

Discovery of a genetic module essential for assigning left-right asymmetry in humans and ancestral vertebrates.

Szenker-Ravi E, Ott T, Khatoor M, de Bellaing AM, Goh WX, Chong YL, Beckers A, Kannesan D, Louvel G, Anujan P, Ravi V, Bonnard C, Moutton S, Schoen P, Fradin M, Colin E, Megarbane A, Daou L, Chehab G, Di Filippo S, Rooryck C, Deleuze JF, Boland A, Arribard N, Eker R, **Tohari S**, Ng AY, Rio M, Lim CT, Eisenhaber B, Eisenhaber F, Venkatesh B, Amiel J, Crollius HR, Gordon CT, Gossler A, Roy S, Attie-Bitach T, Blum M, Bouvagnet P, Reversade B.

Nat Genet. 2022 Jan;54(1):62-72. doi: 10.1038/s41588-021-00970-4. Epub 2021 Dec

13.PMID: 34903892

Whole-Exome Sequencing to Identify Potential Genetic Risk in Substance Use Disorders: A Pilot Feasibility Study.

AshaRani PV, Amron S, Zainulidin NAB, **Tohari S**, Ng AYJ, Song G, Venkatesh B, Mathuru AS.

J Clin Med. 2021 Jun 25;10(13):2810. doi: 10.3390/jcm10132810.PMID: 34202351

Singapore Undiagnosed Disease Program: Genomic Analysis aids Diagnosis and Clinical Management.

Bhatia NS, Lim JY, Bonnard C, Kuan JL, Brett M, Wei H, Cham B, Chin H, Bosso-Lefevre C, Dharuman P, Escande-Beillard N, Devasia AG, Goh CYJ, Kam S, Liew WK, Liew WK, Lin G, Jain K, Ng AY, Subramanian D, Xie M, Tan YM, Tawari NR, Tiang Z, Ting TW, **Tohari S**, Tong CK, Lezhava A, Ng SB, Law HY, Venkatesh B, Tomar S, Sethi R, Tan G, Shanmugasundaram A, Goh DL, Lai PS, Lai A, Tan ES, Ng I, Reversades B, Tan EC, Foo R, Jamuar SS;

SUREKids Working Group. Arch Dis Child. 2021 Jan;106(1):31-37. doi:

10.1136/archdischild-2020-319180. Epub 2020 Aug 20.PMID: 32819910

Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy

Holger Hengel, Céline Bosso-Lefèvre, George Grady, Emmanuelle Szenker-Ravi, Hankun Li, Sarah Pierce, Élise Lebigot, Thong-Teck Tan, Michelle Y Eio, Gunaseelan Narayanan, Kagistia Hana Utami, Monica Yau, Nader Handal, Werner Deigendesch, Reinhard Keimer, Hiyam M Marzouqa, Meral Gunay-Aygun, Michael J Muriello, Helene Verhelst, Sarah Weckhuysen, Sonal Mahida, Sakkubai Naidu, Terrence G Thomas, Jiin Ying Lim, Ee Shien Tan, Damien Haye, Michèle A P Willemsen, Renske Oegema, Wendy G Mitchell, Tyler Mark Pierson, Marisa V Andrews, Marcia C Willing, Lance H Rodan, Tahsin Stefan Barakat, Marjon van Slegtenhorst, Ralitza H Gavrilova, Diego Martinelli, Tal Gilboa, Abdullah M Tamim, Mais O Hashem, Moeenaldeen D AlSayed, Maha M Abdulrahim, Mohammed Al-Owain, Ali Awaji, Adel A H Mahmoud, Eissa A Fageih, Ali Al Asmari, Sulwan M Algain, Lamyaa A Jad, Hesham M Aldhalaan, Ingo Helbig, David A Koolen, Angelika Riess, Ingeborg Kraegeloh-Mann, Peter Bauer, Suleyman Gulsuner, Hannah Stamberger, Alvin Yu Jin Ng, Sha Tang, Sumanty Tohari, Boris Keren, Laura E Schultz-Rogers, Eric W Klee, Sabina Barresi, Marco Tartaglia, Hagar Mor-Shaked, Sateesh Maddirevula, Amber Begtrup, Aida Telegrafi, Rolph Pfundt, Rebecca Schüle, Brian Ciruna, Carine Bonnard, Mahmoud A Pouladi, James C Stewart, Adam Claridge-Chang, Dirk J Lefeber, Fowzan S Alkuraya, Ajay S Mathuru, Byrappa Venkatesh, Joseph J Barycki, Melanie A Simpson, Saumya S Jamuar, Ludger Schöls, Bruno Reversade

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doi: 10.1038/s41467-020-14360-7. doi: 10.1038/s41467-020-14360-7.

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Hengel H, Bosso-Lefèvre C, Grady G, Szenker-Ravi E, Li H, Pierce S, Lebigot É, Tan TT, Eio MY, Narayanan G, Utami KH, Yau M, Handal N, Deigendesch W, Keimer R, Marzouqa HM, Gunay-Aygun M, Muriello MJ, Verhelst H, Weckhuysen S, Mahida S, Naidu S, Thomas TG, Lim JY, Tan ES, Haye D, Willemssen MAAP, Oegema R, Mitchell WG, Pierson TM, Andrews MV, Willing MC, Rodan LH, Barakat TS, van Slegtenhorst M, Gavrilova RH, Martinelli D, Gilboa T, Tamim AM, Hashem MO, AlSayed MD, Abdulrahim MM, Al-Owain M, Awaji A, Mahmoud AAH, Faqeih EA, Asmari AA, Algain SM, Jad LA, Aldhalaan HM, Helbig I, Koolen DA, Riess A, Kraegeloh-Mann I, Bauer P, Gulsuner S, Stamberger H, Ng AYJ, Tang S, **Tohari S**, Keren B, Schultz-Rogers LE, Klee EW, Barresi S, Tartaglia M, Mor-Shaked H, Maddirevula S, Begtrup A, Telegrafi A, Pfundt R, Schüle R, Ciruna B, Bonnard C, Pouladi MA, Stewart JC, Claridge-Chang A, Lefeber DJ, Alkuraya FS, Mathuru AS, Venkatesh B, Barycki JJ, Simpson MA, Jamuar SS, Schöls L, Reversade B.

Nat Commun. 2020 Jan 30;11(1):595. doi: 10.1038/s41467-020-14360-7.PMID: 32001716

Heterozygous missense variant in EIF6 gene: A novel form of Shwachman-Diamond syndrome?

Koh AL, Bonnard C, Lim JY, Liew WK, Thoon KC, Thomas T, Ali NAB, Ng AYJ, Tohari S, Phua KB, Venkatesh B, Reversade B, Jamuar SS.

Am J Med Genet A. 2020 Sep;182(9):2010-2020. doi: 10.1002/ajmg.a.61758. Epub 2020 Jul 13.PMID: 32657013

Bone matrix hypermineralization associated with low bone turnover in a case of Nasu-Hakola disease.

Shboul M, Roschger P, Ganger R, Paschalis L, Rokidi S, Zandieh S, Behunova J, Muschitz C, Fahrleitner-Pammer A, Ng AYJ, **Tohari S**, Venkatesh B, Bonnard C, Reversade B, Klaushofer K, Al Kaissi A.

Bone. 2019 Jun;123:48-55. doi: 10.1016/j.bone.2018.10.008. Epub 2018 Oct 11.PMID: 30316000

Cenani-Lenz syndactyly syndrome - a case report of a family with isolated syndactyly.

Hettiaracchi D, Bonnard C, Jayawardana SMA, Ng AYJ, **Tohari S**, Venkatesh B, Reversade B, Singaraja R, Dissanayake VHW.

BMC Med Genet. 2018 Jul 24;19(1):125. doi: 10.1186/s12881-018-0646-1.PMID: 30041615

Novel mutations in the ciliopathy-associated gene CPLANE1 (C5orf42) cause OFD syndrome type VI rather than Joubert syndrome.

Bonnard C, Shboul M, Tonekaboni SH, Ng AYJ, **Tohari S**, Ghosh K, Lai A, Lim JY, Tan EC, Devisme L, Stichelbout M, Alkindi A, Banu N, Yüksel Z, Ghoumid J, Elkhartoufi N, Boutaud L, Micalizzi A, Brett MS, Venkatesh B, Valente EM, Attié-Bitach T, Reversade B, Kariminejad A.

Eur J Med Genet. 2018 Oct;61(10):585-595. doi: 10.1016/j.ejmg.2018.03.012. Epub 2018 Mar 30.PMID: 29605658

CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays.

Windpassinger C, Piard J, Bonnard C, Alfadhel M, Lim S, Bisteau X, Blouin S, Ali NB, Ng AYJ, Lu H, **Tohari S**, Talib SZA, van Hul N, Caldez MJ, Van Maldergem L, Yigit G, Kayserili H, Youssef SA, Coppola V, de Bruin A, Tessarollo L, Choi H, Rupp V, Roetzer K, Roschger P, Klaushofer K, Altmüller

J, Roy S, Venkatesh B, Ganger R, Grill F, Ben Chehida F, Wollnik B, Altunoglu U, Al Kaissi A, Reversade B, Kaldis P.
Am J Hum Genet. 2017 Sep 7;101(3):391-403. doi: 10.1016/j.ajhg.2017.08.003.PMID: 28886341

Voltage-gated sodium channel gene repertoire of lampreys: gene duplications, tissue-specific expression and discovery of a long-lost gene.
Zakon HH, Li W, Pillai NE, **Tohari S**, Shingate P, Ren J, Venkatesh B.
Proc Biol Sci. 2017 Sep 27;284(1863):20170824. doi: 10.1098/rspb.2017.0824.PMID: 28931746

Lampreys, the jawless vertebrates, contain only two ParaHox gene clusters.
Zhang H, Ravi V, Tay BH, **Tohari S**, Pillai NE, Prasad A, Lin Q, Brenner S, Venkatesh B.
Proc Natl Acad Sci U S A. 2017 Aug 22;114(34):9146-9151. doi: 10.1073/pnas.1704457114. Epub 2017 Aug 7.PMID: 28784804

Loss-of-Function Mutations in LIG4, a Secreted Ligand Involved in Schwann Cell Myelination, Are Responsible for Arthrogryposis Multiplex
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Shifeng Xue, Je'rome Maluenda, Florent Marguet, Mohammad Shboul, Loïc Quevarec, Carine Bonnard, Alvin Yu Jin Ng, **Sumanty Tohari**, Thong Teck Tan, Mung Kei Kong, Kristin G. Monaghan, Megan T. Cho, Carly E. Siskind, Jacinda B. Sampson, Carolina Tesi Rocha, Fawaz Alkazaleh, Marie Gonzales, Luc Rigonnot, Sandra Whalen, Marta Gut, Ivo Gut, Martine Bucourt, Byrappa Venkatesh, Annie Laquerrière, Bruno Reversade, and Judith Melki,
Am J Hum Genet. 2017 Apr 6;100(4):659-665 · Apr 1, 2017

A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome.
Oud MM, Bonnard C, Mans DA, Altunoglu U, **Tohari S**, Ng AYJ, Eskin A, Lee H, Rupar CA, de Wagenaar NP, Wu KM, Lahiry P, Pazour GJ, Nelson SF, Hegele RA, Roepman R, Kayserili H, Venkatesh B, Siu VM, Reversade B, Arts HH.
Cilia. 2016 Apr 11;5:8. doi: 10.1186/s13630-016-0029-1. eCollection 2016.PMID: 27069622

Cyclostomes Lack Clustered Protocadherins.
Ravi V, Yu WP, Pillai NE, Lian MM, Tay BH, **Tohari S**, Brenner S, Venkatesh B.
Mol Biol Evol. 2016 Feb;33(2):311-5. doi: 10.1093/molbev/msv252. Epub 2015 Nov 5.PMID: 26545918

Mutation in TWINKLE in a Large Iranian Family with Progressive External Ophthalmoplegia, Myopathy, Dysphagia and Dysphonia, and Behavior ChangeMutation in TWINKLE in a Large Iranian Family with Progressive External Ophthalmoplegia, Myopathy, Dysphagia and Dysphonia, and Behavior Change
Abbas Tafakhori , Alvin Yu Jin Ng, **Sumanty Tohari** , Byrappa Venkatesh, Hane Lee , Ascia Eskin , Stanley F Nelson , Carine Bonnard , Bruno Reversade , Ariana Kariminejad
Arch Iran Med. 2016 Feb;19(2):87-91 · Feb 1, 2016

Elephant shark genome provides unique insights into gnathostome evolution.
Venkatesh B, Lee AP, Ravi V, Maurya AK, Lian MM, Swann JB, Ohta Y, Flajnik MF, Sutoh Y, Kasahara M, Hoon S, Gangu V, Roy SW, Irimia M, Korzh V, Kondrychyn I, Lim ZW, Tay BH, **Tohari S**, Kong KW, Ho S, Lorente-Galdos B, Quilez J, Marques-Bonet T, Raney BJ, Ingham PW, Tay A, Hillier LW, Minx P, Boehm T, Wilson RK, Brenner S, Warren WC.

Nature. 2014 Jan 9;505(7482):174-9. doi: 10.1038/nature12826.PMID: 24402279

Evidence for at least six Hox clusters in the Japanese lamprey (*Lethenteron japonicum*).

Mehta TK, Ravi V, Yamasaki S, Lee AP, Lian MM, Tay BH, **Tohari S**, Yanai S, Tay A, Brenner S, Venkatesh B. *Proc Natl Acad Sci U S A*. 2013 Oct 1;110(40):16044-9. doi: 10.1073/pnas.1315760110. Epub 2013 Sep 16. PMID: 24043829

A trans-species missense SNP in *Amhr2* is associated with sex determination in the tiger pufferfish, *Takifugu rubripes* (fugu).

Kamiya T, Kai W, Tasumi S, Oka A, Matsunaga T, Mizuno N, Fujita M, Suetake H, Suzuki S, Hosoya S, **Tohari S**, Brenner S, Miyadai T, Venkatesh B, Suzuki Y, Kikuchi K. *PLoS Genet*. 2012;8(7):e1002798. doi: 10.1371/journal.pgen.1002798. Epub 2012 Jul 12. PMID: 22807687

Integration of the genetic map and genome assembly of fugu facilitates insights into distinct features of genome evolution in teleosts and mammals.

Kai W, Kikuchi K, **Tohari S**, Chew AK, Tay A, Fujiwara A, Hosoya S, Suetake H, Naruse K, Brenner S, Suzuki Y, Venkatesh B. *Genome Biol Evol*. 2011;3:424-42. doi: 10.1093/gbe/evr041. Epub 2011 Jun 1. PMID: 21551351

Characterization of a hypoxia-response element in the *Epo* locus of the pufferfish, *Takifugu rubripes*.

Kulkarni RP, **Tohari S**, Ho A, Brenner S, Venkatesh B. *Mar Genomics*. 2010 Jun;3(2):63-70. doi: 10.1016/j.margen.2010.05.001. Epub 2010 Jun 17. PMID: 21798198

Erythropoietin gene from a teleost fish, *Fugu rubripes*.

Chou CF, **Tohari S**, Brenner S, Venkatesh B. *Blood*. 2004 Sep 1;104(5):1498-503. doi: 10.1182/blood-2003-10-3404. Epub 2004 May 13. PMID: 15142879

Detailed deletion mapping at chromosome 11q23 in colorectal carcinoma.

Lee AS, Seo YC, Chang A, **Tohari S**, Eu KW, Seow-Choen F, McGee JO. *Br J Cancer*. 2000 Sep;83(6):750-5. doi: 10.1054/bjoc.2000.1366. PMID: 10952779

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Szenker-Ravi E, Ott T, Khatoor M, de Bellaing AM, Goh WX, Chong YL, Beckers A, Kannesan D, Louvel G, Anujan P, Ravi V, Bonnard C, Moutton S, Schoen P, Fradin M, Colin E, Megarbane A, Daou L, Chehab G, Di Filippo S, Rooryck C, Deleuze JF, Boland A, Arribard N, Eker R, **Tohari S**, Ng AY, Rio M, Lim CT, Eisenhaber B, Eisenhaber F, Venkatesh B, Amiel J, Crollius HR, Gordon CT, Gossler A, Roy S, Attie-Bitach T, Blum M, Bouvagnet P, Reversade B. *Nat Genet*. 2022 Jun;54(6):906. doi: 10.1038/s41588-022-01053-8. PMID: 35304595