

1. Satterstrom, F. K., Walters, R. K., Singh, T., Wigdor, E. M., Lescai, F., Demontis, D., Kosmicki, J. A., Grove, J., Stevens, C., Bybjerg-Grauholm, J., Bækvad-Hansen, M., Palmer, D. S., Maller, J. B., iPSYCH-Broad Consortium, Nordentoft, M., Mors, O., Robinson, E. B., Hougaard, D. M., Werge, T. M., ... Daly, M. J. (2019). Autism spectrum disorder and attention deficit hyperactivity disorder have a similar burden of rare protein-truncating variants. *Nature Neuroscience*, 22(12), 1961–1965. <https://doi.org/10.1038/s41593-019-0527-8>
2. Matey-Hernandez, M. L., Danish Pan Genome Consortium, Brunak, S., & Izarzugaza, J. M. G. (2018). Benchmarking the HLA typing performance of Polysolver and Optitype in 50 Danish parental trios. *BMC Bioinformatics*, 19(1), 239. <https://doi.org/10.1186/s12859-018-2239-6>
3. Ganna, A., Satterstrom, F. K., Zekavat, S. M., Das, I., Kurki, M. I., Churchhouse, C., Alfoldi, J., Martin, A. R., Havulinna, A. S., Byrnes, A., Thompson, W. K., Nielsen, P. R., Karczewski, K. J., Saarentaus, E., Rivas, M. A., Gupta, N., Pietiläinen, O., Emdin, C. A., Lescai, F., ... Neale, B. M. (2018). Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. *American Journal of Human Genetics*, 102(6), 1204–1211. <https://doi.org/10.1016/j.ajhg.2018.05.002>
4. Navarro, A. B., & Lescai, F. (2018). Editorial. *New Biotechnology*, 40(Pt B), 185. <https://doi.org/10.1016/j.nbt.2017.11.001>
5. Jensen, J. M., Villesen, P., Friberg, R. M., Danish Pan-Genome Consortium, Mailund, T., Besenbacher, S., & Schierup, M. H. (2017). Assembly and analysis of 100 full MHC haplotypes from the Danish population. *Genome Research*, 27(9), 1597–1607. <https://doi.org/10.1101/gr.218891.116>
6. Maretty, L., Jensen, J. M., Petersen, B., Sibbesen, J. A., Liu, S., Villesen, P., Skov, L., Belling, K., Theil Have, C., Izarzugaza, J. M. G., Grosjean, M., Bork-Jensen, J., Grove, J., Als, T. D., Huang, S., Chang, Y., Xu, R., Ye, W., Rao, J., ... Schierup, M. H. (2017). Sequencing and de novo assembly of 150 genomes from Denmark as a population reference. *Nature*, 548(7665), 87–91. <https://doi.org/10.1038/nature23264>
7. Lescai, F., Diderichsen, B., Van Montagu, M., & Cole, J. (2017). In memory of Professor Brian Frederic Carl Clark: Contributions from friends. *New Biotechnology*, 38(Pt A), 3–4. <https://doi.org/10.1016/j.nbt.2017.02.005>
8. Lescai, F., Als, T. D., Li, Q., Nyegaard, M., Andorsdottir, G., Biskopstø, M., Hedemand, A., Fiorentino, A., O'Brien, N., Jarram, A., Liang, J., Grove, J., Pallesen, J., Eickhardt, E., Mattheisen, M., Bolund, L., Demontis, D., Wang, A. G., McQuillin, A., ... Børglum, A. D. (2017). Whole-exome sequencing of individuals from an isolated population implicates rare risk variants in bipolar disorder. *Translational Psychiatry*, 7(2), e1034. <https://doi.org/10.1038/tp.2017.3>
9. Lescai, F. (2017). Special issue of new biotechnology in memory of professor Brian F.C. Clark (1936-2014). *New Biotechnology*, 38(Pt A), 1. <https://doi.org/10.1016/j.nbt.2016.11.007>
10. Gregersen, N. O., Lescai, F., Liang, J., Li, Q., Als, T., Buttenschøn, H. N., Hedemand, A., Biskopstø, M., Wang, J., Wang, A. G., Børglum, A. D., Mors, O., & Demontis, D. (2016). Whole-exome sequencing implicates DGKH as a risk gene for panic disorder in the Faroese population. *American Journal of Medical Genetics. Part B, Neuropsychiatric Genetics: The Official Publication of the International Society of Psychiatric Genetics*, 171(8), 1013–1022. <https://doi.org/10.1002/ajmg.b.32464>
11. Demontis, D., Lescai, F., Børglum, A., Glerup, S., Østergaard, S. D., Mors, O., Li, Q., Liang, J., Jiang, H., Li, Y., Wang, J., Lesch, K.-P., Reif, A., Buitelaar, J. K., & Franke, B. (2016). Whole-Exome Sequencing Reveals Increased Burden of Rare Functional and Disruptive Variants in Candidate Risk Genes in Individuals With Persistent Attention-Deficit/Hyperactivity Disorder. *Journal of the American Academy of Child and Adolescent Psychiatry*, 55(6), 521–523. <https://doi.org/10.1016/j.jaac.2016.03.009>
12. Poulsen, J. B., Lescai, F., Grove, J., Bækvad-Hansen, M., Christiansen, M., Hagen, C. M., Maller, J., Stevens, C., Li, S., Li, Q., Sun, J., Wang, J., Nordentoft, M., Werge, T. M., Mortensen, P. B., Børglum, A. D., Daly, M., Hougaard, D. M., Bybjerg-Grauholm, J., & Hollegaard, M. V. (2016). High-Quality Exome Sequencing of Whole-Genome Amplified Neonatal Dried Blood Spot DNA. *PLoS One*, 11(4), e0153253. <https://doi.org/10.1371/journal.pone.0153253>

13. Robinson, E. B., St Pourcain, B., Anttila, V., Kosmicki, J. A., Bulik-Sullivan, B., Grove, J., Maller, J., Samocha, K. E., Sanders, S. J., Ripke, S., Martin, J., Hollegaard, M. V., Werge, T., Hougaard, D. M., iPSYCH-SSI-Broad Autism Group, Neale, B. M., Evans, D. M., Skuse, D., Mortensen, P. B., ... Daly, M. J. (2016). Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. *Nature Genetics*, *48*(5), 552–555. <https://doi.org/10.1038/ng.3529>
14. Liu, S., Huang, S., Rao, J., Ye, W., Genome Denmark Consortium, Krogh, A., & Wang, J. (2015). Discovery, genotyping and characterization of structural variation and novel sequence at single nucleotide resolution from de novo genome assemblies on a population scale. *GigaScience*, *4*, 64. <https://doi.org/10.1186/s13742-015-0103-4>
15. Lescai, F., & Rudelsheim, P. (2015). Editorial. *New Biotechnology*, *32*(6), 533. <https://doi.org/10.1016/j.nbt.2015.07.009>
16. Rajkumar, A. P., Qvist, P., Lazarus, R., Lescai, F., Ju, J., Nyegaard, M., Mors, O., Børglum, A. D., Li, Q., & Christensen, J. H. (2015). Experimental validation of methods for differential gene expression analysis and sample pooling in RNA-seq. *BMC Genomics*, *16*, 548. <https://doi.org/10.1186/s12864-015-1767-y>
17. Williams, H. J., Hurst, J. R., Ocaka, L., James, C., Pao, C., Chanudet, E., Lescai, F., Stanescu, H. C., Kleta, R., GOSgene, Rosser, E., Bacchelli, C., & Beales, P. (2016). The use of whole-exome sequencing to disentangle complex phenotypes. *European Journal of Human Genetics: EJHG*, *24*(2), 298–301. <https://doi.org/10.1038/ejhg.2015.121>
18. Le Quesne Stabej, P., Williams, H. J., James, C., Tekman, M., Stanescu, H. C., Kleta, R., Ocaka, L., Lescai, F., Storr, H. L., Bitner-Glindzicz, M., Bacchelli, C., Conway, G. S., & GOSgene. (2016). STAG3 truncating variant as the cause of primary ovarian insufficiency. *European Journal of Human Genetics: EJHG*, *24*(1), 135–138. <https://doi.org/10.1038/ejhg.2015.107>
19. Besenbacher, S., Liu, S., Izarzugaza, J. M. G., Grove, J., Belling, K., Bork-Jensen, J., Huang, S., Als, T. D., Li, S., Yadav, R., Rubio-García, A., Lescai, F., Demontis, D., Rao, J., Ye, W., Mailund, T., Friborg, R. M., Pedersen, C. N. S., Xu, R., ... Rasmussen, S. (2015). Novel variation and de novo mutation rates in population-wide de novo assembled Danish trios. *Nature Communications*, *6*, 5969. <https://doi.org/10.1038/ncomms6969>
20. Waters, A. M., Asfahani, R., Carroll, P., Bicknell, L., Lescai, F., Bright, A., Chanudet, E., Brooks, A., Christou-Savina, S., Osman, G., Walsh, P., Bacchelli, C., Chapgier, A., Vernay, B., Bader, D. M., Deshpande, C., O'Sullivan, M., Ocaka, L., Stanescu, H., ... Beales, P. L. (2015). The kinetochore protein, CENPF, is mutated in human ciliopathy and microcephaly phenotypes. *Journal of Medical Genetics*, *52*(3), 147–156. <https://doi.org/10.1136/jmedgenet-2014-102691>
21. Thomas, A. C., Williams, H., Setó-Salvia, N., Bacchelli, C., Jenkins, D., O'Sullivan, M., Mengrelis, K., Ishida, M., Ocaka, L., Chanudet, E., James, C., Lescai, F., Anderson, G., Morrogh, D., Ryten, M., Duncan, A. J., Pai, Y. J., Saraiva, J. M., Ramos, F., ... Stanier, P. (2014). Mutations in SNX14 cause a distinctive autosomal-recessive cerebellar ataxia and intellectual disability syndrome. *American Journal of Human Genetics*, *95*(5), 611–621. <https://doi.org/10.1016/j.ajhg.2014.10.007>
22. Lescai, F., Marasco, E., Bacchelli, C., Stanier, P., Mantovani, V., & Beales, P. (2014). Identification and validation of loss of function variants in clinical contexts. *Molecular Genetics & Genomic Medicine*, *2*(1), 58–63. <https://doi.org/10.1002/mgg3.42>
23. Kelberman, D., Islam, L., Lakowski, J., Bacchelli, C., Chanudet, E., Lescai, F., Patel, A., Stupka, E., Buck, A., Wolf, S., Beales, P. L., Jacques, T. S., Bitner-Glindzicz, M., Liasis, A., Lehmann, O. J., Kohlhase, J., Nischal, K. K., & Sowden, J. C. (2014). Mutation of SALL2 causes recessive ocular coloboma in humans and mice. *Human Molecular Genetics*, *23*(10), 2511–2526. <https://doi.org/10.1093/hmg/ddt643>
24. Webb, E. A., AlMutair, A., Kelberman, D., Bacchelli, C., Chanudet, E., Lescai, F., Andoniadou, C. L., Banyan, A., Alsawaid, A., Alrifai, M. T., Alahmesh, M. A., Balwi, M., Mousavy-Gharavy, S. N., Lukovic, B., Burke, D., McCabe, M. J., Kasia, T., Kleta, R., Stupka, E., ... Dattani, M. T. (2013). ARNT2 mutation causes hypopituitarism, post-natal microcephaly, visual and renal anomalies. *Brain: A Journal of Neurology*, *136*(Pt 10), 3096–3105. <https://doi.org/10.1093/brain/awt218>
25. Lescai, F., Bonfiglio, S., Bacchelli, C., Chanudet, E., Waters, A., Sisodiya, S. M., Kasperavičiūtė, D., Williams, J., Harold, D., Hardy, J., Kleta, R., Cirak, S., Williams, R., Achermann, J. C., Anderson, J., Kelsell, D., Vulliamy, T., Houlden, H., Wood, N., ... Stupka, E. (2012). Characterisation and validation of insertions and deletions in 173 patient exomes. *PLoS One*, *7*(12), e51292. <https://doi.org/10.1371/journal.pone.0051292>

26. Su, Z., Gay, L. J., Strange, A., Palles, C., Band, G., Whiteman, D. C., Lescai, F., Langford, C., Nanji, M., Edkins, S., Winkel, A. van der, Levine, D., Sasieni, P., Bellenguez, C., Howarth, K., Freeman, C., Trudgill, N., Tucker, A. T., Pirinen, M., ... Wellcome Trust Case Control Consortium 2. (2012). Common variants at the MHC locus and at chromosome 16q24.1 predispose to Barrett's esophagus. *Nature Genetics*, *44*(10), 1131–1136. <https://doi.org/10.1038/ng.2408>
27. Marchegiani, F., Spazzafumo, L., Cardelli, M., Provinciali, M., Lescai, F., Franceschi, C., & Antonicelli, R. (2012). Paraoxonase-1 55 LL Genotype Is Associated with No ST-Elevation Myocardial Infarction and with High Levels of Myoglobin. *Journal of Lipids*, *2012*, 601796. <https://doi.org/10.1155/2012/601796>
28. Ellinghaus, E., Stanulla, M., Richter, G., Ellinghaus, D., Kronnie, G. te, Cario, G., Cazzaniga, G., Horstmann, M., Panzer Grümayer, R., Cavé, H., Trka, J., Cinek, O., Teigler-Schlegel, A., ElSharawy, A., Häslér, R., Nebel, A., Meissner, B., Bartram, T., Lescai, F., ... Franke, A. (2012). Identification of germline susceptibility loci in ETV6-RUNX1-rearranged childhood acute lymphoblastic leukemia. *Leukemia*, *26*(5), 902–909. <https://doi.org/10.1038/leu.2011.302>
29. Taccioli, C., Tegnér, J., Maselli, V., Gomez-Cabrero, D., Altobelli, G., Emmett, W., Lescai, F., Gustincich, S., & Stupka, E. (2011). ParkDB: A Parkinson's disease gene expression database. *Database: The Journal of Biological Databases and Curation*, *2011*, bar007. <https://doi.org/10.1093/database/bar007>
30. Lescai, F., Chiamenti, A. M., Codemo, A., Pirazzini, C., D'Agostino, G., Ruaro, C., Ghidoni, R., Benussi, L., Galimberti, D., Esposito, F., Marchegiani, F., Cardelli, M., Olivieri, F., Nacmias, B., Sorbi, S., Tagliavini, F., Albani, D., Martinelli Boneschi, F., Binetti, G., ... Franceschi, C. (2011). An APOE haplotype associated with decreased  $\epsilon 4$  expression increases the risk of late onset Alzheimer's disease. *Journal of Alzheimer's Disease: JAD*, *24*(2), 235–245. <https://doi.org/10.3233/JAD-2011-101764>
31. Lescai, F., & Franceschi, C. (2010). The impact of phenocopy on the genetic analysis of complex traits. *PloS One*, *5*(7), e11876. <https://doi.org/10.1371/journal.pone.0011876>
32. Lescai, F., Pirazzini, C., D'Agostino, G., Santoro, A., Ghidoni, R., Benussi, L., Galimberti, D., Federica, E., Marchegiani, F., Cardelli, M., Olivieri, F., Nacmias, B., Sorbi, S., Bagnoli, S., Tagliavini, F., Albani, D., Martinelli Boneschi, F., Binetti, G., Forloni, G., ... Franceschi, C. (2010). Failure to replicate an association of rs5984894 SNP in the PCDH11X gene in a collection of 1,222 Alzheimer's disease affected patients. *Journal of Alzheimer's Disease: JAD*, *21*(2), 385–388. <https://doi.org/10.3233/JAD-2010-091516>
33. Cevenini, E., Bellavista, E., Tieri, P., Castellani, G., Lescai, F., Francesconi, M., Mishto, M., Santoro, A., Valensin, S., Salvioli, S., Capri, M., Zaikin, A., Monti, D., Magalhães, J. P. de, & Franceschi, C. (2010). Systems biology and longevity: An emerging approach to identify innovative anti-aging targets and strategies. *Current Pharmaceutical Design*, *16*(7), 802–813. <https://doi.org/10.2174/138161210790883660>
34. Gravina, S., Lescai, F., Hurteau, G., Brock, G. J., Saramaki, A., Salvioli, S., Franceschi, C., & Roninson, I. B. (2009). Identification of single nucleotide polymorphisms in the p21 (CDKN1A) gene and correlations with longevity in the Italian population. *Aging*, *1*(5), 470–480. <https://doi.org/10.18632/aging.100041>
35. Lescai, F. (2009). Marie Curie fellowships unraveled an interview with Theodosius Lennon, director directorate T, DG Research, European Commission. *New Biotechnology*, *25*(4), 186–187. <https://doi.org/10.1016/j.nbt.2008.12.003>
36. Lescai, F., Marchegiani, F., & Franceschi, C. (2009). PON1 is a longevity gene: Results of a meta-analysis. *Ageing Research Reviews*, *8*(4), 277–284. <https://doi.org/10.1016/j.arr.2009.04.001>
37. Lescai, F., Blanché, H., Nebel, A., Beekman, M., Sahbatou, M., Flachsbar, F., Slagboom, E., Schreiber, S., Sorbi, S., Passarino, G., & Franceschi, C. (2009). Human longevity and 11p15.5: A study in 1321 centenarians. *European Journal of Human Genetics: EJHG*, *17*(11), 1515–1519. <https://doi.org/10.1038/ejhg.2009.54>
38. Lescai, F. (2008). Helping young independent scientists: The EMBO Young Investigator Programme Interview with Gerlind Wallon, Deputy Executive Director, EMBO Young Investigator Programme. *New Biotechnology*, *25*(2-3), 120–121. <https://doi.org/10.1016/j.nbt.2008.08.002>
39. Cevenini, E., Invidia, L., Lescai, F., Salvioli, S., Tieri, P., Castellani, G., & Franceschi, C. (2008). Human models of aging and longevity. *Expert Opinion on Biological Therapy*, *8*(9), 1393–1405. <https://doi.org/10.1517/14712598.8.9.1393>

40. Di Bona, D., Plaia, A., Vasto, S., Cavallone, L., Lescai, F., Franceschi, C., Licastro, F., Colonna-Romano, G., Lio, D., Candore, G., & Caruso, C. (2008). Association between the interleukin-1beta polymorphisms and Alzheimer's disease: A systematic review and meta-analysis. *Brain Research Reviews*, *59*(1), 155–163. <https://doi.org/10.1016/j.brainresrev.2008.07.003>
41. Cardelli, M., Cavallone, L., Marchegiani, F., Oliveri, F., Dato, S., Montesanto, A., Lescai, F., Lisa, R., De Benedictis, G., & Franceschi, C. (2008). A genetic-demographic approach reveals male-specific association between survival and tumor necrosis factor (A/G)-308 polymorphism. *The Journals of Gerontology. Series A, Biological Sciences and Medical Sciences*, *63*(5), 454–460. <https://doi.org/10.1093/gerona/63.5.454>
42. Lescai, F. (2008). The Young European Biotech Network (YEBN). *New Biotechnology*, *25*(1), 34. <https://doi.org/10.1016/j.nbt.2008.04.005>
43. Sevini, F., Santoro, A., Raule, N., Lescai, F., & Franceschi, C. (2007). Role of mitochondrial DNA in longevity, aging and age-related diseases in humans: A reappraisal. *The Italian Journal of Biochemistry*, *56*(4), 243–253.
44. Salvioli, S., Olivieri, F., Marchegiani, F., Cardelli, M., Santoro, A., Bellavista, E., Mishto, M., Invidia, L., Capri, M., Valensin, S., Sevini, F., Cevenini, E., Celani, L., Lescai, F., Gonos, E., Caruso, C., Paolisso, G., De Benedictis, G., Monti, D., & Franceschi, C. (2006). Genes, ageing and longevity in humans: Problems, advantages and perspectives. *Free Radical Research*, *40*(12), 1303–1323. <https://doi.org/10.1080/10715760600917136>
45. Capri, M., Monti, D., Salvioli, S., Lescai, F., Pierini, M., Altilia, S., Sevini, F., Valensin, S., Ostan, R., Bucci, L., & Franceschi, C. (2006). Complexity of anti-immunosenescence strategies in humans. *Artificial Organs*, *30*(10), 730–742. <https://doi.org/10.1111/j.1525-1594.2006.00295.x>
46. Lescai, F., Conti, L., Bartolozzi, M., Ramazzotti, G., Mazzi, M., Sarnicola, V., & Franceschi, C. (2005). Genotype of inflammatory cytokines in limbal stem cell graft in Italian patients. *Biochemical and Biophysical Research Communications*, *332*(1), 95–100. <https://doi.org/10.1016/j.bbrc.2005.04.106>
47. Lescai, F., & Quarta, M. (2003). Young scientist: Italian biotechnologists organize. *Nature*, *425*(6958), 644. <https://doi.org/10.1038/nj6958-644a>
48. Franceschi, C., Valensin, S., Lescai, F., Olivieri, F., Licastro, F., Grimaldi, L. M., Monti, D., De Benedictis, G., & Bonafè, M. (2001). Neuroinflammation and the genetics of Alzheimer's disease: The search for a pro-inflammatory phenotype. *Aging (Milan, Italy)*, *13*(3), 163–170. <https://doi.org/10.1007/BF03351475>
49. Franceschi, C., Motta, L., Valensin, S., Rapisarda, R., Franzone, A., Berardelli, M., Motta, M., Monti, D., Bonafè, M., Ferrucci, L., Deiana, L., Pes, G. M., Carru, C., Desole, M. S., Barbi, C., Sartoni, G., Gemelli, C., Lescai, F., Olivieri, F., ... Baggio, G. (2000). Do men and women follow different trajectories to reach extreme longevity? Italian Multicenter Study on Centenarians (IMUSCE). *Aging (Milan, Italy)*, *12*(2), 77–84. <https://doi.org/10.1007/BF03339894>