

Date Prepared: 12/02/2020
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Place of Birth: Turkey

Education:

1993	B.S.	Agricultural Science	Selcuk University
1996	M.S.	Plant Biotechnology (Suer Yuce, Ph.D.)	The Graduate School of Natural and Applied Sciences, Akdeniz University, Turkey
2000	Ph.D.	Plant Biotechnology (Kenan Turgut, Ph.D.)	The Graduate School of Natural and Applied Sciences, Akdeniz University, Turkey
2004	M.S.	Cellular & Molecular Biology	University of New Haven, West Haven, CT

Postdoctoral Training:

2000-08/01	Intern	Transposable elements- (Elenor Wurtzel, Ph.D.)	Department of Biological Sciences, Lehman College, Bronx, NY
2003-2004	Intern	Gene markers in breast cancer Eva Sapi, Ph.D. & Barry M. Kacinski, M.D.	Department of Therapeutic Radiology, Yale University, School of Medicine, New Haven, CT
06/04-6/10	Postdoctoral Fellow	Genetic mechanisms of neuropsychiatric disorders (Matthew State, MD, Ph.D.)	Yale- School of Medicine, Department of Genetics & Child Study Center

Faculty Academic Appointments:

2010-11/14	Associate Research Scientist	Child Study Center	Yale School of Medicine
11/14-9/2020	Associate Research Scientist	Neurosurgery	Yale School of Medicine
9/2020-Present	Adjunct Assistant Prof	Neurosurgery	Yale School of Medicine
08/18-Present	Adjunct Lecturer	Biology	Utica College
09/18-present	Instructor	Masonic Medical Research Institute	Masonic Medical Research Institute, Utica, NY

Committee Service:

2018-present	Faculty IRB document preparation	Masonic Medical Research Institute, Utica, NY
2020-present	Compliance Committee	Masonic Medical Research Institute, Utica, NY

Editorial Activities:

Ad hoc Reviewer

Journal of Biological Psychiatry, USA (2012)
 Journal of Behavior Genetics, USA (2010)
 Journal of Gene Report, (2021)
 Oncology Letters, (2021)

Other Editorial Roles

07/16-Present	Reviewer	Frontiers in Neuroscience, in <u>Child and Adolescent Psychiatry</u> , USA
2018-Present	Reviewer	Frontiers in Neuroscience, in <u>Molecular Psychiatry</u> USA
01/18-Present	Reviewer	International Journal of Innovative Approaches in Science Research, Turkey
12/20-Present	Reviewer	Journal of Innovative Approaches in Medicine, Turkey

Honors and Prizes:

2005	Top ten scientific breakthroughs of the year	Science Magazine	SIITRK1 first reported gene that is associated with Tourette syndrome
2016	Key Accomplishments and breakthroughs: 20 key discoveries, advances and developments in Tourette and Tic Disorders	Tourette Association in America	HDC is the first gene that identified a link between histaminergic neurotransmission and tics in humans

Report of Funded and Unfunded Projects

Funding Information:

Past

2015-2017	Molecular Genetic Pathogenesis of Intracranial Aneurysms NIH/NINDS 4R01NS057756-10 Non-key/investigator The Goal of this study was identification and functional verification of variants associated with Intracranial aneurysm to demonstrate their biological effects.
2015-2017	Disease Gene Discovery in Structural Brain disorders NIH/NINDS 1R01MH103616-02 Non-key/investigator The major goal is to identify the molecular and genetic basis of structural brain malformations

- 2014-2019 Integrating the genomics of Autism Spectrum Disorders (ASD) in consanguineous and "idiopathic" families
NIMH 1R01MH102342-01A1
Key Personal (PI: Murat Gunel)
The goal of the study is to identify novel, rare genetic variants in ASD by employing homozygosity mapping and whole exome sequencing and further evaluate the overlap risk loci for inbred versus outbred ASD populations.
- 2019-2020 Pilot Grant
Co-PI (PI: Maria Kontaridis)
Grand Lodge of Free and Accepted Masons of the State of New York-Autism
The major goal of this project is to determine the whether the mechanisms that cause CHD also lead to autism.

Current

Role of PTPN11 Mutations in Autism and Heart Pathogenesis

AHA Translational grant

The major goal of this project is to understand the role of *PTPN11* gene in both cardiac and neurodevelopmental abnormalities.

Training Grants and Mentored Trainee Grants

- 2012-2014 Identifying the genes responsible for ASD and/or ID using multiplex families
NCATS NIH UL1 TR000142 & KL2 TR000140
Yale Center for Clinical Investigation (YCCI) Junior Faculty Scholar (PI: Robert Sherwin)
The goal of this study is to find susceptibility genes in individuals with autism spectrum disorder (ASD) and intellectual disability (ID) by studying families in which the disorder is transmitted in a Mendelian fashion

Report of Local Teaching and Training

Teaching of Students in Courses:

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|-----------|---|---|
| 1993-2000 | Molecular Biology
2 nd or 3 rd year undergraduate students | The Graduate School of Natural and Applied Sciences, Akdeniz University, Turkey. Teaching Assistant |
| 2018-Fall | Genomic Approaches for genetic diseases
4 th year undergrad students | Utica College, Biology Department, Utica, NY. Primary teacher |
| 2019-Fall | Bio420- iPSC and CRISPR Technologies
4 th year undergrad students | Utica College, Biology Department, Utica, NY. Primary teacher |

Laboratory and Other Research Supervisory and Training Responsibilities:

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| 2005-2018 | Teaching, Mentoring undergraduate, graduate, and internship students and residents at laboratory environment. | Yale University, Child Study Center & Neurosurgery |
| 2018-Present | Supervision of undergraduate, graduate, research assistants at the laboratory environment | Masonic Medical Research Institute |

Formally Mentored Yale Undergraduate Students:

- 2008-2009 Nicole Oakman, (Undergraduate)/ Pediatrics Resident, Baylor College of Medicine

Nicole was involved in a project called “Assessing the role of SEMA6D gene in language development” under my supervision.

Accomplishment: Ercan-Sencicek AG, Davis Wright NR, Sanders SJ, **Oakman N**, Valdes L, Bakkaloglu B, Doyle N, Yrigollen CM, Morgan TM, Grigorenko EL. (2012). A balanced t(10;15) translocation in a male patient with developmental language disorder. *Eur J Med Genet.* Feb;55(2):128-31.

2009-2011 Lianna Valdes (Undergraduate)/ Ophthalmology Resident, Upstate Medical University
Lianna was involved in a project called “Finding mutations in histaminergic signaling pathway genes in TS patients” and “Assessing the role of SEMA6D gene in language development” under my supervision.

Accomplishment: Ercan-Sencicek AG, Davis Wright NR, Sanders SJ, **Oakman N**, Valdes L, Bakkaloglu B, Doyle N, Yrigollen CM, Morgan TM, Grigorenko EL. (2012). A balanced t(10;15) translocation in a male patient with developmental language disorder. *Eur J Med Genet.* Feb;55(2):128-31.

Other Mentored Trainees and Faculty:

2010 Leire Almandoz Gil (Undergraduate)
Leire completed her summer internship under my supervision by working on the project “Homozygosity Mapping and Whole exome sequencing of a family with Mental Retardation”.

2011 Lorraine Suzuki (Graduate)/ Ingénieur de production at Sogeti, France
Lorraine completed her research part of master thesis under my supervision.
Accomplishment: Expression level of Williams Beuren Syndrome region genes in Patients with autism.

2013 Sinem Sertel (Undergraduate)/ PhD Student, Neuro and Sensory Physiology, Göttingen University Medical Center, Germany
Sinem completed her summer internship under my supervision by working on the project “Generating induced pluripotent stem cells from peripheral blood of patients with autism”
Accomplishment: Dong S, Walker MF, Carriero NJ, DiCola M, Willsey AJ, Ye AY, Waqar Z, Gonzalez LE, Overton JD, Frahm S, Keaney JF 3rd, Teran NA, Dea J, Mandell JD, Hus Bal V, Sullivan CA, DiLullo NM, Khalil RO, Gockley J, Yuksel Z, **Sertel SM**, Ercan-Sencicek AG, Gupta AR, Mane SM, Sheldon M, Brooks AI, Roeder K, Devlin B, State MW, Wei L, Sanders SJ. (2014) De novo insertions and deletions of predominantly paternal origin are associated with autism spectrum disorder. *Cell Rep.* 9;9(1):16- 23. PMID:25284784; PMCID: PMC4194132.

2013 Zafer Yuksel (Resident, Clinical Genetics)/ Clinical Geneticist, MD at Sonic Healthcare, Germany.
Zafer was involved in the project called “Homozygosity Mapping and Whole exome sequencing of families with Autism/ID” under my supervision.
Accomplishment: Ercan-Sencicek AG, Jambi S, Franjic D, Nishimura S, Li M, El-Fishawy P, Morgan TM, Sanders SJ, Bilguvar K, Suri M, Johnson MH, Gupta AR, **Yuksel Z**, Mane S, Grigorenko E, Picciotto M, Alberts AS, Gunel M, Šestan N, State MW. (2015) Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans. *Eur J Hum Genet.* 23(2):165-72. PMID:24781755; PMCID: PMC4297910.

Dong S, Walker MF, Carriero NJ, DiCola M, Willsey AJ, Ye AY, Waqar Z, Gonzalez LE, Overton JD, Frahm S, Keaney JF 3rd, Teran NA, Dea J, Mandell JD, Hus Bal V, Sullivan CA, DiLullo NM, Khalil RO, Gockley J, **Yuksel Z**, Sertel SM, Ercan-Sencicek AG, Gupta AR, Mane SM, Sheldon M, Brooks AI, Roeder K, Devlin B, State MW, Wei L, Sanders SJ. (2014) De novo insertions and deletions of predominantly paternal origin are associated with autism spectrum disorder. *Cell Rep.* 9;9(1):16- 23. PMID:25284784; PMCID: PMC4194132.

- 2015 Bilge Buyukdemirtas (Undergraduate)/ Master student in Koln University, Applied Sciences
Bilge completed her summer internship under my supervision by working on the project “Characterization of brain vasculature in zebrafish”
- 2015 Burcu Gulez, (Postgraduate Associate)/ PhD student in Columbia University Biological sciences
Burcu was involved in the project called “Whole exome sequencing of patients with structural brain disorders” under my supervision.
Accomplishment: Tüysüz B, Ercan-Sencicek AG, Canpolat N, Koparır A, Yılmaz S, Kılıçaslan I, **Güleç B**, Bilguvar K, Günel M. Renal involvement in patients with mucopolidosis IIIalpha/beta: Causal relation or co-occurrence? Am J Med Genet A. 2016 May;170A(5):1187-95. Doi: 10.1002/ajmg.a.37543. Epub 2016 Jan 8. PubMed PMID: 26749367.
- 2016-2017 Yanki Yarman (Postgraduate Associate)/ Medical School Candidate
Yanki was involved in the project called “Whole exome sequencing of patients with structural brain disorders” under my supervision.
Accomplishment: Yarman, Y, Goc, N, Toy F., Meral C, Ercan-Sencicek AG, Gunel M, Novel compound heterozygous mutations in *GPT2* linked to microcephaly, and intellectual developmental disability with or without spastic paraplegia. American Journal of Medical Genetics, 176:2, (2017).

Poster presentation: Exome sequencing of a Turkish cohort with malformations of brain development. **Y Yarman**, C Zhang, AG Ercan-Sencicek, W Koomson, T Barak, Z Erson-Omay, A Louvi, Y Katsuhito, F Toy, N Goc, B Tuysuz, O Caglayan, H Kaymakçalan, M. Gunel, K. Bilguvar. Yale Annual Genetics Department Retreat, 2017, Hancock, MA.

Novel compound heterozygous variants in *GPT2* in a family with microcephaly and intellectual disability. H. Kaymakçalan Çelebiler, A. Ercan-Sencicek, C. Meral, N. Göç, F. Toy, **Y. Yarman**, M. Gunel. ESHG, Copenhagen, May 28, 2017.
- 2016-2018 Nukte Goc (Postgraduate Associate)/ MPH student in the Department of Biostatistics, Yale School of Medicine.
Nukte was involved in the project called “Whole exome sequencing of patients with structural brain disorders” under my supervision.
Accomplishment: Yarman, Y, **Goc, N**, Toy F, Meral C, Ercan-Sencicek AG, Gunel M, Novel compound heterozygous mutations in *GPT2* linked to microcephaly, and intellectual developmental disability with or without spastic paraplegia. American Journal of Medical Genetics, 176:2, (2017).

Poster presentation: Exome sequencing of a Turkish cohort with malformations of brain development. Y Yarman, C Zhang, AG Ercan-Sencicek, W Koomson, T Barak, Z Erson-Omay, A Louvi, Y Katsuhito, F Toy, **N Goc**, B Tuysuz, O Caglayan, H Kaymakçalan, M. Gunel, K. Bilguvar. Yale Annual Genetics Department Retreat, 2017, Hancock, MA.

Novel compound heterozygous variants in *GPT2* in a family with microcephaly and intellectual disability. H. Kaymakçalan Çelebiler, A. Ercan-Sencicek, C. Meral, **N. Göç**, F. Toy, Y. Yarman, M. Gunel. ESHG, Copenhagen, May 28, 2017.

Report of Regional, National and International Invited Teaching and Presentations

International

- 2006 Detection of Microamplification in a Patient, with a PDD-NOS Disorder using Array-Based Comparative Hybridization. World Psychiatric Assoc. Int congress, Turkey.
- 2012 Overview of latest genetic findings on childhood neuropsychiatric disorders Bahcesehir University, Genetic and Bioinformatics, Turkey.
- 2012 Overview of latest genetic findings on autism. Necmettin Erbakan University, Meram Medical School, Turkey.
- 2017 Speeding up Genetic Research to understand Autism 27th Turkish Child and Adolescent psychiatry congress, Izmir Turkey.
- 2019 Autism Spectrum Disorder (ASD) Naval Nuclear Laboratory, Department of Energy, Schenectady, New York
- 2019 Genetics of Autism Spectrum Disorder (ASD) St. John's Day in the Masonic Community, Utica, New York
- 2020 Autism Spectrum Disorder, Autism Gala to benefit Autism Research, Grand Lodge of the State of New York Free & Accepted Masons

Report of Scholarship

Research Investigations

1. Alvero AB, Burtneess BA, **Ercan AG**, Sapi E. Improved method for the detection of cytokeratin 19-positive cells in the peripheral blood of breast cancer patients, Laboratory investigation. 2004 May;84(5):658-61. PubMed PMID: 15105816.
2. Drazinic CM, **Ercan-Sencicek AG**, Gault LM, Hisama FM, Qumsiyeh MB, Nowak NJ, Cubells JF, State MW. Rapid array-based genomic characterization of a subtle structural abnormality: a patient with psychosis and der(18)t(5;18)(p14.1;p11.23). Am J Med Genet A. 2005 Apr 30;134(3):282-9. PubMed PMID:15754353.
3. Abelson JF, Kwan KY, O'Roak BJ, Baek DY, Stillman AA, Morgan TM, Mathews CA, Pauls DL, Rasin MR, Gunel M, Davis NR, **Ercan-Sencicek AG**, Guez DH, Spertus JA, Leckman JF, Dure LS 4th, Kurlan R, Singer HS, Gilbert DL, Farhi A, Louvi A, Lifton RP, Sestan N, State MW. Sequence variants in SLITRK1 are associated with Tourette's syndrome. Science. 2005 Oct 14;310(5746):317-20. PubMed PMID:16224024.
4. Bilguvar K, Bydon M, Bayrakli F, **Ercan-Sencicek AG**, Bayri Y, Mason C, DiLuna ML, Seashore M, Bronen R, Lifton RP, State M, Gunel M. A novel syndrome of cerebral cavernous malformation and Greig cephalopolysyndactyly. Laboratory investigation. J Neurosurg. 2007 Dec;107(6 Suppl):495-9. PubMed PMID: 18154020.
5. Bakkaloglu B, O'Roak BJ, Louvi A, Gupta AR, Abelson JF, Morgan TM, Chawarska K, Klin A, **Ercan-Sencicek AG**, Stillman AA, Tanriover G, Abrahams BS, Duvall JA, Robbins EM, Geschwind DH, Biederer T, Gunel M, Lifton RP, State MW. Molecular cytogenetic analysis and resequencing of contactin associated protein-like 2 in autism spectrum disorders. Am J Hum Genet. 2008 Jan;82(1):165-73. doi:10.1016/j.ajhg.2007.09.017. PubMed PMID: 18179895; PubMed Central PMCID:PMC2253974.
6. Bayrakli F, Guney I, Bayri Y, **Ercan-Sencicek AG**, Ceyhan D, Cankaya T, Mason C, Bilguvar K, Bayrakli S, Mane SM, State MW, Gunel M. A novel heterozygous deletion within the 3' region of the PAX6 gene causing isolated aniridia in a large family group. J Clin Neurosci. 2009 Dec;16(12):1610-4. doi:10.1016/j.jocn.2009.03.022. Epub 2009 Sep 29. PubMed PMID: 19793656.
7. **Ercan-Sencicek AG**, Stillman AA, Ghosh AK, Bilguvar K, O'Roak BJ, Mason CE, Abbott T, Gupta A, King RA, Pauls DL, Tischfield JA, Heiman GA, Singer HS, Gilbert DL, Hoekstra PJ, Morgan TM, Loring E, Yasuno K, Fernandez T, Sanders S, Louvi A, Cho JH, Mane S, Colangelo

- CM, Biederer T, Lifton RP, Gunel M, State MW. L-histidine decarboxylase and Tourette's syndrome. *N Engl J Med*. 2010 May 20;362(20):1901-8. doi: 10.1056/NEJMoa0907006. Epub 2010 May 5. PubMed PMID:20445167; PubMed Central PMCID: PMC2894694.
8. Krusong K, **Ercan-Sencicek AG**, Xu M, Ohtsu H, Anderson GM, State MW, Pittenger C. High levels of histidine decarboxylase in the striatum of mice and rats. *Neurosci Lett*. 2011 May 16;495(2):110-4. doi: 10.1016/j.neulet.2011.03.050. Epub 2011 Apr 1. PubMed PMID: 21440039; PubMed Central PMCID: PMC3081964.
 9. Sanders SJ, **Ercan-Sencicek AG**, Hus V, Luo R, Murtha MT, Moreno-De-Luca D, Chu SH, Moreau MP, Gupta AR, Thomson SA, Mason CE, Bilguvar K, Celestino-Soper PB, Choi M, Crawford EL, Davis L, Wright NR, Dhodapkar RM, DiCola M, DiLullo NM, Fernandez TV, Fielding-Singh V, Fishman DO, Frahm S, Garagaloyan R, Goh GS, Kammela S, Klei L, Lowe JK, Lund SC, McGrew AD, Meyer KA, Moffat WJ, Murdoch JD, O'Roak BJ, Ober GT, Pottenger RS, Raubeson MJ, Song Y, Wang Q, Yaspan BL, Yu TW, Yurkiewicz IR, Beaudet AL, Cantor RM, Curland M, Grice DE, Günel M, Lifton RP, Mane SM, Martin DM, Shaw CA, Sheldon M, Tischfield JA, Walsh CA, Morrow EM, Ledbetter DH, Fombonne E, Lord C, Martin CL, Brooks AI, Sutcliffe JS, Cook EH Jr, Geschwind D, Roeder K, Devlin B, State MW. Multiple recurrent de novo CNVs, including duplications of the 7q11.23 Williams syndrome region, are strongly associated with autism. *Neuron*. 2011 Jun 9;70(5):863-85. doi:10.1016/j.neuron.2011.05.002. PubMed PMID: 21658581; PubMed Central PMCID:PMC3939065.
 10. Celestino-Soper PB, Shaw CA, Sanders SJ, Li J, Murtha MT, **Ercan-Sencicek AG**, Davis L, Thomson S, Gambin T, Chinault AC, Ou Z, German JR, Milosavljevic A, Sutcliffe JS, Cook EH Jr, Stankiewicz P, State MW, Beaudet AL. Use of array CGH to detect exonic copy number variants throughout the genome in autism families detects a novel deletion in TMLHE. *Hum Mol Genet*. 2011 Nov 15;20(22):4360-70. doi: 10.1093/hmg/ddr363. Epub 2011 Aug 24. PubMed PMID: 21865298; PubMed Central PMCID: PMC3196886.
 11. Fernandez TV, Sanders SJ, Yurkiewicz IR, **Ercan-Sencicek AG**, Kim YS, Fishman DO, Raubeson MJ, Song Y, Yasuno K, Ho WS, Bilguvar K, Glessner J, Chu SH, Leckman JF, King RA, Gilbert DL, Heiman GA, Tischfield JA, Hoekstra PJ, Devlin B, Hakonarson H, Mane SM, Günel M, State MW. Rare copy number variants in Tourette syndrome disrupt genes in histaminergic pathways and overlap with autism. *Biol Psychiatry*. 2012 Mar 1;71(5):392-402. doi: 10.1016/j.biopsych.2011.09.034. Epub 2011 Dec 14. PubMed PMID: 22169095; PubMed Central PMCID: PMC3282144.
 12. Sanders SJ, Murtha MT, Gupta AR, Murdoch JD, Raubeson MJ, Willsey AJ, **Ercan-Sencicek AG**, DiLullo NM, Parikshak NN, Stein JL, Walker MF, Ober GT, Teran NA, Song Y, El-Fishawy P, Murtha RC, Choi M, Overton JD, Bjornson RD, Carriero NJ, Meyer KA, Bilguvar K, Mane SM, Sestan N, Lifton RP, Günel M, Roeder K, Geschwind DH, Devlin B, State MW. De novo mutations revealed by whole-exome sequencing are strongly associated with autism. *Nature*. 2012 Apr 4;485(7397):237-41. doi: 10.1038/nature10945. PubMed PMID: 22495306; PubMed Central PMCID: PMC3667984.
 13. Novarino G, El-Fishawy P, Kayserili H, Meguid NA, Scott EM, Schroth J, Silhavy JL, Kara M, Khalil RO, Ben-Omran T, **Ercan-Sencicek AG**, Hashish AF, Sanders SJ, Gupta AR, Hashem HS, Matern D, Gabriel S, Sweetman L, Rahimi Y, Harris RA, State MW, Gleeson JG. Mutations in BCKD-kinase lead to a potentially treatable form of autism with epilepsy. *Science*. 2012 Oct 19;338(6105):394-7. doi: 10.1126/science.1224631. Epub 2012 Sep 6. PubMed PMID: 22956686; PubMed Central PMCID: PMC3704165.
 14. Willsey AJ, Sanders SJ, Li M, Dong S, Tebbenkamp AT, Muhle RA, Reilly SK, Lin L, Fertuzinhos S, Miller JA, Murtha MT, Bichsel C, Niu W, Cotney J, **Ercan-Sencicek AG**, Gockley J, Gupta AR, Han W, He X, Hoffman EJ, Klei L, Lei J, Liu W, Liu L, Lu C, Xu X, Zhu Y, Mane SM, Lein ES, Wei L, Noonan JP, Roeder K, Devlin B, Sestan N, State MW. Coexpression networks implicate human midfetal deep cortical projection neurons in the pathogenesis of autism. *Cell*.

2013 Nov 21;155(5):997-1007. doi: 10.1016/j.cell.2013.10.020. PubMed PMID: 24267886; PubMed Central PMCID: PMC3995413.

15. Baldan LC, Williams KA, Gallezot JD, Pogorelov V, Rapanelli M, Crowley M, Anderson GM, Loring E, Gorczyca R, Billingslea E, Wasyluk S, Panza KE, **Ercan-Sencicek AG**, Krusong K, Leventhal BL, Ohtsu H, Bloch MH, Hughes ZA, Krystal JH, Mayes L, de Araujo I, Ding YS, State MW, Pittenger C. Histidine decarboxylase deficiency causes tourette syndrome: parallel findings in humans and mice. *Neuron*. 2014 Jan 8;81(1):77-90. doi: 10.1016/j.neuron.2013.10.052. Erratum in: *Neuron*. 2014 Jun 4;82(5):1186-7. PubMed PMID: 24411733; PubMed Central PMCID: PMC3894588.
16. **Ercan-Sencicek AG**, Jambi S, Franjic D, Nishimura S, Li M, El-Fishawy P, Morgan TM, Sanders SJ, Bilguvar K, Suri M, Johnson MH, Gupta AR, Yuksel Z, Mane S, Grigorenko E, Picciotto M, Alberts AS, Gunel M, Sestan N, State MW. Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans. *Eur J Hum Genet*. 2015 Feb;23(2):165-72. doi: 10.1038/ejhg.2014.82. Epub 2014 Apr 30. PubMed PMID: 24781755; PubMed Central PMCID: PMC4297910.
17. Gupta AR, Pirruccello M, Cheng F, Kang HJ, Fernandez TV, Baskin JM, Choi M, Liu L, **Ercan-Sencicek AG**, Murdoch JD, Klei L, Neale BM, Franjic D, Daly MJ, Lifton RP, De Camilli P, Zhao H, Sestan N, State MW. Rare deleterious mutations of the gene EFR3A in autism spectrum disorders. *Mol Autism*. 2014 Apr 29;5:31. doi: 10.1186/2040-2392-5-31. eCollection 2014. PubMed PMID: 24860643; PubMed Central PMCID: PMC4032628.
18. Dong S, Walker MF, Carriero NJ, DiCola M, Willsey AJ, Ye AY, Waqar Z, Gonzalez LE, Overton JD, Frahm S, Keaney JF 3rd, Teran NA, Dea J, Mandell JD, Hus Bal V, Sullivan CA, DiLullo NM, Khalil RO, Gockley J, Yuksel Z, Sertel SM, **Ercan-Sencicek AG**, Gupta AR, Mane SM, Sheldon M, Brooks AI, Roeder K, Devlin B, State MW, Wei L, Sanders SJ. De novo insertions and deletions of predominantly paternal origin are associated with autism spectrum disorder. *Cell Rep*. 2014 Oct 9;9(1):16-23. doi: 10.1016/j.celrep.2014.08.068. Epub 2014 Oct 2. PubMed PMID: 25284784; PubMed Central PMCID: PMC4194132.
19. Sanders SJ, He X, Willsey AJ, **Ercan-Sencicek AG**, Samocha KE, Cicek AE, Murtha MT, Bal VH, Bishop SL, Dong S, Goldberg AP, Jinlu C, Keaney JF 3rd, Klei L, Mandell JD, Moreno-DeLuca D, Poultney CS, Robinson EB, Smith L, Solli-Nowlan T, Su MY, Teran NA, Walker MF, Werling DM, Beaudet AL, Cantor RM, Fombonne E, Geschwind DH, Grice DE, Lord C, Lowe JK, Mane SM, Martin DM, Morrow EM, Talkowski ME, Sutcliffe JS, Walsh CA, Yu TW; Autism Sequencing Consortium, Ledbetter DH, Martin CL, Cook EH, Buxbaum JD, Daly MJ, Devlin B, Roeder K, State MW. Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. *Neuron*. 2015 Sep 23;87(6):1215-1233. doi:10.1016/j.neuron.2015.09.016. PubMed PMID: 26402605; PubMed Central PMCID: PMC4624267.
20. Bai H, Harmancı AS, Erson-Omay EZ, Li J, Coşkun S, Simon M, Kirschek B, Özdoğan K, Omay SB, Sorensen EA, Turcan Ş, Bakırcıoğlu M, Carrión-Grant G, Murray PB, Clark VE, **Ercan-Sencicek AG**, Knight J, Sencar L, Altınok S, Kaulen LD, Gülez B, Timmer M, Schramm J, Mishra-Gorur K, Henegariu O, Moliterno J, Louvi A, Chan TA, Tannheimer SL, Pamir MN, Vortmeyer AO, Bilguvar K, Yasuno K, Günel M. Integrated genomic characterization of IDH1-mutant glioma malignant progression. *Nat Genet*. 2016 Jan;48(1):59-66. doi: 10.1038/ng.3457. Epub 2015 Nov 30. PubMed PMID: 26618343; PubMed Central PMCID: PMC4829945.
21. Tüysüz B, **Ercan-Sencicek AG**, Canpolat N, Koparır A, Yılmaz S, Kılıçaslan I, Gülez B, Bilguvar K, Günel M. Renal involvement in patients with mucopolidosis IIIalpha/beta: Causal relation or co-occurrence? *Am J Med Genet A*. 2016 May;170A(5):1187-95. doi: 10.1002/ajmg.a.37543. Epub 2016 Jan 8. PubMed PMID: 26749367.
22. Anazi S, Maddirevula S, Faqeih E, Alsedairy H, Alzahrani F, Shamseldin HE, Patel N, Hashem M, Ibrahim N, Abdulwahab F, Ewida N, Alsaif HS, Al Sharif H, Alamoudi W, Kentab A, Bashiri FA, Alnaser M, AlWadei AH, Alfadhel M, Eyaid W, Hashem A, Al Asmari A, Saleh MM, AlSaman A, Alhasan KA, Alsughayir M, Al Shammari M, Mahmoud A, Al-Hassnan ZN, Al-Husain M,

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