

PUBLICATIONS

27) First mitochondrial genome wide association study with metabolomics.

Brahim Aboulmaouahib, Gabi Kastenmüller, Karsten Suhre, Sebastian Zöllner, Hansi Weissensteiner, Cornelia Prehn, Jerzy Adamski, Christian Gieger, Rui Wang-Sattler, Peter Lichtner, Konstantin Strauch, **Antònia Flaquer.** (2021). Human Molecular Genetics.

<https://doi.org/10.1093/hmg/ddab312>

26) Biomolecular insights into North African-related ancestry, mobility and diet in eleventh-century Al-AndalusMari.

Marina Silva, Gonzalo Oteo-García, Rui Martiniano, João Guimarães, Matthew von Tersch, Ali Madour, Tarek Shoeib, Alessandro Fichera, Pierre Justeau, M. George B. Foody, Krista McGrath, Amparo Barrachina, Vicente Palomar, Katharina Dulias, Bobby Yau, Francesca Gandini, Douglas J. Clarke, Alexandra Rosa, António Brehm, **Antònia Flaquer,** Teresa Rito, Anna Olivieri, Alessandro Achilli, Antonio Torroni, Alberto Gómez-Carballa, Antonio Salas, Jaroslaw Bryk, Peter W. Ditchfield, Michelle Alexander, Maria Pala, Pedro A. Soares, Ceiridwen J. Edwards & Martin B. Richards.(2021). Scientific Reports. 11:18121

<https://doi.org/10.1038/s41598-021-95996-3>

25) The conserved ASTN2/BRINP1 locus at 9q33.1–33.2 is associated with major psychiatric disorders in a large pedigree from Southern Spain.

Josep Pol-Fuster, Francesca Cañellas, Laura Ruiz-Guerra, Aina Medina-Dols, Bàrbara Bisbal-Carrió, Bernat Ortega-Vila, Jaume Llinàs, Jessica Hernandez-Rodriguez, Jerònia Lladó, Gabriel Olmos, Konstantin Strauch, Damià Heine-Suñer, Cristòfol Vives-Bauzà, **Antònia Flaquer.** (2021). Scientific Reports. 11:14529

<https://doi.org/10.1038/s41598-021-93555-4>

24) Prediction of type 2 diabetes mellitus based on nutrition data.

Andreas Katsimpris, Aboulmaouahib Brahim, Wolfgang Rathmann, Anette Peters, Konstantin Strauch, **Antònia Flaquer.** (2021). Journal of Nutritional Science. vol. 10, e46.

<https://doi.org/10.1017/jns.2021.36>

23) Familial Psychosis Associated With a Missense Mutation at MACF1 Gene Combined With the Rare Duplications DUP3p26.3 and DUP16q23.3. Affecting the CNTN6 and CDH13 Genes

Pol-Fuster Josep, Cañellas Francesca, Ruiz-Guerra Laura, Medina-Dols Aina, Bisbal-Carrió Bàrbara, Asensio Víctor, Ortega-Vila Bernat, Marzese Diego, Vidal Carme, Santos Carmen, Lladó Jerònia, Olmos Gabriel, Heine-Suñer Damià, Strauch Konstantin, **Flaquer Antònia,** Vives-Bauzà Cristòfol. (2021). Frontiers in Genetics12: 497.

<https://doi.org/10.3389/fgene.2021.622886>

- 22) Mitochondrial GWA analysis of lipid profile identifies genetic variants to be associated with HDL cholesterol and triglyceride levels
Flaquer A, Rospleszcz S, Reischl E, Zeilinger S, Prokisch H, Meitinger T, Meisinger C, Peters A, Waldenberger M, Grallert H, Strauch K. (2015). *PLoS ONE* 10(5): e0126294. doi:10.1371/journal.pone.0126294.
- 21) Mitochondrial genetic variants identified to be associated with posttraumatic stress disorder
Flaquer A, Baumbach C, Ladwig KH, Kriebel J, Waldenberger M, Grallert H, Baumert J, Meitinger T, Kruse J, Peters A, Emeny R, Strauch K. (2015). *Transl Psychiatry*, 5, e524. doi:10.1038/tp.20.
- 20) Mitochondrial Genetic Variants Identified to Be Associated with BMI in Adults
Flaquer A, Baumbach C, Kriebel J, Meitinger T, Peters A, Waldenberger M, Grallert H, Strauch K. (2014). *PLoS ONE* 9(8): e105116. doi:10.1371/journal.pone.0105116.
- 19) Association study of mitochondrial genetic polymorphisms in asthmatic children
Flaquer A, Heinzmann A, Rospleszcz S, Mailaparambil B, Dietrich H, Strauch K, Grychtol R. (2014). *Mitochondrion*, 14(1):49-53. doi: 10.1016/j.mito.2013.11.002.
- 18) Genome-wide linkage analysis of congenital heart defects using MOD score analysis identifies two novel loci
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- 17) A comparison of different linkage statistics in small to moderate sized pedigrees with complex diseases.
Flaquer A and Strauch K. (2012) *BMC Research Notes* 5:411.
- 16) A new susceptibility locus for bipolar affective disorder in PAR1 on Xp22.3/Yp11.3
Flaquer A, Jamra RA, Etterer K, Díaz GO, Rivas F, Rietschel M, Cichon S, Nöthen MM, Strauch K. (2010) *American Journal of Medical Genetics. Part B. Neuropsychiatric Genetics*. 153B(5):1110–14.
- 15) A new sex-specific genetic map of the human pseudoautosomal regions (PAR1 and PAR2)
Flaquer A, Fischer C, Wienker TF. (2009) *Human Heredity* 68(3):192-200.
- 14) Evaluation of potential power gain with imputed genotypes in genome-wide association studies.
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- 13) Genetic association study of the P-type ATPase ATP13A2 in late-onset Parkinson's disease.
Rakovic A, Stiller B, Djarmati A, **Flaquer A**, Freudenberg J, Toliat MR, Linnebank M, Kostic V, Lohmann K, Paus S, Nürnberg P, Kubisch C, Klein C, Wüllner U, Ramirez A. (2009) *Movement Disorders: official journal of the Movement Disorder Society* 24(3):429-33.
- 12) Susceptibility variants for male-pattern baldness on chromosome 20p11.
Hillmer AM, Brockschmidt FF, Hanneken S, Eigelshoven S, Steffens M, **Flaquer A**, Herms S, Becker T, Kortüm AK, Nyholt DR, Zhao ZZ, Montgomery GW, Martin NG, Mühleisen TW, Alblas MA, Moebus S, Jöckel KH, Bröcker-preuss M, Erbel R, Reinartz R. et al. (2008) *Nature genetics* 40(11):1279-81.
- 11) Genome-wide SNP-based linkage scan identifies a locus on 8q24 for an age-related hearing impairment trait.
Huyghe JR, Van laer L, Hendrickx JJ, Fransen E, Demeester K, Topsakal V, Kunst S, Manninen M, Jensen M, Bonaconsa A, Mazzoli M, Baur M, Hannula S, Mäki-torkko E, Espeso A, Van eyken E, **Flaquer A**, Becker C, Stephens D, Sorri M, et al. (2008) *American Journal of Human Genetics* 83(3):401-07.
- 10) The human pseudoautosomal regions: a review for genetic epidemiologists.

- Flaquer A**, Rappold GA, Wienker TF, Fischer C. (2008) *European Journal of Human Genetics* 16: 771-779.
- 9) Genome-wide scan and fine-mapping linkage study of androgenetic alopecia reveals a locus on chromosome 3q26.
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- 8) The R620W polymorphism in PTPN22 confers general susceptibility for the development of alopecia areata.
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- 6) Linkage analysis of alcohol dependence using MOD scores
Strauch K, Fürst R, Rüschenhoff F, Windemuth C, Dietter J, **Flaquer A**, Baur MP, Wienker TF. (2006) *BMC Genetics Suppl* 1:S162.
- 5) Genomewide scan and fine-mapping linkage studies in four European samples with bipolar affective disorder suggest a new susceptibility locus on chromosome 1p35-p36 and provides further evidence of loci on chromosome 4q31 and 6q24.
Schumacher J, Kaneva R, Jamra RA, Diaz GO, Ohlraun S, Milanova V, Lee YA, Rivas F, Mayoral F, Fuerst R, **Flaquer A**, Windemuth C, Gay E, Sanz S, González MJ, Gil S, Cabaleiro F, Del rio F, Perez F, Haro J, et al. (2005) *American Journal of Human Genetics* 77(6):1102-11.
- 4) Genetic variation in the human androgen receptor gene is the major determinant of common early-onset androgenetic alopecia.
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- 3) Fine mapping of 10q and 18q for familial Alzheimer's disease in Caribbean Hispanics.
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- 2) Genetic influences on life span and survival among elderly African-Americans, Caribbean Hispanics, and Caucasians.
Lee JH, **Flaquer A**, Costa R, Andrews H, Cross P, Lantigua R, Schupf N, Tang MX, Mayeux R. (2004) *American Journal of Medical Genetics: Part A* 128A(2):159-6.
- 1) Genetic influences on memory performance in familial Alzheimer disease.
Lee JH, **Flaquer A**, Stern Y, Tycko B, Mayeux R. (2004) *Neurology* 62(3):414-22.
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BIBLIOGRAPHICALLY CITABLE ABSTRACTS OF LECTURES AND POSTERS

Mitochondrial genome-wide association study of pancreatic cancer

Brahim Aboulmaouhib, **Antònia Flaquer**, Martina Müller-Nurasyid, Peter Lichtner, Detlef K. Bartsch, Emily P. Slater, Konstantin Strauch. IGES 2022, Paris, France.

Mitochondrial GWA analysis in several complex diseases using the KORA population

Antònia Flaquer, Hanna-Theresa Heier, Karl-Heinz Ladwig, Melanie Waldenberger, Harald Grallert, Christa Meisinger, Thomas Meitinger, Annette Peters, Konstantin Strauch. (2016) Journal of World Mitochondria Society 2(2)

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Antònia Flaquer, Karl-Heinz Ladwig, Rebecca Emeny, Melanie Waldenberger, Harald Grallert, Stephan Weidinger, Christa Meisinger, Thomas Meitinger, Annette Peters, Konstantin Strauch. 2014. International Genetic Epidemiology Society (IGES). Vienna, Austria. ISBN 978-1-940377-01-8. Available through: <http://www.geneticepi.org/meeting-abstracts>

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Why does linkage analysis often fail with complex diseases

Antonia Flaquer and Konstantin Strauch. International Genetic Epidemiology Society (IGES). September 18, 2011. Heidelberg, Germany. *Genetic Epidemiology* 36(2):S130. <http://onlinelibrary.wiley.com/doi/10.1002/gepi.2012.36.issue-2/issuetoc>

Pooled DNA in whole genome association studies with single nucleotide polymorphism (SNP) arrays.

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A genome-wide association study identifies new susceptibility variants for male pattern baldness on chromosome 20.

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A genetic epidemiologist's view on the human pseudoautosomal regions

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Lee JH, Mayeux R, Mayo D, Santana V, Williamson J, **Flaquer A**, Ciappa A, Rondon H, Estevez P, Lantigua R, Medrano M, Torres M, Stern Y, Tycko B, Knowles JA. Meeting of The American Society of Human Genetics. November 4, 2003. Los Angeles, USA.

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Mitochondrial GWA analysis in several complex diseases using the KORA population

Antònia Flaquer, Hanna-Theresa Heier, Karl-Heinz Ladwig, Melanie Waldenberger, Harald Grallert, Christa Meisinger, Thomas Meitinger, Annette Peters, Konstantin Strauch. 2014. DZNE Deutsches Zentrum für Neurodegenerative Erkrankungen in der Helmholtz-Gemeinschaft. Germany.

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