



Publications **X-omics** consortium

2023

## Publications of the X-omics consortium

### December

*A monoacylglycerol lipase inhibitor showing therapeutic efficacy in mice without central side effects or dependence.*

Jiang M, Huizenga MCW, Wirt JL, Paloczi J, Amedi A, van den Berg RJBHN, Benz J, Collin L, Deng H, Di X, Driever WF, Florea BI, Grether U, Janssen APA, **Hankemeier T**, Heitman LH, Lam TW, Mohr F, Pavlovic A, Ruf I, van den Hurk H, Stevens AF, van der Vliet D, van der Wel T, Wittwer MB, van Boeckel CAA, Pacher P, Hohmann AG, van der Stelt M.

Nat Commun. 2023 Dec 5;14(1):8039. doi: 10.1038/s41467-023-43606-3. PMID: 38052772

<https://pubmed.ncbi.nlm.nih.gov/38052772/>

*Application of a capillary electrophoresis-mass spectrometry metabolomics workflow in zebrafish larvae reveals new effects of cortisol.*

van Mever M, Mamani-Huanca M, Faught E, López-González Á, **Hankemeier T**, Barbas C, Schaaf MJM, Ramautar R.

Electrophoresis. 2023 Dec 10. doi: 10.1002/elps.202300186. Online ahead of print. PMID: 38072651

<https://pubmed.ncbi.nlm.nih.gov/38072651/>

*Into the Dark Serum Proteome: Personalized Features of IgG1 and IgA1 Repertoires in Severe COVID-19 Patients.*

Bondt A, Hoek M, Dingess K, Tamara S, de Graaf B, Peng W, den Boer MA, Damen M, Zwart C, Barendregt A, van Rijswijk DMH, Schulte D, Grobber M, Tejjani K, van Rijswijk J, Völlmy F, Snijder J, Fortini F, Papi A, Volta CA, Campo G, Contoli M, van Gils MJ, Spadaro S, Rizzo P, **Heck AJR**.

Mol Cell Proteomics. 2023 Dec 6;23(1):100690. doi: 10.1016/j.mcpro.2023.100690. Online ahead of print. PMID: 38065436

<https://pubmed.ncbi.nlm.nih.gov/38065436/>

*CD5L is a canonical component of circulatory IgM.*

Oskam N, den Boer MA, Lukassen MV, Ooijevaar-de Heer P, Veth TS, van Mierlo G, Lai SH, Derksen NIL, Yin V, Streutker M, Franc V, Šiborová M, Damen MJA, Kos D, Barendregt A, Bondt A, van Goudoever JB, de Haas CJC, Aerts PC, Muts RM, Rooijackers SHM, Vidarsson G, Rispens T, **Heck AJR**.

Proc Natl Acad Sci U S A. 2023 Dec 12;120(50):e2311265120. doi: 10.1073/pnas.2311265120. Epub 2023 Dec 6. PMID: 38055740

<https://pubmed.ncbi.nlm.nih.gov/38055740/>

*Proteoform-resolved profiling of plasminogen activation reveals novel abundant phosphorylation site and primary N-terminal cleavage site.*

Cramer DAT, Yin V, Caval T, Franc V, Yu D, Wu G, Lloyd G, Langendorf C, Whisstock JC, Law RHP, **Heck AJR**.

Mol Cell Proteomics. 2023 Dec 13:100696. doi: 10.1016/j.mcpro.2023.100696. Online ahead of print. PMID: 38101751

<https://pubmed.ncbi.nlm.nih.gov/38101751/>

*Direct Comparison of the Hinge-Cleaving Proteases IgdE and BdpK for LC-MS-Based IgG1 Clonal Profiling.*

van Rijswijck DMH, Bondt A, de Kat N, Lood R, **Heck AJR**.

Anal Chem. 2023 Dec 17. doi: 10.1021/acs.analchem.3c03712. Online ahead of print. PMID: 38105593

<https://pubmed.ncbi.nlm.nih.gov/38105593/>

*The relationship of school performance with self-control and grit is strongly genetic and weakly causal.*

Kevenaar ST, van Bergen E, Oldehinkel AJ, **Boomsma DI**, Dolan CV.

NPJ Sci Learn. 2023 Dec 4;8(1):53. doi: 10.1038/s41539-023-00198-3. PMID: 38049407

<https://pubmed.ncbi.nlm.nih.gov/38049407/>

*Genome-wide association study meta-analysis of dizygotic twinning illuminates genetic regulation of female fecundity.*

Mbarek H, Gordon SD, Duffy DL, Hubers N, Mortlock S, Beck JJ, Hottenga JJ, Pool R, Dolan CV, Actkins KV, Gerring ZF, Van Dongen J, Ehli EA, Iacono WG, Mcgue M, Chasman DI, Gallagher CS, Schilit SLP, Morton CC, Paré G, Willemsen G, Whiteman DC, Olsen CM, Derom C, Vlietinck R, Gudbjartsson D, Cannon-Albright L, Krapohl E, Plomin R, Magnusson PKE, Pedersen NL, Hysi P, Mangino M, Spector TD, Palviainen T, Milaneschi Y, Penninx BW, Campos AI, Ong KK, Perry JRB, Lambalk CB, Kaprio J, Ólafsson Í, Durore K, Revenu C, Rentería ME, Yengo L, Davis L, Derks EM, Medland SE, Stefansson H, Stefansson K, Del Bene F, Reversade B, Montgomery GW, **Boomsma DI**, Martin NG.

Hum Reprod. 2023 Dec 5:dead247. doi: 10.1093/humrep/dead247. Online ahead of print. PMID: 38052102

<https://pubmed.ncbi.nlm.nih.gov/38052102/>

*New insights into the (epi)genetics of twinning.*

van Dongen J, Hubers N, **Boomsma DI**.

Hum Reprod. 2023 Dec 5;dead131. doi: 10.1093/humrep/dead131. Online ahead of print. PMID: 38052159

<https://pubmed.ncbi.nlm.nih.gov/38052159/>

*Longitudinal multi-omics study reveals common etiology underlying association between plasma proteome and BMI trajectories in adolescent and young adult twins.*

Drouard G, Hagenbeek FA, Whipp AM, Pool R, Hottenga JJ, Jansen R, Hubers N, Afonin A; BIOS Consortium, BBMRI-N. L. Metabolomics Consortium; Willemsen G, de Geus EJC, Ripatti S, Pirinen M, Kanninen KM, **Boomsma DI**, van Dongen J, Kaprio J.

BMC Med. 2023 Dec 21;21(1):508. doi: 10.1186/s12916-023-03198-7. PMID: 38129841

<https://pubmed.ncbi.nlm.nih.gov/38129841/>

*Trajectories of kidney function and risk of mortality.*

van der Burgh AC, Sedaghat S, **Ikram MA**, Hoorn EJ, Chaker L.

Int J Epidemiol. 2023 Dec 25;52(6):1959-1967. doi: 10.1093/ije/dyad111. PMID: 37649343

<https://pubmed.ncbi.nlm.nih.gov/37649343/>

*WOMBAT-P: Benchmarking Label-Free Proteomics Data Analysis Workflows.*

Bouyssié D, Altiner P, Capella-Gutierrez S, Fernández JM, Hagemeyer YP, **Horvatovich P**, Hubálek M, Levander F, Mauri P, Palmblad M, Raffelsberger W, Rodríguez-Navas L, Di Silvestre D, Kunkli BT, Uszkoreit J, Vandenbrouck Y, Vizcaíno JA, Winkelhardt D, Schwämmle V.

J Proteome Res. 2023 Dec 1. doi: 10.1021/acs.jproteome.3c00636. Online ahead of print. PMID: 38038272

<https://pubmed.ncbi.nlm.nih.gov/38038272/>

*PANDORA v2.0: Benchmarking peptide-MHC II models and software improvements.*

Parizi FM, Marzella DF, Ramakrishnan G, **'t Hoen PAC**, Karimi-Jafari MH, Xue LC.

Front Immunol. 2023 Dec 8;14:1285899. doi: 10.3389/fimmu.2023.1285899. eCollection 2023. PMID: 38143769

<https://pubmed.ncbi.nlm.nih.gov/38143769/>

*Serum bile acids associate with liver volume in polycystic liver disease and decrease upon treatment with lanreotide.*

Dekker SEI, Bierau J, **Giera M**, Blomberg N, Drenth JPH, Mayboroda OA, de Fijter JW, Soonawala D; DIPAK Consortium.

*Eur J Clin Invest.* 2023 Dec 9:e14147. doi: 10.1111/eci.14147. Online ahead of print. PMID: 38071418

<https://pubmed.ncbi.nlm.nih.gov/38071418/>

*Totum-070, a Polyphenol-Rich Plant Extract, Prevents Hypercholesterolemia in High-Fat Diet-Fed Hamsters by Inhibiting Intestinal Cholesterol Absorption.*

Langhi C, Vallier M, Otero YF, Maura M, Le Joubioux F, Groult H, Achour O, Pebriana RB, **Giera M**, Guigas B, Maugard T, Chassaing B, Peltier S, Bard JM, Sirvent P.

*Nutrients.* 2023 Dec 9;15(24):5056. doi: 10.3390/nu15245056. PMID: 38140315

<https://pubmed.ncbi.nlm.nih.gov/38140315/>

*Assessment of IgG-Fc glycosylation from individual RhD-specific B cell clones reveals regulation at clonal rather than clonotypic level.*

de Graaf EL, Larsen MD, van der Bolt N, Visser R, Verhagen OJHM, Hipgrave Ederveen AL, Koeleman CAM, van der Schoot CE, **Wuhrer M**, Vidarsson G.

*Immunology.* 2023 Dec 14. doi: 10.1111/imm.13737. Online ahead of print. PMID: 38097893

<https://pubmed.ncbi.nlm.nih.gov/38097893/>

*Mapping of the gene network that regulates glycan clock of ageing.*

Frkatović-Hodžić A, Mijakovac A, Miškec K, Nostaeva A, Sharapov SZ, Landini A, Haller T, Akker EVD, Sharma S, Cuadrat RRC, Mangino M, Li Y, Keser T, Rudman N, Štambuk T, Pučić-Baković M, Trbojević-Akmačić I, Gudelj I, Štambuk J, Pribić T, Radovani B, Tominac P, Fischer K, Beekman M, **Wuhrer M**, Gieger C, Schulze MB, Wittenbecher C, Polasek O, Hayward C, Wilson JF, Spector TD, Köttgen A, Vučković F, Aulchenko YS, Vojta A, Krištić J, Klarić L, Zoldoš V, Lauc G.

*Ageing (Albany NY).* 2023 Dec 26;15. doi: 10.18632/aging.205106. Online ahead of print. PMID: 38149987

<https://pubmed.ncbi.nlm.nih.gov/38149987/>

*DNA methylation epesignature, extension of the clinical features and comparative epigenomic profiling of Hao-Fountain syndrome caused by variants in USP7.*

van der Laan L, Karimi K, Rooney K, Lauffer P, McConkey H, Caro P, Relator R, Levy MA, Bhai P, Mignot C, Keren B, Briuglia S, Sobering AK, Li D, **Vissers LELM**, Dingemans AJM, Valenzuela I, Verberne EA, Misra-Isrie M, Zwijnenburg PJG, Waisfisz Q, Alders M, Sailer S, Schaaf CP, Mannens MMAM, Sadikovic B, van Haelst MM, Henneman P.

Genet Med. 2023 Dec 18:101050. doi: 10.1016/j.gim.2023.101050. Online ahead of print.  
PMID: 38126281

<https://pubmed.ncbi.nlm.nih.gov/38126281/>

## November

*Cytogenetics Is a Science, Not a Technique! Why Optical Genome Mapping Is So Important to Clinical Genetic Laboratories.*

Smith AC, **Hoischen A**, Raca G.

Cancers (Basel). 2023 Nov 19;15(22):5470. doi: 10.3390/cancers15225470. PMID: 38001730

<https://pubmed.ncbi.nlm.nih.gov/38001730/>

*Biallelic MAD2L1BP (p31comet) mutation is associated with mosaic aneuploidy and juvenile granulosa cell tumors.*

Abdel-Salam GMH, Hellmuth S, Gradhand E, Käseberg S, Winter J, Pabst AS, Eid MM, Thiele H, Nürnberg P, Budde BS, Toliat MR, Brecht IB, Schroeder C, Gschwind A, Ossowski S, Häuser F, Rossmann H, Abdel-Hamid MS, Hegazy I, Mohamed AG, Schneider DT, Bertoli-Avella A, Bauer P, Pearing JN, Pfundt R, **Hoischen A**, **Gilissen C**, Strand D, Zechner U, Tashkandi SA, Faqeh EA, Stemmann O, Strand S, Bolz HJ.

JCI Insight. 2023 Nov 22;8(22):e170079. doi: 10.1172/jci.insight.170079. PMID: 37796616

<https://pubmed.ncbi.nlm.nih.gov/37796616/>

*Prevalence of chromosomal alterations in first-trimester spontaneous pregnancy loss.*

Essers R, Lebedev IN, Kurg A, Fonova EA, Stevens SJC, Koeck RM, von Rango U, Brandts L, Deligiannis SP, Nikitina TV, Sazhenova EA, Tolmacheva EN, Kashevarova AA, Fedotov DA, Demeneva VV, Zhigalina DI, Drozdov GV, Al-Nasiry S, Macville MVE, van den Wijngaard A, Dreesen J, Paulussen A, **Hoischen A**, Brunner HG, Salumets A, Zamani Esteki M.

Nat Med. 2023 Nov 23. doi: 10.1038/s41591-023-02645-5. Online ahead of print. PMID: 37996709

<https://pubmed.ncbi.nlm.nih.gov/37996709/>

*Higher plasma levels of endocannabinoids and analogues correlate with a worse cardiometabolic profile in young adults.*

Di X, Martinez-Tellez B, Krekels EHJ, Jurado-Fasoli L, Osuna-Prieto FJ, Ortiz-Alvarez L, **Hankemeier T**, Rensen PCN, Ruiz JR, Kohler I.

J Clin Endocrinol Metab. 2023 Nov 15:dgad668. doi: 10.1210/clinem/dgad668. Online ahead of print. PMID: 37967236

<https://pubmed.ncbi.nlm.nih.gov/37967236/>

*Artificial surface labelling of Escherichia coli with StrepTagII antigen to study how monoclonal antibodies drive complement-mediated killing.*

Muts RM, den Boer MA, Bardoel BW, Aerts PC, de Haas CJC, Heck AJR, Rooijackers SHM, Heesterbeek DAC.

Sci Rep. 2023 Nov 1;13(1):18836. doi: 10.1038/s41598-023-46026-x. PMID: 37914798

<https://pubmed.ncbi.nlm.nih.gov/37914798/>

*Trans-ancestry epigenome-wide association meta-analysis of DNA methylation with lifetime cannabis use.*

Fang F, Quach B, Lawrence KG, van Dongen J, Marks JA, Lundgren S, Lin M, Odintsova VV, Costeira R, Xu Z, Zhou L, Mandal M, Xia Y, Vink JM, Bierut LJ, Ollikainen M, Taylor JA, Bell JT, Kaprio J, Boomsma DI, Xu K, Sandler DP, Hancock DB, Johnson EO.

Mol Psychiatry. 2023 Nov 7. doi: 10.1038/s41380-023-02310-w. Online ahead of print. PMID: 37935791

<https://pubmed.ncbi.nlm.nih.gov/37935791/>

*Gene-educational attainment interactions in a multi-population genome-wide meta-analysis identify novel lipid loci.*

de Las Fuentes L, Schwander KL, Brown MR, Bentley AR, Winkler TW, Sung YJ, Munroe PB, Miller CL, Aschard H, Aslibekyan S, Bartz TM, Bielak LF, Chai JF, Cheng CY, Dorajoo R, Feitosa MF, Guo X, Hartwig FP, Horimoto A, Kolčič I, Lim E, Liu Y, Manning AK, Marten J, Musani SK, Noordam R, Padmanabhan S, Rankinen T, Richard MA, Ridker PM, Smith AV, Vojinovic D, Zonderman AB, Alver M, Boissel M, Christensen K, Freedman BI, Gao C, Giulianini F, Harris SE, He M, Hsu FC, Kühnel B, Laguzzi F, Li X, Lyytikäinen LP, Nolte IM, Poveda A, Rauramaa R, Riaz M, Robino A, Sofer T, Takeuchi F, Tayo BO, van der Most PJ, Verweij N, Ware EB, Weiss S, Wen W, Yanek LR, Zhan Y, Amin N, Arking DE, Ballantyne C, Boerwinkle E, Brody JA, Broeckel U, Campbell A, Canouil M, Chai X, Chen YI, Chen X, Chitrala KN, Concas MP, de Faire U, de Mutsert R, de Silva HJ, de Vries PS, Do A, Faul JD, Fisher V, Floyd JS, Forrester T, Friedlander Y, Girotto G, Gu CC, Hallmans G, Heikkinen S, Heng CK, Homuth G, Hunt S, Ikram MA, Jacobs DR Jr, Kavousi M, Khor CC, Kilpeläinen TO, Koh WP, Komulainen P, Langefeld CD, Liang J, Liu K, Liu J, Lohman K, Mägi R, Manichaikul AW, McKenzie CA, Meitinger T, Milaneschi Y, Nauck M, Nelso... See abstract for full author list →

Front Genet. 2023 Nov 2;14:1235337. doi: 10.3389/fgene.2023.1235337. eCollection 2023. PMID: 38028628

<https://pubmed.ncbi.nlm.nih.gov/38028628/>

*Bone mineral density and the risk of incident dementia: A meta-analysis.*

Lary CW, Ghatan S, Gerety M, Hinton A, Nagarajan A, Rosen C, Ross RD, Bennett DA, DeStefano AL, Ikram MA, Rivadeneira F, Kiel DP, Seshadri S, Beiser A.

J Am Geriatr Soc. 2023 Nov 7. doi: 10.1111/jgs.18638. Online ahead of print. PMID: 37933827

<https://pubmed.ncbi.nlm.nih.gov/37933827/>



*Serum immunoglobulins and biomarkers of dementia: a population-based study.*

Yaqub A, Khan SR, Vernooij MW, van Hagen PM, Peeters RP, **Ikram MA**, Chaker L, Dalm VASH.

*Alzheimers Res Ther.* 2023 Nov 7;15(1):194. doi: 10.1186/s13195-023-01333-3.  
PMID: 37936180

<https://pubmed.ncbi.nlm.nih.gov/37936180/>

*Plasma circulating microRNAs associated with blood-based immune markers: a population-based study.*

Leonard S, Karabegović I, **Ikram MA**, Ahmad S, Ghanbari M.

*Clin Exp Immunol.* 2023 Nov 9:uxad126. doi: 10.1093/cei/uxad126. Online ahead of print.  
PMID: 37950349

<https://pubmed.ncbi.nlm.nih.gov/37950349/>

*Where is VALDO? VAScular Lesions Detection and segmentatiOn challenge at MICCAI 2021.*

Sudre CH, Van Wijnen K, Dubost F, Adams H, Atkinson D, Barkhof F, Birhanu MA, Bron EE, Camarasa R, Chaturvedi N, Chen Y, Chen Z, Chen S, Dou Q, Evans T, Ezhov I, Gao H, Girones Sanguesa M, Gispert JD, Gomez Anson B, Hughes AD, **Ikram MA**, Ingala S, Jaeger HR, Kofler F, Kuijf HJ, Kutnar D, Lee M, Li B, Lorenzini L, Menze B, Molinuevo JL, Pan Y, Puybareau E, Rehwald R, Su R, Shi P, Smith L, Tillin T, Tochon G, Urien H, van der Velden BHM, van der Velpen IF, Wiestler B, Wolters FJ, Yilmaz P, de Groot M, Vernooij MW, de Bruijne M; ALFA study.

*Med Image Anal.* 2023 Nov 19;91:103029. doi: 10.1016/j.media.2023.103029. Online ahead of print. PMID: 37988921

<https://pubmed.ncbi.nlm.nih.gov/37988921/>

*Advanced glycation end products measured by skin autofluorescence and subclinical cardiovascular disease: the Rotterdam Study.*

Chen J, Arshi B, Waqas K, Lu T, Bos D, **Ikram MA**, Uitterlinden AG, Kavousi M, Zillikens MC.

*Cardiovasc Diabetol.* 2023 Nov 28;22(1):326. doi: 10.1186/s12933-023-02052-7.  
PMID: 38017418

<https://pubmed.ncbi.nlm.nih.gov/38017418/>

*A bronchial gene signature specific for severe COPD that is retained in the nose.*

van Nijnatten J, Faiz A, Timens W, **Guryev V**, Slebos DJ, Klooster K, Hartman JE, Kole T, Choy DF, Chakrabarti A, Grimbaldeston M, Rosenberger CM, Kerstjens H, Brandsma CA, van den Berge M.

ERJ Open Res. 2023 Nov 27;9(6):00354-2023. doi: 10.1183/23120541.00354-2023. eCollection 2023 Nov. PMID: 38020574

<https://pubmed.ncbi.nlm.nih.gov/38020574/>

*FastCAR: fast correction for ambient RNA to facilitate differential gene expression analysis in single-cell RNA-sequencing datasets.*

Berg M, Petoukhov I, van den Ende I, Meyer KB, **Guryev V**, Vonk JM, Carpaij O, Banchemo M, Hendriks RW, van den Berge M, Nawijn MC.

BMC Genomics. 2023 Nov 29;24(1):722. doi: 10.1186/s12864-023-09822-3. PMID: 38030970

<https://pubmed.ncbi.nlm.nih.gov/38030970/>

*Fatty acid desaturation and lipoxygenase pathways support trained immunity.*

Ferreira AV, Alarcon-Barrera JC, Domínguez-Andrés J, Bulut Ö, Kilic G, Debisarun PA, Röring RJ, Özhan HN, Terschlüsen E, Ziogas A, Kostidis S, Mohammed Y, Matzaraki V, Renieris G, Giamarellos-Bourboulis EJ, Netea MG, **Giera M**.

Nat Commun. 2023 Nov 15;14(1):7385. doi: 10.1038/s41467-023-43315-x. PMID: 37968313

<https://pubmed.ncbi.nlm.nih.gov/37968313/>

*Impaired gut microbiota-mediated short-chain fatty acid production precedes morbidity and mortality in people with HIV.*

Sereti I, Verburgh ML, Gifford J, Lo A, Boyd A, Verheij E, Verhoeven A, Wit FWNM, Schim van der Loeff MF, **Giera M**, Kootstra NA, Reiss P, Vujkovic-Cvijin I.

Cell Rep. 2023 Nov 28;42(11):113336. doi: 10.1016/j.celrep.2023.113336. Epub 2023 Nov 1. PMID: 37918403

<https://pubmed.ncbi.nlm.nih.gov/37918403/>

*Unraveling the impact of sialic acids on the immune landscape and immunotherapy efficacy in pancreatic cancer.*

Boelaars K, Goossens-Kruijssen L, Wang D, de Winde CM, Rodriguez E, Lindijer D, Springer B, van der Haar Àvila I, de Haas A, Wehry L, Boon L, Mebius RE, van Montfoort N, **Wuhrer M**, den Haan JMM, van Vliet SJ, van Kooyk Y.

J Immunother Cancer. 2023 Nov;11(11):e007805. doi: 10.1136/jitc-2023-007805.  
PMID: 37940346

<https://pubmed.ncbi.nlm.nih.gov/37940346/>

*A multi-platform approach to identify a blood-based host protein signature for distinguishing between bacterial and viral infections in febrile children (PERFORM): a multi-cohort machine learning study.*

Jackson HR, Zandstra J, Menikou S, Hamilton MS, McArdle AJ, Fischer R, Thorne AM, Huang H, Tanck MW, Jansen MH, De T, Agyeman PKA, Von Both U, Carrol ED, Emonts M, Eleftheriou I, Van der Flier M, Fink C, **Gloerich J**, De Groot R, Moll HA, Pokorn M, Pollard AJ, Schlapbach LJ, Tsolia MN, Usuf E, Wright VJ, Yeung S, Zavadska D, Zenz W, Coin LJM, Casals-Pascual C, Cunningham AJ, Martinon-Torres F, Herberg JA, de Jonge MI, Levin M, Kuijpers TW, Kaforou M; PERFORM consortium.

Lancet Digit Health. 2023 Nov;5(11):e774-e785. doi: 10.1016/S2589-7500(23)00149-8.  
PMID: 37890901

<https://pubmed.ncbi.nlm.nih.gov/37890901/>

*Targeted perturbation of signaling-driven condensates.*

Gui T, Fleming C, Manzato C, Bourgeois B, Sirati N, Heuer J, Papadionysiou I, Montfort DIV, Gijzen MV, Smits LMM, **Burgering BMT**, Madl T, Schuijers J.

Mol Cell. 2023 Nov 16;83(22):4141-4157.e11. doi: 10.1016/j.molcel.2023.10.023. Epub 2023 Nov 16. PMID: 37977121

<https://pubmed.ncbi.nlm.nih.gov/37977121/>

## October

*Biallelic MAD2L1BP (p31comet) mutation is associated with mosaic aneuploidy and juvenile granulosa cell tumors.*

Abdel-Salam GMH, Hellmuth S, Gradhand E, Käseberg S, Winter J, Pabst AS, Eid MM, Thiele H, Nürnberg P, Budde BS, Toliat MR, Brecht IB, Schroeder C, Gschwind A, Ossowski S, Häuser F, Rossmann H, Abdel-Hamid MS, Hegazy I, Mohamed AG, Schneider DT, Bertoli-Avella AM, Bauer P, Pearing JN, Pfundt R, **Hoischen A**, **Gilissen C**, Strand D, Zechner U, Tashkandi SA, Faqeih EA, Stemmann O, Strand S, Bolz HJ.

JCI Insight. 2023 Oct 5:e170079. doi: 10.1172/jci.insight.170079. Online ahead of print. PMID: 37796616

<https://pubmed.ncbi.nlm.nih.gov/37796616/>

*Systematic analysis of paralogous regions in 41,755 exomes uncovers clinically relevant variation.*

Steyaert W, Haer-Wigman L, Pfundt R, Hellebrekers D, Steehouwer M, Hampstead J, de Boer E, Stegmann A, Yntema H, Kamsteeg EJ, Brunner H, **Hoischen A**, **Gilissen C**.

Nat Commun. 2023 Oct 27;14(1):6845. doi: 10.1038/s41467-023-42531-9. PMID: 37891200

<https://pubmed.ncbi.nlm.nih.gov/37891200/>

*Higher-order structural organization of the mitochondrial proteome charted by in situ cross-linking mass spectrometry.*

Hevler JF, **Heck AJR**.

Mol Cell Proteomics. 2023 Oct 5:100657. doi: 10.1016/j.mcpro.2023.100657. Online ahead of print. PMID: 37805037

<https://pubmed.ncbi.nlm.nih.gov/37805037/>

*A cyclin-dependent kinase-mediated phosphorylation switch of disordered protein condensation.*

Valverde JM, Dubra G, Phillips M, Haider A, Elena-Real C, Fournet A, Alghoul E, Chahar D, Andrés-Sánchez N, Paloni M, Bernadó P, van Mierlo G, Vermeulen M, van den Toorn H, **Heck AJR**, Constantinou A, Barducci A, Ghosh K, Sibille N, Knipscheer P, Krasinska L, Fisher D, Altelaar M.

Nat Commun. 2023 Oct 9;14(1):6316. doi: 10.1038/s41467-023-42049-0. PMID: 37813838

<https://pubmed.ncbi.nlm.nih.gov/37813838/>

*Epigenetic and Metabolomic Biomarkers for Biological Age: A Comparative Analysis of Mortality and Frailty Risk.*

Kuiper LM, Polinder-Bos HA, Bizzarri D, Vojinovic D, Vallerga CL, Beekman M, Dollé ET, Ghanbari M, Voortman T, Reinders MJT, Verschuren WMM, **Slagboom PE**, van den Akker EB, van Meurs JBJ.

J Gerontol A Biol Sci Med Sci. 2023 Oct 9;78(10):1753-1762. doi: 10.1093/gerona/glad137. PMID: 37303208

<https://pubmed.ncbi.nlm.nih.gov/37303208/>

*Metabolomic biomarkers of habitual B vitamin intakes unveil novel differentially methylated positions in the human epigenome.*

Costeira R, Evangelista L, Wilson R, Yan X, Hellbach F, Sinke L, Christiansen C, Villicaña S, Masachs OM, Tsai PC, Mangino M, Menni C, Berry SE, Beekman M, van Heemst D, **Slagboom PE**, Heijmans BT, Suhre K, Kastenmüller G, Gieger C, Peters A, Small KS, Linseisen J, Waldenberger M, Bell JT.

Clin Epigenetics. 2023 Oct 19;15(1):166. doi: 10.1186/s13148-023-01578-7. PMID: 37858220

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*M-protein diagnostics in multiple myeloma patients using ultra-sensitive targeted mass spectrometry and an off-the-shelf calibrator.*

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## August

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