



Publications X-omics consortium

2022

Publications of the X-omics consortium

December

Identification of de novo variants in nonsyndromic cleft lip with/without cleft palate patients with low polygenic risk scores.

Ishorst N, Henschel L, Thieme F, Drichel D, Sivalingam S, Mehrem SL, Fechtner AC, Fazaal J, Welzenbach J, Heimbach A, Maj C, Borisov O, Hausen J, Raff R, **Hoischen A**, Dixon M, Rada-Iglesias A, Bartusel M, Rojas-Martinez A, Aldharae K, Braumann B, Kruse T, Kirschneck C, Spanier G, Reutter H, Nowak S, Gölz L, Knapp M, Buness A, Krawitz P, Nöthen MM, Nothnagel M, Becker T, Ludwig KU, Mangold E.

Mol Genet Genomic Med. 2022 Dec 5:e2109. doi: 10.1002/mgg3.2109. Online ahead of print. PMID: 36468602

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Effect of exogenous IL-37 on immune cells from a patient carrying a potential IL37 loss-of-function variant: A case study.

Teufel LU, van der Made CI, Klück V, Simons A, **Hoischen A**, Vernimmen V, Joosten LAB, Arts RJW.

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Optical genome mapping and revisiting short-read genome sequencing data reveal previously overlooked structural variants disrupting retinal disease-associated genes.

de Bruijn SE, Rodenburg K, Corominas J, Ben-Yosef T, Reurink J, Kremer H, Whelan L, Plomp AS, Berger W, Farrar GJ, Ferenc Kovács Á, Fajardy I, Hitti-Malin RJ, Weisschuh N, Weener ME, Sharon D, Pennings RJE, Haer-Wigman L, Hoyng CB, **Nelen MR**, **Vissers LELM**, van den Born LI, **Gilissen C**, Cremers FPM, **Hoischen A**, Neveling K, Roosing S.

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Newborn screening for Cerebrotendinous Xanthomatosis: A retrospective biomarker study using both flow-injection and UPLC-MS/MS analysis in 20,000 newborns.

Vaz FM, Jamal Y, Barto R, Gelb MH, DeBarber AE, Wevers RA, **Nelen MR**, Verrips A, Bootsma AH, Bouva MJ, Kleise N, van der Zee W, He T, Salomons GS, Huidekoper HH.

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Detection of spontaneous preterm birth by maternal urinary volatile organic compound analysis: A prospective cohort study.

Ronde E, Frerichs NM, Brantenaar S, El Manouni El Hassani S, Wicaksono AN, Covington JA, De Boer NKH, De Meij TG, **Hankemeier T**, Reiss IKM, Schoenmakers S.

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Cerebrospinal fluid and plasma amine profiles in interictal migraine.

Onderwater GLJ, van Dongen RM, **Harms AC**, Zielman R, van Oosterhout WPJ, van Klinken JB, Goeman JJ, Terwindt GM, van den Maagdenberg AMJM, **Hankemeier T**, Ferrari MD.

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Mukherjee S, Jankevics A, Busch F, Lubeck M, Zou Y, Kruppa G, **Heck AJR**, Scheltema RA, Reiding KR.

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Association of Torquetenovirus viremia with physical frailty and cognitive impairment in three independent European cohorts.

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Post A, Dam WA, Sokooti S, Groothof D, **Gloerich J, van Gool AJ**, Kremer D, Gansevoort RT, van den Born J, Kema IP, Franssen CFM, Dullaart RPF, Bakker SJL.

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