

Barış Salman

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Curriculum Vitae
E-mail Phone Website
Github Linkedin Biostars Orcid

technical skills

Wet lab and Bioinformatics

DNA/RNA/Protein Isolation, **Sequencing** WGS, WES, Panels, quantative/qualitative RNAseq, scRNAseq,

quantative/qualitative PCRs, Blotting, Cytogenetics and Karyotyping

Functional StudiesCell Culture, *in vivo*models, Microscopy

Micro-arrays
Expression

, 17

Recombinant DNA technologies

Computers and Data Analyses

Genetic Engineering

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

Toolkit(numpy, pandas, scipy etc.), R language,

Sanger

Shell Scripting,
Bioinformatic tools
Machine Learning,
PCA, Clustering

Workflow Linux, Doom Emacs,

Git, Jupyter

Statistics and Probability

Hypothesis testing, Bayesian Theorem,

Data distributions

Writing Orgmode, Zotero,

Bibtex, LATEX

Visualization Ma

Matplotlib, Plotly/Dash,

MTEX

Web Development Django, Javascript

education

Doctorate, 2021-2024 Genetics, Institute of Health Sciences, Istanbul 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, Istanbul 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, Istanbul 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 CounselingFall 2018Use and care ofMar 21 - Apr 08rotationIU Cerrahpaşa,laboratory animalsIU HADYEK 46.Department ofLaboratory Animal Use

Pediatric Genetics and Training

Teratology

Genetic Counseling Spring 2018 robot Counseling Spring 2018 robot Hamilton Venus Online IU Cerrahpasa. Advanced Software

Department of Medical Training
Genetics 9-12 Aug

9-12 August 2021

Cell Culture August 2016 Hamilton Venus Online
Basic Software

internship IU Cerrahpasa Training

Department of Medical

Genomics June-July 2015
internship IU A.S. Institute of

Exprmental Medicine

Department of Genetics

Biology

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
- 2. 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

- 3. 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
- 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

• Epi25 Collaborative

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.

 $\mathbf{X}\Sigma\Delta$ Cross-symbol checker

This tool checks Ensembl and NCBI annotation files for different genome versions and shows which gene symbol is used. This way a more appropriate gene set can be used to avoid the false negatives in

the variant discovery process.

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

- 1. 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)
- 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)
- 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

These are the languages I am still learning

French Beginner

Japanese Beginner

Latin Beginner

Fluent

Basic words and phrases Basic words and phrases

Basic words and phrases