

Barış Salman

Curriculum Vitae

technical skills

Wet Lab

• Bioinformatics

DNA/RNA/Protein Isolation,

quantative/qualitative PCRs, Blotting, Cytogenetics and Karyotyping Sequencing

WGS, WES, Panels, RNAseq, scRNAseq,

Sanger

Functional Studies Cell Culture, in vivo

models, Microscopy

Micro-arrays Genotyping,

Expression

Genetic Engineering F

Recombinant DNA technologies

Computers

Writing

• Data Analyses

Development Python, R, Unix Shell **Analysis** Python Scientific

Toolkit(numpy, pandas,

Workflow Linux, Doom Emacs, Scip Git, Jupyter lang

scipy etc.), R language, Shell Scripting,

Bioinformatic tools

Machine Learning, PCA, Clustering

Web Development Django, Javascript Statistics and Probability

Orgmode, Zotero,

Bibtex, LATEX

Hypothesis testing, Bayesian Theorem, Data distributions

Visualization Matplotlib, Plotly/Dash,

EX

education

Doctorate, 2021-2024 Genetics, Institute of Health Sciences, İstanbul University, Turkey **GPA 3.5**

Thesis Work in progress

Adviser Prof. Dr. Sibel Uğur İşeri

Notable Courses Advanced Molecular Genetics, Mendelian and non Mendelian

Diseases, Chromosomal Diseases, Systems Biology

Master, 2016-2019 Genetics, Institute of Health Sciences, İstanbul University, Turkey **GPA 2.14**

Thesis Study of Split Hand/Foot Malformation with Genomic Techniques

and Bioinformatic Approaches

Adviser Assoc Prof. Dr. Sibel Uğur İşeri

Notable Courses Bioinformatics, Genetic Counseling, Medical Genetics, Hospital

Rotation for Genetic Counseling

Bachelor, 2011-2016 Molecular Biology and Genetics, İstanbul University, Turkey

Thesis Metabolic Regulation in Prokaryotes with Small Non-Coding

RNAs

Adviser Asst. Prof. Dr. Semian Karaer Uzuner

Notable Courses Methods Used in Molecular Biology(lab), Molecular Genetics,

Genetic Engineering

iobs

Refgen Biotechnology Development of platform for Company

bioinformatics

Date Mar 2021-Ongoing Pipeline research and development

Position Genetics and Bioinformatics Specialist

Company Gen-Era Diagnostics Development of pipelines for NGS Oct. 2018-Mar 2021 Date

Variant pathogenity classification

automatization

Position Genetics and Bioinformatics Specialist

Company Aktif Gen Development of bioinformatic pipelines

Date Jan. 2018-Oct 2018 **Position** R&D Personnel

GPA 2.44

• Internships & Rotations

• Certificates

Genetic Counseling	Fall 2018	Use and care of	Mar 21 - Apr 08
rotation	IU Cerrahpaşa, Department of Pediatric Genetics and	laboratory animals	IU HADYEK 46. Laboratory Animal Use Training
	Teratology	Liquid handling	2-5 November 2021
Genetic Counseling	Spring 2018	robot	Hamilton Venus Online
rotation	IU Cerrahpaşa, Department of Medical Genetics		Advanced Software Training
O-II O-It			9-12 August 2021
Cell Culture	August 2016		Hamilton Venus Online
internship	IU Cerrahpasa Department of Medical Biology		Basic Software Training
Genomics	June-July 2015		
internship	IU A.S. Inst. of Expr. Medicine Department of Genetics		
Cytogenetics	June-July 2014		
internship	IU Department of Medical Genetics		
Molecular Dynamics	2012-2013		
internship	IU Faculty of Science Quantum Technologies Laboratory		

publications

- Mercan S, Akcakaya NH, Salman B, Yapici Z, Ozbek U, Ugur Iseri SA. Clinical and genetic analyses in syndromic intellectual disability with primary microcephaly reveal biallelic and de novo variants in patients with parental consanguinity. Genes Genom. Published online November 12, 2022. doi:10.1007/s13258-022-01344-8
- Oguz-Akarsu E, Salman B, Ugur-Iseri S, Baykan B. An Extraordinary EEG Phenomenon Misdiagnosed as Nonconvulsive Status Epilepticus: Frequent Subclinical Periodic Discharges Terminated by Sudden Auditory Stimuli. Clin EEG Neurosci. Published online October 5, 2022:15500594221129964. doi:10.1177/15500594221129965
- Salman B., Yucesan E., Samanci B., Bilgic B., Hanagasi H., Gurvit H., Ozbek U., Ugur Iseri S. Combined Analysis Of Linkage And Whole Exome Sequencing Reveals CIC As A Candidate Gene For Isolated Dystonia. Journal of Istanbul Faculty of Medicine. 2021;84(4):457-463. doi:10.26650/IUITFD.2021.913346
- 4. Haryanyan G, Ozdemir O, Tutkavul K, Salman B, et al. The rare rs769301934 variant in NHLRC1

- is a common cause of Lafora disease in Turkey. J Hum Genet. 2021;66(12):1145-1151. doi:10.1038/s10038-021-00944-8
- Akçakaya NH, Salman B, Ugur İşeri SA., et al. A Novel and Mosaic WDR45 Nonsense Variant Causes Beta-Propeller Protein-Associated Neurodegeneration Identified Through Whole Exome Sequencing and X chromosome Heterozygosity Analysis. Neuromolecular Med. 2019;21(1):54-59. doi:10.1007/s12017-018-08522-6
- 6. Bekdik Şirinocak P, Salman B, Kesim FY, Bebek N, Baykan B, Ugur İşeri SA. Susceptibility to Juvenile Myoclonic Epilepsy Associated with the EFHC1 Gene: First Case Report in Turkey. tnd. 2019;25(4):233-236. doi:10.4274/tnd.galenos.2019.61214

• in pipeline

- Exome sequencing reveals a treatable neurometabolic origin in two siblings with an undiagnosed form of neurodevelopmental disorder: From exon to neuron (in review)
- ELP2 Mutation Leads Developmental and Epileptic Encephalopathy: Case Report Consanguineous Family from Turkey and Review of Literature (in review)
- Variant Version Control: A Git Framework For Keeping Track Of Variant Annotation Changes (in prep.)

Collaborative

- 1. Niestroj LM, Perez-Palma E, Howrigan DP, et al. Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17,458 subjects. Brain. 2020;143(7):2106-2118. doi:10.1093/brain/awaa171
- 2. Stevelink R, Luykx JJ, Lin BD, et al. Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia. 2021;62(7):1518-1527. doi:10.1111/epi.16922
- 3. Leu C, Stevelink R, Smith AW, et al. Polygenic burden in focal and generalized epilepsies. Brain. 2019;142(11):3473-3481. doi:10.1093/brain/awz292
- 4. Koko M, Krause R, Sander T, et al. Distinct gene-set burden patterns underlie common generalized and focal epilepsies. EBioMedicine. 2021;72:103588. doi:10.1016/j.ebiom.2021.103588
- Epi25 Collaborative. Electronic address: s.berkovic@unimelb.edu.au, Epi25 Collaborative. Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. Am J Hum Genet. 2019;105(2):267-282. doi:10.1016/j.ajhg.2019.05.020
- 6. Epi25 Collaborative. Electronic address: jm4279@cumc.columbia.edu, Epi25 Collaborative. Subgenic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. Am J Hum Genet. 2021;108(10):2024. doi:10.1016/j.ajhg.2021.08.008

Posters

- Susgun S., Kesim Y., Salman B., Yucesan E., Khalilov D., Sirin G., Baykan B, Bebek N., Iseri Ugur S., Two candidates bi-allelic variant to neurodevelopmental disorder in a consanguineous family from Turkey, ESHG 2022
- 2. Acar A., Say M., Salman B., Dulger M.V., Comparison of Established Microsatellite Instability Detection Tools in Next Generation Sequencing, ESHG 2021

3. Ugur Iseri S., Akçakaya N. H., Salman B., et al., Exome Sequencing Identifies a FBXO38 Variant Inherited from a Mosaic Mother to cause Distal Hereditary Neuronopathy Type IID with Distinct Features, ASHG 2017

Congresses

1. Investigation of Gene Variants Associated with Syndromic Intellectual Disability, 7. International Congress of Cerebral Palsy and Developmental Disorders, February 23-25 2018, Istanbul, Turkey

projects

- 1. (in review) TUBITAK 1001 Project, **Advanced Molecular Autopsy Protocol for Sudden Deaths** under 40 Years Research Fellow
- 2. 2021-2022 TUBITAK 1501 Project, 3210420, Modeling and Automatic Analysis of Genotype-Phenotype Related Copy Number Variations Researcher
- 3. 2021-2022 Research Fund of Istanbul University, ONAP-37862, **Transcriptomics Approaches To Biomarker Potential Of B Cell Expression Analyses In Anti-Neuronal Antibody Related Autoimmune Epilepsy** Researcher
- 4. 2018-2019 Research Fund of Istanbul University, TYL-2018-30315, Study of Split Hand/Foot Malformation with Genomic Techniques and Bioinformatics Approaches Researcher
- 5. 2018–2018 TUBITAK 1512 Project, 3210420 **Development of Cloud Based Software for Next Generation Amplicon Sequencing Technologies**, Jan 2018- Oct 2018 Research Fellow
- 6. 2017-2018 Research Fund of Istanbul University, TDP-2017-25510, **Genetic Analyses of Progressive Myoclonic Epilepsy** Researcher
- 7. 2016–2018 TUBITAK 1001 Project, **Investigation of Epileptogenesis After Febrile Seizures**Research Fellow

software

VVC Variant Version Control (in development)

Tool for tracking changes in variant annotation using git.

Dove Downstream VCF Evaluation

Tool for annotating VCF files, multi genome analysis and filtering

variants.

Pigeon Pipelining Genomic Operations

Tool for pipelining bioinformatics tools written considering NGS

applications.

Picus Pointed Interpretation of Clinical Variant Significance

Tool for classifying sequence variants according to ACMG/AMP

criteria.

awards

III. Rare Neurological Diseases Symposium and Neurogenetics Course, **Second place project award**, Istanbul, Turkey, 2017

congresses&courses

- 2022 Turkish Society of Medical Genetics, Nov 9-13, Genome-Level Evaluation of Copy Number Changes, One of the Important Risk Factors for Genetic Epilepsy (Oral Presentation)
- 2. 2020 ESHG 2020, June 6-9, Virtual Conference, **GenerAVI: Variant Interpreter and Genetic Analysis Summary Generator** (Poster Presentation)
- 3. 2019 7th International Congress of the Molecular Biology Association of Turkey, Sep 27-29, Istanbul, Turkey, **PICUS: Pointed Interpretation of Clinical Variant Significance** (Poster Presentation)
- 4. 2019 Bioinformatics Days III, Gazi University Faculty of Medicine, May 25, Ankara, Turkey, **NGS Pipelines with Python** (Oral Presentation)
- 5. 2019 Erciyes University Faculty of Medicine Genetics Days, February 21-23, Kayseri, Turkey, **Making Sense of Human Genome with Databases and Bioinformatic Tools** (Oral Presentation)
- 6. 2018 IV. Rare Neurological Diseases Symposium and Neurogenetics Course, May 31-July 1, Istanbul, Turkey, **Analysis of Neurogenetics Data Interactive Training and Practice** (Instructor)
- 7. 2018 11. National Epilepsy Congress, May 3–6, 2018, Dalaman, Turkey, **Meta Analysis of SCN1A Gene Variants of 114 Patients from Epi25 Exome Data** (Oral Presentation)
- 8. 2018 7. International Congress of Cerebral Palsy and Developmental Disorders, February 23-25, Istanbul, Turkey, Investigation of Gene Variants Associated with Syndromic Intellectual Disability

- 9. 2017 53. National Neurology Congress, November 24–30, Antalya, Turkey, **Genomic Approaches to Intellectual Disability Case with Epilepsy** (Oral Presentation)
- 10. 2017 III. Rare Neurological Diseases Symposium and Neurogenetics Course, July 1–2, Istanbul, Turkey, **Profiling Anti-Epileptic Drug Resistance an Genetic Diagnosis with Epilepsy Panel** (Poster Presentation)

general skills

Languages

Turkish Mother Tongue

English Advanced Fluent

These are the languages I am still learning

French BeginnerBasic words and phrasesJapanese BeginnerBasic words and phrases

Latin Beginner Basic words and phrases