleber RG, **Ott J**, Acott TS (2003) Lack of an association of apolipoprotein E gene polymorphisms with familial age-related macular degeneration. *Arch Ophthalmol* 121, 679-683
- 218) Nojonen-Hietala N, Kyllonen E, Mannikko M, Ilkko E, Karppinen J, **Ott J**, Ala-Kokko L (2003) Sequence variations in the collagen IX and XI genes are associated with degenerative lumbar spinal stenosis. *Ann Rheum Dis* 62, 1208-1214
- 219) Levenstien MA, Yang Y, **Ott J** (2003) Statistical significance for hierarchical clustering in genetic association and microarray expression studies. *BMC Bioinformatics* 4(1), 62
- 220) DeAngelis MM, Lane AM, Shah CP, **Ott J**, Dryja TP, Miller JW (2004) Extremely discordant sib-pair study design to determine risk factors for neovascular age-related macular degeneration. *Arch Ophthalmol* 122, 575-580
- 221) Bart G, Heilig M, LaForge KS, Pollak L, Leal SM, **Ott J**, Kreek MJ (2004) Substantial attributable risk related to a functional mu-opioid receptor gene polymorphism in association with heroin addiction in central Sweden. *Mol Psychiatry* 9, 547-549
- 222) Kang SJ, Gordon D, Brown AM, **Ott J**, Finch SJ (2004) Tradeoff between no-call reduction in genotyping error rate and loss of sample size for genetic case/control association studies. *Pac Symp Biocomput* 2004;:116-127
- 223) Gordon D, Haynes C, Johnnidis C, Patel SB, Bowcock AM, **Ott J** (2004) A transmission disequilibrium test for general pedigrees that is robust to the presence of random genotyping errors and any number of untyped parents. *Eur J Hum Genet* 12, 752-761
- 224) Kim H, Klein R, Majewski J, **Ott J** (2004) Estimating rates of alternative splicing in mammals and invertebrates. *Nat Genet* 36, 915-916
- 225) Hoh J, **Ott J** (2004) Genetic dissection of diseases: design and methods. *Curr Opin Genet Dev* 14, 229-232
- 226) DeWan A, **Ott J** (2004) Reanalysis of a genome scan for schizophrenia loci using multigenic methods. *Hum Hered* 57, 191-194
- 227) Bart G, Kreek MJ, **Ott J**, Laforge KS, Proudnikov D, Pollak L, Heilig M (2004) Increased attributable risk related to a functional mu-opioid receptor gene polymorphism in association with alcohol dependence in Central Sweden. *Neuropsychopharmacology* 2004, 1-6
- 228) Yuferov V, Fussell D, LaForge KS, Nielsen DA, Gordon D, Ho A, Leal SM, **Ott J**, Kreek MJ (2004) Redefinition of the human kappa opioid receptor gene (OPRK1) structure and association of haplotypes with opiate addiction. *Pharmacogenetics* 14, 793-804

- 229) Sherriff A, **Ott J**; ALSPAC Study Team (2004) Artificial neural networks as statistical tools in epidemiological studies: analysis of risk factors for early infant wheeze. *Paediatr Perinat Epidemiol* 18, 456-463
- 230) **Ott J** (2004) Issues in association analysis: error control in case-control association studies for disease gene discovery. *Hum Hered* 58, 171-174
- 231) Noponen-Hietala N, Virtanen I, Karttunen R, Schwenke S, Jakkula E, Li H, Merikivi R, Barral S, **Ott J**, Karppinen J, Ala-Kokko L (2005) Genetic variations in IL6 associate with intervertebral disc disease characterized by sciatica. *Pain* 114, 186-194
- 232) Klein RJ, Zeiss C, Chew EY, Tsai JY, Sackler RS, Haynes C, Henning AK, Sangiovanni JP, Mane SM, Mayne ST, Bracken MB, Ferris FL, **Ott J**, Barnstable C, Hoh J (2005) Complement Factor H Polymorphism in Age-Related Macular Degeneration. *Science* 308, 385-389
- 233) Xu J, Yang Y, **Ott J** (2005) Survival analysis of microarray expression data by transformation models. *Comput Biol Chem* 29, 91-94
- 234) Zarepari S, Branham KE, Li M, Shah S, Klein RJ, **Ott J**, Hoh J, Abecasis GR, Swaroop A (2005) Strong Association of the Y402H Variant in Complement Factor H at 1q32 with Susceptibility to Age-Related Macular Degeneration. *Am J Hum Genet* 77, 149-153
- 235) Francis PJ, Schultz DW, Gregory AM, Schain MB, Barra R, Majewski J, **Ott J**, Acott T, Weleber RG, Klein ML (2005) Genetic and phenotypic heterogeneity in pattern dystrophy. *Br J Ophthalmol* 89, 1115-1119
- 236) Levrán O, Attwooll C, Henry RT, Milton KL, Neveling K, Rio P, Batish SD, Kalb R, Velleuer E, Barral S, **Ott J**, Petrini J, Schindler D, Hanenberg H, Auerbach AD (2005) The BRCA1-interacting helicase BRIP1 is deficient in Fanconi anemia. *Nat Genet* 37, 931-933
- 237) Markovic D, Tang X, Guruju M, Levenstien MA, Hoh J, Kumar A, **Ott J** (2005) Association of Angiotensinogen Gene Polymorphisms with Essential Hypertension in African-Americans and Caucasians. *Hum Hered* 60, 89-96
- 238) Bleiber G, May M, Martinez R, Meylan P, **Ott J**, Beckmann JS, Telenti A; Swiss HIV Cohort Study (2005) Use of a combined ex vivo/in vivo population approach for screening of human genes involved in the human immunodeficiency virus type 1 life cycle for variants influencing disease progression. *J Virol* 79, 12674-12680
- 239) Jim JJ, Noponen-Hietala N, Cheung KM, **Ott J**, Karppinen J, Sahraravand A, Luk KD, Yip SP, Sham PC, Song YQ, Leong JC, Cheah KS, Ala-Kokko L, Chan D (2005) The TRP2 allele of COL9A2 is an age-dependent risk factor for the development and severity of intervertebral disc degeneration. *Spine* 30, 2735-2742
- 240) Helms C, Saccone NL, Cao L, Daw JA, Cao K, Hsu TM, Taillon-Miller P, Duan S, Gordon D, Pierce B, **Ott J**, Rice J, Fernandez-Vina MA, Kwok PY, Menter A, Bowcock AM (2005) Localization of PSORS1 to a haplotype block harboring HLA-C and distinct from corneodesmosin and HCR. *Hum Genet* 118, 466-476
- 241) Proudnikov D, LaForge KS, Hofflich H, Levenstien M, Gordon D, Barral S, **Ott J**, Kreek MJ (2006) Association analysis of polymorphisms in serotonin 1B receptor (HTR1B) gene with heroin addiction: a comparison of molecular and statistically estimated haplotypes. *Pharmacogenet Genomics* 16, 25-36
- 242) Haider AS, Peters SB, Kaporis H, Cardinale I, Fei J, **Ott J**, Blumenberg M, Bowcock AM, Krueger JG, Carucci JA (2006) Genomic analysis defines a cancer-specific gene expression signature for human squamous cell carcinoma and distinguishes malignant hyperproliferation from benign hyperplasia. *J Invest Dermatol* 126, 869-881
- 243) Cheung KM, Chan D, Karppinen J, Chen Y, Jim JJ, Yip SP, **Ott J**, Wong KK, Sham P, Luk KD, Cheah KS, Leong JC, Song YQ (2006) Association of the Taq I allele in vitamin D receptor with degenerative disc disease and disc bulge in a Chinese population. *Spine* 31, 1143-1148
- 244) Tomic M, **Ott J**, Barral S, Bovet P, Deppen P, Gheorghita F, Matthey M-L, Parnas J, Preisig M, Saraga M, Solida A, Timm S, Wang AG, Werge T, Cuenod M, Do KQ (2006) Schizophrenia and Oxidative Stress: Glutamate Cysteine

Ligase Modifier as a Susceptibility Gene. *Am J Hum Genet* **79**, 586-592

- 245) Levenstien MA, **Ott J**, Gordon D (2006) Are Molecular Haplotypes Worth the Time and Expense? A Cost-Effective Method for Applying Molecular Haplotypes. *PLoS Genet* **18**;2(8)
- 246) **Ott J** (2006) Unknown parental phase--lod score versus information. A comment to Prof. Edwards' note. *Ann Hum Genet* **70**, 974
- 247) Barral S, Francis PJ, Schultz DW, Schain MB, Haynes C, Majewski J, Ott J, Acott T, Weleber RG, Klein ML. Expanded genome scan in extended families with age-related macular degeneration. *Invest Ophthalmol Vis Sci*. 2006 Dec;47(12):5453-5459. PMID: 17122136
- 248) Martinez-Mir A, Zlotogorski A, Gordon D, Petukhova L, Mo J, Gilliam TC, Londono D, Haynes C, Ott J, Hordinsky M, Nanova K, Norris D, Price V, Duvic M, Christiano AM. Genomewide scan for linkage reveals evidence of several susceptibility loci for alopecia areata. *Am J Hum Genet*. 2007 Feb;80(2):316-328. PMID: 17236136
- 249) Virtanen IM, Noponen N, Barral S, Karppinen J, Li H, Vuoristo M, Niinimäki J, Ott J, Ala-Kokko L, Mannikko M. A Putative Susceptibility Locus on Chromosome 21q for Lumbar Disc Disease (LDD) in the Finnish Population. *J Bone Miner Res*. 2007 May;22(5):701-7. PMID: 17266399
- 250) Xu MQ, St Clair D, Ott J, Feng GY, He L. Brain-derived neurotrophic factor gene C-270T and Val66Met functional polymorphisms and risk of schizophrenia: A moderate-scale population-based study and meta-analysis. *Schizophr Res*. 2007 Mar;91(1-3):6-13. PMID: 17289348
- 251) Francis PJ, George S, Schultz DW, Rosner B, Hamon S, Ott J, Weleber RG, Klein ML, Seddon JM. The LOC387715 Gene, Smoking, Body Mass Index, Environmental Associations with Advanced Age-Related Macular Degeneration. *Hum Hered*. 2007;63(3-4):212-218. PMID: 17347568
- 252) Kankova K, Stejskalova A, Pacal L, Tschoplova S, Hertlova M, Krusova D, Izakovicova-Holla L, Beranek M, Vasku A, Barral S, Ott J. Genetic risk factors for diabetic nephropathy on chromosomes 6p and 7q identified by the set-association approach. *Diabetologia*. 2007 May;50(5):990-999. PMID: 17345061
- 253) Virtanen IM, Karppinen J, Taimela S, Ott J, Barral S, Kaikkonen K, Heikkilä O, Mutanen P, Noponen N, Mannikko M, Tervonen O, Natri A, Ala-Kokko L. Occupational and genetic risk factors associated with intervertebral disc disease. *Spine*. 2007 May 1;32(10):1129-34. PMID: 17471097
- 254) Williams TJ, Laforge KS, Gordon D, Bart G, Kellogg S, Ott J, Kreek MJ. Prodynorphin gene promoter repeat associated with cocaine/alcohol codependence. *Addict Biol*. 2007 Sep;12(3-4):496-502. PMID: 17559549
- 255) Perez CA, Ott J, Mays DJ, Pietenpol JA. p63 consensus DNA-binding site: identification, analysis and application into a p63MH algorithm. *Oncogene*. 2007 Nov 15;26(52):7363-70. PMID: 17563751
- 256) DeAngelis MM, Ji F, Kim IK, Adams S, Capone A Jr, Ott J, Miller JW, Dryja TP. Cigarette smoking, CFH, APOE, ELOVL4, and risk of neovascular age-related macular degeneration. *Arch Ophthalmol*. 2007 Jan;125(1):49-54. PMID: 17210851
- 257) Francis PJ, Schultz DW, Hamon S, Ott J, Weleber RG, Klein ML. Haplotypes in the Complement Factor H (CFH) Gene: Associations with Drusen and Advanced Age-Related Macular Degeneration. *PLoS ONE*. 2007 Nov 28;2(11):e1197. PMID: 18043728
- 258) Haider AS, Cohen J, Fei J, Zaba LC, Cardinale I, Toyoko K, Ott J, Krueger JG. Insights into Gene Modulation by Therapeutic TNF and IFN γ Antibodies: TNF Regulates IFN γ Production by T Cells and TNF-Regulated Genes Linked to Psoriasis Transcriptome. *J Invest Dermatol*. 2008 Mar;128(3):655-66. PMID: 17928893
- 259) DeAngelis MM, Ji F, Adams S, Morrison MA, Harring AJ, Sweeney MO, Capone A Jr, Miller JW, Dryja TP, Ott J, Kim IK. Alleles in the HtrA Serine Peptidase 1 Gene Alter the Risk of Neovascular Age-Related Macular Degeneration. *Ophthalmology*. 2008 Jul;115(7):1209-1215.e7. PMID: 18164066

- 260) Nielsen DA, Barral S, Proudnikov D, Kellogg S, Ho A, Ott J, Kreek MJ. TPH2 and TPH1: Association of Variants and Interactions with Heroin Addiction. *Behav Genet.* 2008 Mar;38(2):133-50. PMID: 18181017
- 261) Nielsen DA, Ji F, Yuferov V, Ho A, Chen A, Levran O, Ott J, Kreek MJ. Genotype patterns that contribute to increased risk for or protection from developing heroin addiction. *Mol Psychiatry.* 2008, 13(4):417-428. PMID: 18195715
- 262) Oosterhuis BE, Laforge KS, Proudnikov D, Ho A, Nielsen DA, Gianotti R, Barral S, Gordon D, Leal SM, Ott J, Kreek MJ. Catechol-O-methyltransferase (COMT) gene variants: Possible association of the Val158Met variant with opiate addiction in hispanic women. *Am J Med Genet B Neuropsychiatr Genet.* 2008 Sep 5;147B(6):793-8. PMID: 18270997
- 263) Proudnikov D, Hamon S, Ott J, Kreek MJ. Association of polymorphisms in the melanocortin receptor type 2 (MC2R, ACTH receptor) gene with heroin addiction. *Neurosci Lett.* 2008 Apr 25;435(3):234-9. PMID: 18359160
- 264) Bacolod MD, Schemmann GS, Wang S, Shattock R, Giardina SF, Zeng Z, Shia J, Stengel RF, Gerry N, Hoh J, Kirchhoff T, Gold B, Christman MF, Offit K, Gerald WL, Notterman DA, Ott J, Paty PB, Barany F. The signatures of autozygosity among patients with colorectal cancer. *Cancer Res.* 2008 Apr 15;68(8):2610-21. PMID: 18375840
- 265) Klein ML, Francis PJ, Rosner B, Reynolds R, Hamon SC, Schultz DW, Ott J, Seddon JM. CFH and LOC387715/ARMS2 Genotypes and Treatment with Antioxidants and Zinc for Age-Related Macular Degeneration. *Ophthalmology.* 2008 Jun;115(6):1019-25. PMID: 18423869
- 266) Levran O, O'Hara K, Peles E, Li D, Barral S, Ray B, Borg L, Ott J, Adelson M, Kreek MJ. ABCB1 (MDR1) Genetic Variants are Associated with Methadone Doses Required for Effective Treatment of Heroin Dependence. *Hum Mol Genet.* 2008 Jul 15;17(14):2219-27. PMID: 18424454
- 267) Levran O, Londono D, O'Hara K, Nielsen DA, Peles E, Rotrosen J, Casadonte P, Linzy S, Randesi M, Ott J, Adelson M, Kreek MJ. Genetic susceptibility to heroin addiction; a candidate-gene association study. *Genes Brain Behav.* 2008 Oct;7(7):720-9. PMID: 18518925
- 268) Francis PJ, Appukuttan B, Simmons E, Landauer N, Stoddard J, Hamon S, Ott J, Ferguson B, Klein M, Stout JT, Neuringer M. Rhesus monkeys and humans share common susceptibility genes for age-related macular disease. *Hum Mol Genet.* 2008 Sep 1;17(17):2673-80. PMID: 18535016
- 269) Zhang Q, Wang S, Ott J. Combining identity by descent and association in genetic case-control studies. *BMC Genet.* 2008 Jul 5;9:42. PMID: 18601744
- 270) Matthews AG, Haynes C, Liu C, Ott J. Collapsing SNP genotypes in case-control genome-wide association studies increases the type I error rate and power. *Stat Appl Genet Mol Biol.* 2008;7:Article23. PMID: 18673292
- 271) Kim IK, Ji F, Morrison MA, Adams S, Zhang Q, Lane AM, Capone A, Dryja TP, Ott J, Miller JW, DeAngelis MM. Comprehensive analysis of CRP, CFH Y402H and environmental risk factors on risk of neovascular age-related macular degeneration. *Mol Vis.* 2008 Aug 11;14:1487-95. PMID: 18704199
- 272) Xu J, Yang Y, Ying Z, Ott J. Testing linkage disequilibrium from pooled DNA: a contingency table perspective. *Stat Med.* 2008 Dec 10;27(28):5801-15. PMID: 18712782
- 273) Cheng YW, Pincas H, Bacolod MD, Schemmann G, Giardina SF, Huang J, Barral S, Idrees K, Khan SA, Zeng Z, Rosenberg S, Notterman DA, Ott J, Paty P, Barany F. CpG island methylator phenotype associates with low-degree chromosomal abnormalities in colorectal cancer. *Clin Cancer Res.* 2008 Oct 1;14(19):6005-13. PMID: 18829479
- 274) Yuferov V, Ji F, Nielsen DA, Levran O, Ho A, Morgello S, Shi R, Ott J, Kreek MJ. A Functional Haplotype Implicated in Vulnerability to Develop Cocaine Dependence is Associated with Reduced PDYN Expression in Human Brain. *Neuropsychopharmacology.* 2009 Apr;34(5):1185-97. PMID: 18923396

- 275) Nielsen DA, Yuferov V, Hamon S, Jackson C, Ho A, Ott J, Kreek MJ. Increased OPRM1 DNA Methylation in Lymphocytes of Methadone-Maintained Former Heroin Addicts. *Neuropsychopharmacology*. 2009 Mar;34(4):867-73. PMID: 18650805
- 276) Yang Y, He C, Ott J. Testing Association with Interactions by Partitioning Chi-Squares. *Ann Hum Genet*. 2009 Jan;73(1):109-17. PMID: 18798840
- 277) Francis PJ, Hamon SC, Ott J, Weleber RG, Klein ML. Polymorphisms in C2, CFB and C3 are associated with progression to Advanced Age-Related Macular Degeneration associated with visual loss. *J Med Genet*. 2009 May;46(5):300-7. PMID: 19015224
- 278) Wang S, Haynes C, Barany F, Ott J. Genome-wide autozygosity mapping in human populations. *Genet Epidemiol*. 2009 Feb;33(2):172-80. PMID: 18814273
- 279) Shaaban S, Matsuo T, Fujiwara H, Itoshima E, Furuse T, Hasebe S, Zhang Q, Ott J, Ohtsuki H. Chromosomes 4q28.3 and 7q31.2 as new susceptibility loci for comitant strabismus. *Invest Ophthalmol Vis Sci*. 2009 Feb;50(2):654-61. PMID: 18824738
- 280) Yang Y, He C, Ott J. Testing association with interactions by partitioning chi-squares. *Ann Hum Genet*. 2009 Jan;73(Pt 1):109-17
- 281) He C, Hamon S, Li D, Barral-Rodriguez S, Ott J; Diabetes Genetics Consortium. MHC fine mapping of human type 1 diabetes using the T1DGC data. *Diabetes Obes Metab*. 2009 Feb;11 Suppl 1:53-59. PMID: 19143815
- 282) Long Q, Zhang Q, Ott J. Detecting disease-associated genotype patterns. *BMC Bioinformatics*. 2009 Jan 30;10 Suppl 1:S75. PMID: 19208180
- 283) Levran O, Londono D, O'Hara K, Randesi M, Rotrosen J, Casadonte P, Linzy S, Ott J, Adelson M, Kreek MJ. Heroin addiction in African Americans: a hypothesis-driven association study. *Genes Brain Behav*. 2009 Jul;8(5):531-40. PMID: 19500151
- 284) Risch N, Herrell R, Lehner T, Liang KY, Eaves L, Hoh J, Griem A, Kovacs M, Ott J, Merikangas KR. Interaction between the serotonin transporter gene (5-HTTLPR), stressful life events, and risk of depression: a meta-analysis. *JAMA*. 2009 Jun 17;301(23):2462-71. PMID: 19531786
- 285) Matthews AG, Li J, He C, Ott J, Andrade M. Adjusting for HLA-DRbeta1 in a genome-wide association analysis of rheumatoid arthritis and related biomarkers. *BMC Proc*. 2009 Dec 15;3 Suppl 7:S12. PMID: 20017985
- 286) Nash GM, Gimbel M, Cohen AM, Zeng ZS, Ndubuisi MI, Nathanson DR, Ott J, Barany F, Paty PB. KRAS mutation and microsatellite instability: two genetic markers of early tumor development that influence the prognosis of colorectal cancer. *Ann Surg Oncol*. 2010 Feb;17(2):416-24. PMID: 19813061
- 287) Proudnikov D, Krosiak T, Sipe JC, Randesi M, Li D, Hamon S, Ho A, Ott J, Kreek MJ. Association of polymorphisms of the cannabinoid receptor (CNR1) and fatty acid amide hydrolase (FAAH) genes with heroin addiction: impact of long repeats of CNR1. *Pharmacogenomics J*. 2010 Jun;10(3):232-42. PMID: 20010914
- 288) Briant JA, Nielsen DA, Proudnikov D, Londono D, Ho A, Ott J, Kreek MJ. Evidence for association of two variants of the nociceptin/orphanin FQ receptor gene OPRL1 with vulnerability to develop opiate addiction in Caucasians. *Psychiatr Genet*. 2010 Apr;20(2):65-72. PMID: 20032820
- 289) Nielsen DA, Hamon S, Yuferov V, Jackson C, Ho A, Ott J, Kreek MJ. Ethnic diversity of DNA methylation in the OPRM1 promoter region in lymphocytes of heroin addicts. *Hum Genet*. 2010 Jun;127(6):639-49. PMID: 20237803
- 290) Klein ML, Ferris FL 3rd, Francis PJ, Lindblad AS, Chew EY, Hamon SC, Ott J. Progression of Geographic Atrophy and Genotype in Age-Related Macular Degeneration. *Ophthalmology*. 2010 Apr 8;117:1554-1559. PMID: 20381870

- 291) Wang G, Yang Y, Ott J. Genome-wide conditional search for epistatic disease-predisposing variants in human association studies. *Hum Hered.* 2010 Apr 23;70(1):34-41. PMID: 20413980
- 292) Kramer PL, Xu H, Woltjer RL, Westaway SK, Clark D, Erten-Lyons D, Kaye JA, Welsh-Bohmer KA, Troncoso JC, Markesbery WR, Petersen RC, Turner RS, Kukull WA, Bennett DA, Galasko D, Morris JC, Ott J. Alzheimer disease pathology in cognitively healthy elderly: A genome-wide study. *Neurobiol Aging.* 2010 May 6. PMID: 20452100
- 293) Wu C, Xu B, Yuan P, Ott J, Guan Y, Liu Y, Liu Z, Shen Y, Yu D, Lin D. Genome-wide examination of genetic variants associated with response to platinum-based chemotherapy in patients with small-cell lung cancer. *Pharmacogenet Genomics.* 2010 Jun;20(6):389-95. PMID: 20463552
- 294) Nielsen DA, Ji F, Yuferov V, Ho A, He C, Ott J, Kreek MJ. Genome-wide association study identifies genes that may contribute to risk for developing heroin addiction. *Psychiatr Genet.* 2010 Oct;20(5):207-14. PMID: 20520587
- 295) Ott J, Macciardi F, Shen Y, Carta MG, Murru A, Triunfo R, Robledo R, Rinaldi A, Contu L, Siniscalco M. Pilot Study on Schizophrenia in Sardinia. *Hum Hered.* 2010 Jun 17;70(2):92-96. PMID: 20558996
- 296) Cho S, Kim K, Kim YJ, Lee JK, Cho YS, Lee JY, Han BG, Kim H, Ott J, Park T. Joint identification of multiple genetic variants via elastic-net variable selection in a genome-wide association analysis. *Ann Hum Genet.* 2010 Sep 1;74(5):416-28. PMID: 20642809
- 297) Shen Y, Liu Z, Ott J. Systematic Removal of Outliers to Reduce Heterogeneity in Case-Control Association Studies. *Hum Hered.* 2010 Oct 6;70(4):227-231. PMID: 20924194
- 298) Wu L, Xi B, Zhang M, Shen Y, Zhao X, Cheng H, Hou D, Sun D, Ott J, Wang X, Mi J. Associations of six single nucleotide polymorphisms in obesity-related genes with body mass index and risk of obesity in the Chinese children. *Diabetes.* 2010 Sep 15. PMID: 20843981
- 299) Korvala J, Hartikka H, Pihlajamaki H, Solovieva S, Ruohola JP, Sahi T, Barral S, Ott J, Ala-Kokko L, Mannikko M. Genetic Predisposition For Femoral Neck Stress Fractures In Military Conscripts. *BMC Genet.* 2010 Oct 21;11(1):95. PMID: 20961463
- 300) Lee KT, Byun MJ, Kang KS, Park EW, Lee SH, Cho S, Kim H, Kim KW, Lee T, Park JE, Park W, Shin D, Park HS, Jeon JT, Choi BH, Jang GW, Choi SH, Kim DW, Lim D, Park HS, Park MR, Ott J, Schook LB, Kim TH, Kim H. Neuronal genes for subcutaneous fat thickness in human and pig are identified by local genomic sequencing and combined SNP association study. *PLoS One.* 2011 Feb 2;6(2):e16356. PMID: 21311593
- 301) Ott J. Writings on genetic linkage in the Annals. *Ann Hum Genet.* 2011 May;75(3):344-7. PMID: 21488851
- 302) Ott J. William Allan Award Address: On the role and soul of a statistical geneticist. *Am J Hum Genet.* 2011 Mar 11;88(3):264-8. PMID: 21516615
- 303) Ott J, Kamatani Y, Lathrop M. Family-based designs for genome-wide association studies. *Nat Rev Genet.* 2011 Jun 1;12(7):465-74. doi: 10.1038/nrg2989. PMID: 21629274
- 304) Ott J, Wang J. Multiple phenotypes in genome-wide genetic mapping studies. *Protein Cell.* 2011 Jul;2(7):519-22. PMID: 21647556
- 305) Schäffer AA, Lemire M, Ott J, Lathrop GM, Weeks DE. Coordinated conditional simulation with SLINK and SUP of many markers linked or associated to a trait in large pedigrees. *Hum Hered.* 2011;71(2):126-34. PMID: 21734403
- 306) Liu Z, Shen Y, Ott J. Multilocus association mapping using generalized ridge logistic regression. *BMC Bioinformatics.* 2011 Sep 29;12:384. PMID: 21958005
- 307) Zhang L, Chang S, Li Z, Zhang K, Du Y, Ott J, Wang J. ADHDgene: a genetic database for attention deficit hyperactivity disorder. *Nucleic Acids Res.* 2012 Jan;40(Database issue):D1003-9. PMID: 22080511
- 308) Proudnikov D, Randesi M, Levran O, Crystal H, Dorn M, Ott J, Ho A, Kreek MJ. Association of polymorphisms of

- the mu opioid receptor gene with severity of HIV infection and response to HIV treatment. *J Infect Dis.* 2012 Jun;205(11):1745-56. PMID: 22457278
- 309) Barral S, Bird T, Goate A, Farlow MR, Diaz-Arrastia R, Bennett DA, Graff-Radford N, Boeve BF, Sweet RA, Stern Y, Wilson RS, Foroud T, Ott J, Mayeux R; National Institute on Aging Late-Onset Alzheimer's Disease Genetics Study. Genotype patterns at PICALM, CR1, BIN1, CLU, and APOE genes are associated with episodic memory. *Neurology.* 2012 May 8;78(19):1464-71. PMID: 22539578
- 310) Ott J, Sun D. Multilocus association analysis under polygenic models. *Int J Data Min Bioinform.* 2012; 6(5):482-489. PMID: 23155777
- 311) Shen Y, Liu Z, Ott J. Support vector machines with L1 penalty for detecting gene-gene interactions. *Int J Data Min Bioinform.* 2012; 6(5):463-470. PMID: 23155775
- 312) Park T, Ott J. Data mining for high throughput data from genome-wide association studies. *Int J Data Min Bioinform.* 2012;6(5):461-2. PMID: 23155774
- 313) Guo L, Zhang W, Chang S, Zhang L, Ott J, Wang J. MK4MDD: A Multi-Level Knowledge Base and Analysis Platform for Major Depressive Disorder. *PLoS One.* 2012;7(10):e46335. doi: 10.1371/journal.pone.0046335. PMID: 23071556
- 314) Ott J, Hoh J. Scan statistics in human gene mapping. *Am J Hum Genet.* 2012 Nov 2;91(5):970. PMID: 23122592
- 315) Ott J, Liu Z, Shen Y. Challenging False Discovery Rate: A Partition Test Based on p Values in Human Case-Control Association Studies. *Hum Hered.* 2012 Nov 13;74(1):45-50. PMID: 23154528
- 316) Yuferov V, Ho A, Morgello S, Yang Y, Ott J, Kreek MJ. Expression of Ephrin Receptors and Ligands in Postmortem Brains of HIV-Infected Subjects With and Without Cognitive Impairment. *J Neuroimmune Pharmacol.* 2013 Mar;8(1):333-44. doi: 10.1007/s11481-012-9429-1. Epub 2013 Jan 12. PMID:23314923, PMC3587720
- 317) Thomas BN, Thakur TJ, Yi L, Guindo A, Diallo DA, Ott J. Extensive ethnogenomic diversity of endothelial nitric oxide synthase (eNOS) polymorphisms. *Gene Regul Syst Bio.* 2013;7:1-10. doi: 10.4137/GRSB.S10857. Epub 2013 Jan 15. PMID:23400313
- 318) Proudnikov D, Randesi M, Levran O, Yuferov V, Crystal H, Ho A, Ott J, Kreek MJ. Polymorphisms of the Kappa Opioid Receptor and Prodynorphin Genes: HIV risk and HIV Natural History. *J Acquir Immune Defic Syndr.* 2013 Feb 7. [Epub ahead of print] PMID:23392455
- 319) Suo C, Touloupoulou T, Bramon E, Walshe M, Picchioni M, Murray R, Ott J. Analysis of multiple phenotypes in genome-wide genetic mapping studies. *BMC Bioinformatics.* 2013 May 2;14:151. doi: 10.1186/1471-2105-14-151. PMID: 23639181
- 320) Levran O, Peles E, Randesi M, Shu X, Ott J, Shen PH, Adelson M, Kreek MJ. Association of genetic variation in pharmacodynamic factors with methadone dose required for effective treatment of opioid addiction. *Pharmacogenomics.* 2013 May;14(7):755-68. doi: 10.2217/pgs.13.58. PMID: 23651024
- 321) Xi B, Shen Y, Zhao X, Chandak GR, Cheng H, Hou D, Li Y, Ott J, Zhang Y, Wang X, Mi J. Association of common variants in/near six genes (ATP2B1, CSK, MTHFR, CYP17A1, STK39 and FGF5) with blood pressure/hypertension risk in Chinese children. *J Hum Hypertens.* 2013 Jun 13. doi: 10.1038/jhh.2013.50. [Epub ahead of print] PMID: 23759979
- 322) Negi S, Juyal G, Senapati S, Prasad P, Gupta A, Singh S, Kashyap S, Kumar A, Kumar U, Gupta R, Kaur S, Agrawal S, Aggarwal A, Ott J, Jain S, Juyal RC, Thelma BK. A genome-wide association study reveals ARL15, a novel non-HLA susceptibility gene for Rheumatoid arthritis in north Indians. *Arthritis Rheum.* 2013 Aug 5. doi: 1002/art.38110. [Epub ahead of print] PMID: 23918589
- 323) Li Y, Zhang K, Chen H, Sun F, Xu J, Wu Z, Li P, Zhang L, Du Y, Luan H, Li X, Wu L, Li H, Wu H, Li X, Li X, Zhang X,

- Gong L, Dai L, Sun L, Zuo X, Xu J, Gong H, Li Z, Tong S, Wu M, Li X, Xiao W, Wang G, Zhu P, Shen M, Liu S, Zhao D, Liu W, Wang Y, Huang C, Jiang Q, Liu G, Liu B, Hu S, Zhang W, Zhang Z, You X, Li M, Hao W, Zhao C, Leng X, Bi L, Wang Y, Zhang F, Shi Q, Qi W, Zhang X, Jia Y, Su J, Li Q, Hou Y, Wu Q, Xu D, Zheng W, Zhang M, Wang Q, Fei Y, Zhang X, Li J, Jiang Y, Tian X, Zhao L, Wang L, Zhou B, Li Y, Zhao Y, Zeng X, Ott J, Wang J, Zhang F. A genome-wide association study in Han Chinese identifies a susceptibility locus for primary Sjögren's syndrome at 7q11.23. *Nat Genet.* 2013 Nov;45(11):1361-5. doi: 10.1038/ng.2779. Epub 2013 Oct 6. PMID: 24097066
- 324) Erdmann J, Stark K, Esslinger UB, Rumpf PM, Koesling D, de Wit C, Kaiser FJ, Braunholz D, Medack A, Fischer M, Zimmermann ME, Tennstedt S, Graf E, Eck S, Aherrahou Z, Nahrstaedt J, Willenborg C, Bruse P, Brænne I, Nöthen MM, Hofmann P, Braund PS, Mergia E, Reinhard W, Burgdorf C, Schreiber S, Balmforth AJ, Hall AS, Bertram L, Steinhagen-Thiessen E, Li SC, März W, Reilly M, Kathiresan S, McPherson R, Walter U; CARDIoGRAM, Ott J, Samani NJ, Strom TM, Meitinger T, Hengstenberg C, Schunkert H. Dysfunctional nitric oxide signalling increases risk of myocardial infarction. *Nature.* 2013 Dec 19;504(7480):432-6. doi: 10.1038/nature12722. Epub 2013 Nov 10. PMID: 24213632
- 325) Mayer-Blackwell B, Schlussman SD, Butelman ER, Ho A, Ott J, Kreek MJ, Zhang Y. Self administration of oxycodone by adolescent and adult mice affects striatal neurotransmitter receptor gene expression. *Neuroscience.* 2014 Jan 31;258:280-91. doi: 10.1016/j.neuroscience.2013.10.062. Epub 2013 Nov 9. PMID: 24220688
- 326) Zhang Y, Mayer-Blackwell B, Schlussman SD, Randesi M, Butelman ER, Ho A, Ott J, Kreek MJ. Extended access oxycodone self-administration and neurotransmitter receptor gene expression in the dorsal striatum of adult C57BL/6 J mice. *Psychopharmacology (Berl).* 2014 Apr;231(7):1277-87. doi: 10.1007/s00213-013-3306-3. Epub 2013 Nov 13. PMID: 24221825
- 327) Levran O, Randesi M, Li Y, Rotrosen J, Ott J, Adelson M, Jeanne Kreek M. Drug addiction and stress-response genetic variability: association study in African Americans. *Ann Hum Genet.* 2014 Jul;78(4):290-8. doi: 10.1111/ahg.12064. Epub 2014 Apr 26. PMID: 24766650
- 328) Juyal G, Negi S, Sood A, Gupta A, Prasad P, Senapati S, Zaneveld J, Singh S, Midha V, van Sommeren S, Weersma RK, Ott J, Jain S, Juyal RC, Thelma BK. Genome-wide association scan in north Indians reveals three novel HLA-independent risk loci for ulcerative colitis. *Gut.* 2014 May 16. pii: gutjnl-2013-306625. doi: 10.1136/gutjnl-2013-306625. [Epub ahead of print] PMID: 24837172
- 329) Levran O, Peles E, Randesi M, Li Y, Rotrosen J, Ott J, Adelson M, Kreek MJ. Stress-related genes and heroin addiction: a role for a functional FKBP5 haplotype. *Psychoneuroendocrinology.* 2014 Jul;45:67-76. doi: 10.1016/j.psyneuen.2014.03.017. Epub 2014 Apr 6. PMID: 24845178
- 330) Zhang Q, Long Q, Ott J. AprioriGWAS, a New Pattern Mining Strategy for Detecting Genetic Variants Associated with Disease through Interaction Effects. *PLoS Comput Biol.* 2014 Jun 5;10(6):e1003627. doi: 10.1371/journal.pcbi.1003627. eCollection 2014 Jun. PMID: 24901472
- 331) Zhang Y, Brownstein AJ, Buonora M, Niikura K, Ho A, Correa da Rosa J, Kreek MJ, Ott J. Self administration of oxycodone alters synaptic plasticity gene expression in the hippocampus differentially in male adolescent and adult mice. *Neuroscience.* 2015 Jan 29;285:34-46. doi: 10.1016/j.neuroscience.2014.11.013. Epub 2014 Nov 14. PMID:25446355
- 332) Levran O, Peles E, Randesi M, Correa da Rosa J, Ott J, Rotrosen J, Adelson M, Kreek MJ. Dopaminergic pathway polymorphisms and heroin addiction: further support for association of CSNK1E variants. *Pharmacogenomics.* 2014 Dec;15(16):2001-9. doi: 10.2217/pgs.14.145. PMID:25521358
- 333) Onengut-Gumuscu S, Chen WM, Burren O, Cooper NJ, Quinlan AR, Mychaleckyj JC, Farber E, Bonnie JK, Szpak M, Schofield E, Achuthan P, Guo H, Fortune MD, Stevens H, Walker NM, Ward LD, Kundaje A, Kellis M, Daly MJ, Barrett JC, Cooper JD, Deloukas P; Type 1 Diabetes Genetics Consortium, Todd JA, Wallace C, Concannon P, Rich SS. Fine mapping of type 1 diabetes susceptibility loci and evidence for colocalization of causal variants with

lymphoid gene enhancers. *Nat Genet.* 2015 Apr;47(4):381-6. doi: 10.1038/ng.3245. Epub 2015 Mar 9. PMID:25751624

- 334) Ott J, Wang J, Leal SM. Genetic linkage analysis in the age of whole-genome sequencing. *Nat Rev Genet.* 2015 May;16(5):275-84. doi: 10.1038/nrg3908. Epub 2015 Mar 31. Review. PMID:25824869
- 335) Levran O, Randesi M, da Rosa JC, Ott J, Rotrosen J, Adelson M, Kreek MJ. Overlapping dopaminergic pathway genetic susceptibility to heroin and cocaine addictions in African Americans. *Ann Hum Genet.* 2015 May; 79(3):188-98. doi: 10.1111/ahg.12104. Epub 2015 Feb 27. PMID:25875614
- 336) Maass PG, Aydin A, Luft FC, Schächterle C, Weise A, Stricker S, Lindschau C, Vaegler M, Qadri F, Toka HR, Schulz H, Krawitz PM, Parkhomchuk D, Hecht J, Hollfinger I, Wefeld-Neuenfeld Y, Bartels-Klein E, Mühl A, Kann M, Schuster H, Chitayat D, Bialer MG, Wienker TF, Ott J, Rittscher K, Liehr T, Jordan J, Plessis G, Tank J, Mai K, Naraghi R, Hodge R, Hopp M, Hattenbach LO, Busjahn A, Rauch A, Vandeput F, Gong M, Rüschenhoff F, Hübner N, Haller H, Mundlos S, Bilginturan N, Movsesian MA, Klussmann E, Toka O, Bähring S. PDE3A mutations cause autosomal dominant hypertension with brachydactyly. *Nat Genet.* 2015 Jun;47(6):647-53. doi: 10.1038/ng.3302. Epub 2015 May 11. PMID:25961942
- 337) Wang J, Tao Y, Song F, Sun Y, Ott J, Saffen D. Common Regulatory Variants of CYFIP1 Contribute to Susceptibility for Autism Spectrum Disorder (ASD) and Classical Autism. *Ann Hum Genet.* 2015 Jun 19. doi: 10.1111/ahg.12121. PMID:26094621
- 338) Guan X, Song Y, Ott J, Zhang Y, Li C, Xin T, Li Z, Gan Y, Li J, Zhou S, Zhou Y. The ADAMTS1 Gene Is Associated with Familial Mandibular Prognathism. *J Dent Res.* 2015 Sep;94(9):1196-201. doi: 10.1177/0022034515589957. Epub 2015 Jun 29. PMID:26124221
- 339) Imai A, Nakaya A, Fahiminiya S, Tétreault M, Majewski J, Sakata Y, Takashima S, Lathrop M, Ott J. Beyond Homozygosity Mapping: Family-Control analysis based on Hamming distance for prioritizing variants in exome sequencing. *Sci Rep.* 2015 Jul 6;5:12028. doi: 10.1038/srep12028. PMID:26143870
- 340) Li Y, Cagirici HB, Horpaopan S, Ott J, Imai A, Majewski J, Lathrop M. Leveling the Playing Field in Homozygosity Mapping Using Map Distances. *Ann Hum Genet.* 2015 Jul 15. doi: 10.1111/ahg.12125. PMID:26179257
- 341) Levran O, Peles E, Randesi M, Correa da Rosa J, Ott J, Rotrosen J, Adelson M, Kreek MJ. Susceptibility loci for heroin and cocaine addiction in the serotonergic and adrenergic pathways in populations of different ancestry. *Pharmacogenomics.* 2015 Aug;16(12):1329-42. doi: 10.2217/pgs.15.86. PMID:26227246
- 342) Levran O, Peles E, Randesi M, Correa da Rosa J, Ott J, Rotrosen J, Adelson M, Kreek MJ. Glutamatergic and GABAergic susceptibility loci for heroin and cocaine addiction in subjects of African and European ancestry. *Prog Neuropsychopharmacol Biol Psychiatry.* 2016 Jan 4;64:118-23. doi: 10.1016/j.pnpbp.2015.08.003. PMID:26277529
- 343) Xu T, Wang Y, Li Z, Huang J, Lui SS, Tan SP, Yu X, Cheung EF, He MG, Ott J, Gur RE, Gur RC, Chan RC. Heritability and familiarity of neurological soft signs: evidence from healthy twins, patients with schizophrenia and non-psychotic first-degree relatives. *Psychol Med.* 2015 Sep 8:1-7. PMID:26347209
- 344) Levran O, Peles E, Randesi M, Correa da Rosa J, Ott J, Rotrosen J, Adelson M, Kreek MJ. Synaptic Plasticity and Signal Transduction Gene Polymorphisms and Vulnerability to Drug Addictions in Populations of European or African Ancestry. *CNS Neurosci Ther.* 2015 Sep 19. doi: 10.1111/cns.12450. PMID:26384852

EDITORIALS, BOOK CHAPTERS, CONFERENCE REPORTS, LETTERS

- 1) Ott J (1979) Ascertainment in the Seattle lipid studies. In: *Genetic Analysis of Common Diseases and Applications to Predictive Factors in Coronary Disease*, edited by C.F. Sing and M. Skolnick. New York, Alan R. Liss, pp. 383-388
- 2) Ott J (1979) Genetic linkage studies in man. *Transplant Proc* 11, 1689-1691
- 3) Weber W, Ott J, Gencik A, Muller HJ (1983) Familial cancer - genetically determined? *Anticancer Research* 3, 133-142
- 4) Ott J (1984) Mathematical models and methods in human pedigree analysis. In: *Modeling and Analysis in Biomedicine*, edited by C. Nicolini. Singapore, World Scientific Publishing Co, pp. 185-200
- 5) Conneally PM, Edwards JH, Kidd KK, Lalouel J-M, Morton NE, Ott J, White R (1985) Report of the committee on methods of linkage analysis and reporting. *Cytogenet Cell Genet* 40, 356-359
- 6) Ott J (1986) A short guide to linkage analysis. In: *Analysis of Human Genetic Diseases*, edited by Kay Davies, IRL Press, Great Britain
- 7) Ott J, Aston C, Baur M, Bishop T, Chakravarti A, Clayton J, Edwards JH, Elston RC, Keats B, Lathrop M, Neugebauer M, Pascoe L (1987) Detection and estimation of linkage, especially multipoint mapping. In: *Human Genetics, Proceedings of the 7th International Congress Berlin 1986*, Friedrich Vogel and Karl Sperling (eds.). Springer, Berlin and New York, pp. 188-189
- 8) Ott J (1988) Letter to the editor. *Genomics* 3, 91
- 9) Lehner T, Sandkuyl LA, **Ott J** (1989) Linkage analysis for a disease with a complex mode of inheritance in a simulated data set. In: *Multipoint Mapping and Linkage Based Upon Affected Pedigree Members: Genetic Analysis Workshop 6* (Elston RC, Spence MA, Hodge SE, MacCluer JW, Eds). New York: Alan R. Liss
- 10) Keats B, Ott J, Conneally M (1989) Human Gene Mapping 10 - Report of the committee on linkage and gene order. *Cytogenet Cell Genet* 51, 459-502
- 11) Ott J (1990) Genetic linkage and complex diseases: A comment. *Genet Epidemiol* 7, 35-36
- 12) Ott J (1990) Invited editorial: Cutting a Gordian knot in the linkage analysis of complex human traits. *Am J Hum Genet* 46, 219-221
- 13) Ott J (1990) Genetic interpretation of disease clustering. In: *Convergent issues in genetics and demography*, edited by J. Adams, D.A. Lam, A.I. Hermalin, and P.E. Smouse. New York: Oxford University Press, pp. 245-255
- 14) Keats BJB, Sherman SL, Ott J (1990) Human Gene Mapping 10.5 - Report of the committee on linkage and gene order. *Cytogenet Cell Genet* 55, 387-94
- 15) Baron M, Endicott J, Ott J (1990) Genetic linkage in mental illness. Limitations and prospects. *Brit J Psychiatry* 157, 645-655
- 16) Ott J (1990) Genetic linkage analysis under uncertain disease definition. In *Banbury Report 33: Genetics and Biology and Alcoholism*, edited by C.R. Cloninger and H. Begleiter. Cold Spring Harbor, New York: Cold Spring Harbor Laboratory Press, pp. 327-331
- 17) Keats BJB, Sherman SL, Morton NE, Robson EB, Buetow KH, Cartwright PE, Chakravarti A, Francke U, Green PP, Ott J (1991) Guidelines for Human Linkage Maps: An International System for Human Linkage Maps (ISLM, 1990). *Genomics* 9, 557-560
- 18) Ott J (1991) Computer simulation methods in human linkage analysis. In: *Recent Progress in the Genetic Epidemiology of Cancer*, edited by H.T. Lynch and P. Tautu. Berlin: Springer Verlag, pp. 135-139
- 19) Lernmark Å, Ducat L, Eisenbarth G, Ott J, Perlmutter MA, Rubinstein P, Spielman R (1991) Human cell lines from families available for diabetes research. *Diabetologia* 34, 61 (letter to the editor)

- 20) Ott J (1991) Computer programs for linkage analysis. In: *Recent Progress in the Genetic Epidemiology of Cancer*, edited by H.T. Lynch and P. Tautu. Berlin: Springer Verlag, pp. 140-144
- 21) Ott J (1991) Principles of human genetic linkage analysis. In: *Molecular Genetic Approaches to Neuropsychiatric Diseases*, edited by J. Brosius and R.T. Freneau. New York: Academic Press, pp. 35-53
- 22) Ott J, Terwilliger JD (1992) Assessing the evidence for linkage in psychiatric genetics. In: *Genetic Research in Psychiatry*, edited by J. Mendlewicz and H. Hippus. New York: Springer, pp. 245-249
- 23) Ott J (1992) Introductory remarks: genetic models and statistical approaches. *Ann Med* 24, 375-377
- 24) Ott J (1992) The future of multilocus linkage analysis. *Ann Med* 24, 401-403
- 25) LeBeau MM, Overhauser J, Straub RE, Silverman G, Gilliam TC, Ott J, O'Connell P, Francke U, Geurts van Kessel A (1993) Report of the first international workshop on human chromosome 18 mapping. *Cytogenet Cell Genet* 63, 78-95
- 26) Ott J (1993) Recent developments in the theoretical aspects of linkage analysis. In *Human Population Genetics*, edited by P.P. Majumder. New York: Plenum Press, pp. 165-179
- 27) Ott J (1994) Choice of genetic models for linkage analysis of psychiatric traits. In: *Genetic Approaches to Mental Disorders*, edited by E.S. Gershon and C.R. Cloninger. American Psychiatric Press, Washington DC, pp. 63-75
- 28) Vieland VJ, Knowles JA, Fyer AJ, Stefanovich M, Freimer NF, Lish J, Adams P, Woodley K, Rassnick H, Heiman GA, White P, Das K, Klein DF, Ott J, Weissman MM, Gilliam TC (1994) Linkage study of panic disorder: A preliminary report. In: *Genetic Approaches to Mental Disorders*, edited by E.S. Gershon and C.R. Cloninger. American Psychiatric Press, Washington DC, pp. 345-354
- 29) Ott J, Donis-Keller H (1994) Statistical methods in genetic mapping. *Genomics* 22, 496-497
- 30) Shugart YY, Banerjee P, Knowles JA, Lewis CA, Jacobson SG, Matise TC, Penchaszadeh G, Gilliam TC, Ott J (1995) Fine genetic mapping of a gene for autosomal recessive retinitis pigmentosa on chromosome 6p21. *Am J Hum Genet* 57, 499-502
- 31) Antonarakis SE, Blouin J-L, Pulver AE, Wolyniec P, Lasseter VK, Nestadt G, Kasch L, Babb R, Kazazian HH, Dombroski B, Kimberland M, Ott J, Housman D, Karayiorgou M, MacLean CJ (1995) Schizophrenia susceptibility and chromosome 6p24-22. *Nature Genetics* 11, 235-236
- 32) Ott J (1995) How do you compute a lod score? *Nature Genetics* 11, 354-355
- 33) Speer MC, Terwilliger JD, Ott J (1995) Data simulation for GAW9 problems 1 and 2. *Genet Epidemiol* 12, 561-564
- 34) Gambacorti-Passerini C, Ott J, Bergen A (1995) The Human Molecular Genetics Network. *New Engl J Med* 333, 1573
- 35) Ott J (1996) Complex traits on the map. *Nature* 379, 772-773
- 36) Pauls DL, Ott J, Paul SM, Allen CR, Fann CSJ, Carulli JP, Falls KM, Bouthillier CA, Gravius TC, Keith TP, Egeland JA, Ginns EI (1996) Chromosome 18 markers: linked or not linked to bipolar affective disorders in the Old Order Amish? A reply to Gershon et al. *Am J Hum Genet* 58, 1384-1385
- 37) Ott J (1996) Human linkage maps. In: *Encyclopedia of Molecular Biology and Molecular Medicine*, Vol. 3, Meyers RA, ed. VCH Verlagsgesellschaft, Einheim Germany, pp. 233-240
- 38) Simonic I, Ott J (1996) Novel etiological hypotheses imply new analysis methods for schizophrenia genetics. *Schizophrenia Research* 20, 235-237
- 39) Ott J (1996) Estimating crossover frequencies and testing for numerical interference with highly polymorphic markers. In: *Genetic Mapping and DNA Sequencing*, Vol. 81 in "The IMA Volumes in Mathematics and its Applications," eds. Terry Speed and Michael S. Waterman. New York: Springer, pp 49-63

- 40) Shugart YY, Ott J (1996) Using lod score peak length to distinguish true and false positives, in: *Proc Eur Math Genet Meeting*, Clementi M, Forabusco P, eds. Padova University Press, Padova, Italy, pp. 127-133
- 41) Matisse TC, Donis-Keller H, Ott J (1996) Statistical methods in genetic mapping. *Genomics* 36, 223-225
- 42) Mérette C, Ott J (1997) Finding and excluding gene locations by linkage analysis. In *The Molecular and Genetic Basis of Neurological Disease, second edition*, edited by R.N. Rosenberg, S. Prusiner, S. DiMauro, and R.L. Barchi. Boston MA: Butterworth-Heinemann, pp. 29-32
- 43) Ott J (1997) Testing for interference in human genetic maps. *J Mol Med* 75, 414-419
- 44) Ott J (1997) Genetic mapping in complex disorders. In *Genetic Mapping of Disease Genes*, edited by I-H Pawlowitzki, JH Edwards, and EA Thompson. New York: Academic Press, pp. 23-30
- 45) Lernmark Å, Eisenbarth G, Ducat L, Erlich HA, Faustman D, Maclaren NK, Ott J, Permutt MA, She J-X, She J-X, Todd J (1997) Family cell lines available for research - an endangered resource? *Am J Hum Genet* 61, 778-779
- 46) Lernmark Å, Ott J (1998) Sometimes it's hot, sometimes it's not. *Nature Genetics* 19, 213-214
- 47) Ott J, Lucek P (1998) Complex traits on the map. In *Genes and Environment in Cancer*, edited by Schwab M, Rabes H, Munk K, Hofschneider PH. Heidelberg: Springer, pp. 285-291
- 48) Ott J (1999) Methods of analysis and resources available for genetic trait mapping. *J Heredity* 90, 68-70
- 49) Schaid DJ, Buetow K, Weeks DE, Wijsman E, Guo SW, Ott J, Dahl C (1999) Discovery of cancer susceptibility genes: study designs, analytic approaches, and trends in technology. *J Natl Cancer Inst Monogr* 26, 1-16
- 50) Ott J (2000) Predicting the range of linkage disequilibrium. *Proc Natl Acad Sci USA* 97, 2-3
- 51) Ott J (2001) Major strengths and weaknesses of the lod score method. *Adv Genet* 42, 125-132
- 52) Sherriff A, Ott J (2001) Applications of neural networks for gene finding. *Adv Genet* 42, 287-297
- 53) Nothnagel M, Ott J (2002) Statistical gene mapping of traits in humans---hypertension as a complex trait: Is it amenable to genetic analysis? *Semin Nephrol* 22, 105-114
- 54) Ott J (2004) Association of genetic loci – Replication or not, that is the question (editorial). *Neurology* 63, 955-958
- 55) Hoh, J. and Ott, J. 2009. Scan statistics in genome-wide scan for complex trait loci. In *Scan Statistics: Methods and Applications* (Statistics for Industry and Technology), Vol 1 (ed. J. Glaz, V. Pozdnyakov, and S. Wallenstein), pp. 197-204. Birkhäuser Boston, Boston
- 56) Ott J. 2009. Richard S. Spielman 1946-2009 (obituary). *Nature Genetics* 41, 1159