Assistant Professor, Sinem FIRTINA

Education:

2011-2017, Istanbul University, Aziz Sancar Institute for Experimental Research, Genetics Department, ***PhD****,* Identification of Genetic Background in Primary Immunodeficiencies with Next Generation Sequencing

Supervisor: Prof. Dr. Muge Sayitoglu, Second Supervisor: Assist. Prof. Dr. Yuk Yin Ng

2007-2010, Istanbul University, Aziz Sancar Institute for Experimental Research, Immunology Department, ***MsC*****,** Analysis of *PAX5* gene in B-Lineage Acute Lymphoblastic Leukemia

Supervisor: Prof. Dr. Ugur Ozbek

2003-2007, Marmara University, Faculty of Science and Literature, Biology Department, ***Bachelor***

Research Area:

Primary Immunodeficiencies, Next Generation Sequencing, Pediatric Leukemia

Courses:

PSI104- Introduction to General Biology

PRE003- Genetic

Publications:

- Sinem Firtina, Funda Cipe, Yuk Yin Ng, Ayca Kiykim, Ozden Hatirnaz Ng, Tugce Sudutan, Cigdem Aydogmus, Safa Baris, Gulyuz Ozturk, Elif Aydiner, Ahmet Ozen, Muge Sayitoglu, A Novel FOXN1 Variant Is Identified in Two Siblings with Nude Severe Combined Immunodeficiency, J Clin Immunol. 2019 Mar 22. doi: 10.1007/s10875-019-00615-6
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- Ozkaya HM, Comunoglu N, Sayitoglu M, Keskin FE, Firtina S, Khodzhaev K, Apaydin T, Gazioglu N, Tanriover N, Oz B, Kadioglu P, Germline mutations of aryl hydrocarbon

receptor-interacting protein (AIP) gene and somatostatin receptor 1-5 and AIP immunostaining in patients with sporadic acromegaly with poor versus good response to somatostatin analogues,., Pituitary. 2018 Feb 17. doi: 10.1007/s11102-018-0876-4

- Sinem Firtina, Yuk Yin Ng, Ozden Hatirnaz Ng, Serdar Nepesov, Osman Yesilbas, Meltem Kilercik, Nihan Burtecene, Suzan Cinar, Yildiz Camcioglu, Ugur Ozbek, Muge Sayitoglu, A Novel Pathogenic Frameshift Variant of *CD3E* gene in two T- B+ NK+ SCID patients from Turkey, Immunogenetics, 2017, 10.1007/s00251-017-1005-7
- Firtina S, Hatirnaz Ng Ö, Erbilgin Y, Özbek U, Sayitoğlu M..Dysregulation of the *DKK1* gene in pediatric B-cell acute lymphoblastic leukemia., Turk J Med Sci. 2017 Feb 27;47(1):357-363. doi: 10.3906/sag-1507-106
- Ozden HATIRNAZ NG, Sinem FIRTINA, Yucel ERBILGIN, Ugur OZBEK, Muge SAYITOGLU, Aberrant Hypermethylation of *APC* Tumor Supressor Gene in Acute Leukemia Patients, International Journal of Hematology and Oncology, 2017, 27-1, doi: 10.4999/uhod.171663
- Özden Hatırnaz Ng, Sinem Fırtına, İsmail Can, Zeynep Karakaş, Leyla Ağaoğlu, Ömer Doğru, Tiraje Celkan, Arzu Akçay, Yıldız Yıldırmak, Çetin Timur, Uğur Özbek, Müge Sayitoğlu, A possible role for *WNT5A* hypermethylation in Pediatric Acute Lymphoblastic Leukemia, 127-135 | DOI:
- Ng OH, Erbilgin Y, **Firtina S**, Celkan T, Karakas Z, Aydogan G, Turkkan E, Yildirmak Y, Timur C, Zengin E, van Dongen JJ, Staal FJ, Ozbek U, Sayitoglu M., Deregulated WNT signaling in childhood T-cell acute lymphoblastic leukemia. Blood Cancer J. 2014 Mar 14;4:e192. doi: 10.1038/bcj.2014.12.
- Niyazoglu M, Sayitoglu M, **Firtina S**, Hatipoglu E, Gazioglu N, Kadioglu P.,Familial acromegaly due to aryl hydrocarbon receptor-interacting protein (AIP) gene mutation in a Turkish cohort., Pituitary. 2013 Jun 7.
- **Firtina S,** Sayitoglu M, Hatirnaz O, Erbilgin Y, Oztunc C, Cinar S, Yildiz I, Celkan T, Anak S, Unuvar A, Devecioglu O, Timur C, Aydogan G, Akcay A, Atay D, Turkkan E, Karaman S, Orhaner B, Sarper N, Deniz G, Ozbek U,Evaluation of *PAX5* gene in the early stages of leukemic B cells in the childhood Bcell acute lymphoblastic leukemia.. Leuk Res. 2011 Aug
- Hatırnaz Ng Özden, Can İsmail, Sinem Fırtına, Erbilgin Yücel, Özbek Uğur, Sayitoğlu Müge (2015). 8q24 bölgesi çocukluk çağı lenfoblastik lösemiler için potansiyel risk oluşturur mu? Deneysel Tıp Dergisi, 5(9)
- Yuk Yin Ng, Sinem Şişko, Özden Hatırnaz Ng, Suzin Çatal Tatonyan, Dilek Sever Kaya, Sinem Fırtına, Müge Sayitoğlu, Uğur Özbek, First Steps of the Genetic Monitorization in Primary Immune Deficiencies in the Lead of Prof. Dr. Işil Barlan in Turkey, Turkish Journal of Immunology, Turk J Immunol 2015;3(2):47

Book Chapter Translations

Essential Medical Genetics, Chapter 18, ISBN -978-605- 4499-82-3, Istanbul Medikal Yayincilik, 2014.

Presentations

International Oral Presentations

- Yucel Erbilgin, Ozden Hatirnaz Ng, Sinem Firtina, Tiraje Celkan, Sema Anak, Zeynep Karakas, Aykan Ozguven, Nazan Sarper, Emine Zengin, Emine Turkkan, Didem Yalcin Atay, Yuk Yin Ng, Omer Dogru, Muge Sayitoglu, Ugur Ozbek, 2013/11/15 American Society of Hematology
- Yucel Erbilgin, Ahmet Emre Eskazan, Ozden Hatirnaz Ng, Ayse Salihoglu, Tugrul Elverdi, Sinem Firtina, Suzin Catal Tatonyan, Seniz Ongoren Aydin, Muhlis Cem Ar, Zafer Baslar, Muge Sayitoglu, A. Burhan Ferhanoglu, Yildiz Aydin, Ugur Ozbek and Teoman Soysal, 2013/11/15 American Society of Hematology

International Selected Posters:

- E. Sun, Ö. Hatırnaz Ng, Y. Erbilgin, S. Fırtına, M. Sayitoğlu, Comprehensive analysis of transcriptomic portrait of T-cell acute lymphoblastic leukemia by RNA sequencing, IXth International Eurasian Hematology Oncology Congress,2018
- Yücel Erbilgin, Sinem Firtina, Ozden Hatirnaz Ng, Tiraje Celkan, Zeynep Karakas, Seniye Sema Anak, Nazan Sarper, Emine Zengin, Ahmet Eskazan, Muge Sayitoglu, Ugur Ozbek, IKZF1 Deletions at Diagnose and Relapse of Childhood B-ALL, American Society of Hematology (ASH) 59th Annual Meeting, 2017
- Y. Erbilgin, O.Hatirnaz ng, S.Firtina, K.Khodzhaev, U.Ozbek, M.Sayitoglu, S.Ugur iseri, Research Biobank for Leukemia, Towards Harmony in Biobanking, Sweeden, 2017
- S. Firtina, M. Sayitoglu, O. Hatirnaz Ng, A. Kiykim, E. Karakoc Aydiner, S. Baris, A. Ozen, S. Nepesov, Y. Camcioglu, E. Hazar Sayar, I. Reisli, S. H. Torun, D. Uygun, F. Cipe, S. Cekic, U. Ozbek, Y. Y. Ng, Molecular Dıagnosıs Of Scıd Patıents By A Custom Desıgned Targeted Next Generatıon Sequencıng Panel, IPIC,2017, Dubai
- Yuk Yin Ng, Sinem Firtina, Özden Hatırnaz Ng, Yıldız Camcıoğlu, Elif Aydıner, Ayça Kiykim, Ayper Somer, Manolya Kara, Alisan Yildiran, Seyhan Kutlug, İsmail Reisli, Şule Haskoloğlu, Funda Çipe, Uğur Özbek, Müge Sayitoğlu Diagnostics Of Primary Antibody Deficiencies Through Targeted Next Generation Sequencing Panel, IPIC,2017, Dubai
- Kiykim, S. Firtina, Y. Zhang,E. Nain,M.Sayitoglu,S.Baris, E.Karakoc-Aydiner, A.Ozen, Hypomorfic *JAK3* and *IL2RG* Mutations Presenting with a Predominantly Antibody DEficiency Phenotype, Meeting of the European Society for Immunodeficiencies (ESID) Meeting, Edinburgh, 2017
- Yucel Erbilgin, Sinem Fırtına, Ozden Hatırnaz, Zeynep Karakas, Tiraje Celkan, Nazan Sarpen, Gonul Aydogan, Khusan Khodzaev, Muge Sayitoglu, Ugur Ozbek, Prognostic significance of IKZF1 deletions in Childhood B-ALL, 10th Biennal of Childhood Leukemia Symposium, Greece, 2016
- S. Firtina, M. Sayitoglu, O. Hatırnaz Ng, A. Kiykim, E. Karakoc Aydıner, S. Nepesov, Y. Camcioglu, İ. Reisli, E. Hazar Sayar, A. Kaya, M.T. Cogurlu, F. Cipe, U. Ozbek, Y.Y. Ng., Identıfıcatıon Of Genetıc Varıatıon By Targeted Next Generatıon Sequencıng In

Turkısh Scıd Patıents, 17th Biennial Meeting of the European Society for Immunodeficiencies (ESID), Barcelona, 2016

- Yuk Yin Ng, Sinem Fırtına, Özden Hatırnaz Ng, Ayça Kıykım, Elif Aydıner, Serdar Nepesov, Yıldız Camcıoğlu, Funda Çipe, Ayşenur Kaya, Müjde Tuba Çöğürlü, Uğur Özbek, Müge Sayitoğlu, Detection of genetic variations in severe combined immunodeficiency (SCID) patients by targeted amplicon sequencing, Molecular Immunology & Immunogenetics (MIMIC) Congress, 2016, Antalya, Turkey
- Serdar Nepesov, Deniz Aygun, Emre Tasdemir, Haluk Çokuğraş, Ozden Hatırnaz Ng, Sinem Sisko, Sinem Fırtına, Ugur Ozbek, Yıldız Camcioglu, A Case with TACI mutation, Molecular Immunology & Immunogenetics (MIMIC) Congress, 2016, Antalya, Turkey
- Y. Erbilgin, B. Islek, S. Firtina, O. Hatirnaz Ng, A. Inal, P. Dogan, T. Celkan, Z. Karakas, N. Sarper, M. Sayitoglu, U. Ozbek; Gene-expression patterns in relapsed B-cell acute lymphoblastic leukemia and potential therapeutic targets ESHG 2016, Barcelona
- Erbilgin Yücel, Hatırnaz Ng Özden, Burak İşlek, Fırtına Sinem, Anak Seniye Sema, Karakaş Zeynep, Ng Yuk Yın, Sayitoğlu Müge, Özbek Uğur. Pathways associated with relapse and high risk in childhood acute lymphoblastic leukemia. EUROPEAN SOCIETY OF HUMAN GENETICS, 23(1), 2015 Glasgow, UK
- Erbilgin Y, Ng O, Fırtına S, Celkan T, Anak S, Karakas Z, Guven A, Sarper N, Zengin E, Turkkan E, Atay D, Ng Y, Dogru O, Sayitoğlu M, Ozbek U. Epigenetic profile of early relapsed Childhood ALL. European Society of Human Genetics, Milan, Italy, May 31-June 3, 2014.
- Erbilgin Y, Ng O, Firtina S, Celkan T, Anak S, Karakas Z, Ozguven A, Sarper N, Zengin E, Turkkan E, Atay D, Ng YY, Dogru O, Sayitoglu M, Ozbek U. Whole genome methylation profile of relapsed childhood leukemia, Translating Epigenomes into Function: a Next-Generation Challenge for Human Disease. FEBS Workshop, Capri, Italy, October 13-16, 2013.
- Sinem Firtina, Muge Sayitoglu, Ozden Hatirnaz Ng, Yucel Erbilgin, Ceren Oztunc, Suzan Cinar, Zeynep Karakas, Tiraje Celkan, Omer Devecioglu, Cetin Timur, Gonul Aydogan, Gunnur Deniz, Ugur Ozbek. Evaluation of *PAX5* gene in the early stages of leukemic B cells in the childhood B cell acute lymphoblastic leukemia, Molecular Immunology & Immunogenetics Congress, 27-29 April 2012, Antalya,Turkey
- Muge Sayitoglu, Ozden Hatirnaz, Yucel Erbilgin, Sinem Ozdemirli, Machteld M. Tiemessen, Jacques J.M van Dongen, Frank J.T. Staal, Ugur Ozbek. Deregulated WNT Signaling in Childhood T- Cell Acute Lymphoblastic Leukemia, T- Cell Acute Lymphoblastic Leukemia (T- ALL) Meets Normal T-Cell Development, 7-9 May, 2010, Mandelieu, France (P29).
- Sinem Ozdemirli, Muge Aydın Sayitoglu, Yucel Erbilgin, Nurhan Mavi, Ozden Hatırnaz, Ugur Ozbek, Pax5 Expression Levels are increased in B-Cell acute lymphoblastic leukemia patients. MediMed Gen, 28 June-1 July 2009, Bilkent-Ankara, Turkey (P40).

Selected National Oral Presentations

- Yuk Yin Ng, Ozden Hatirnaz Ng, Sinem Firtina, Suzan Adin Çinar, H. Haluk Akar, Turkan Patiroglu, Yildiz Camcioglu, Muge Sayitoglu, Ugur Ozbek, Correction Of B-Cell Defficiency In X Linked Agammaglobulinemia (XLA) Mouse Model, Preliminary Data XXIV. National Immunology Congress, 2017, Istanbul
- Yuk Yin Ng, Sinem Firtina, Ozden Hatirnaz Ng, Serdar Nepesov, Ayça Kiykim, Elif Karakoç Aydiner, Yildiz Camcioglu, Ugur Ozbek, Muge Sayitoglu, Analysis of TREC/KREC copy levels in SCID and clinic correlations 3. Clinical Immunology Congress, 2017, Izmir
- Sinem Firtina, Yuk Yin Ng, Ozden Hatirnaz Ng, Yildiz Camcioglu, Elif Aydiner, Manolya Kara, Ayper Somer, Ismail Reisli, Sule Haskologlu, Funda Çipe, Ugur Ozbek, Muge Sayitoglu, Identification of Disease-causing Variants in PAD by NGS, 3. Clinical Immunology Congress, 2017, Izmir
- Sinem Firtina, Yuk Yin Ng, Ozden Hatirnaz Ng, Serdar Nepesov, Yildiz Camcioglu, Ayça Kiykim, Elif Aydiner, Osman Yesilbas, Aysenur Kaya, Funda Çipe, Mujde Tuba Çogurlu, Ugur Ozbek, Muge Sayitoglu, Determination of novel variants in SCID patients : Example of CD3E, XXIII. National Allergy and Immunology Congress, 2016
- Ozdemirli S, Sayitoglu M, Hatirnaz O, Erbilgin Y, Cinar S, Mavi N, Deniz G, Ozbek U. Analysis of *PAX5* Gene in B-Lineage Acute Lymphoblastic Leukemia" 9. Society of Medical Genetics, 2010, Istanbul
- Sinem Ozdemirli, Muge Aydin Sayitoglu, Ozden Hatirnaz, Yucel Erbilgin, Nurhan Mavi, Çetin Timur, Gonul Aydogan, Inci Yildiz, Sema Anak, Fugen Pekun, Arzu Akçay, Ugur Ozbek. Molecular Analysis of *PAX5* Gene in B-cell Acute Leukemia, 35. National Immunology Congress, 2009, Antalya, (S032)

Selected National Posters

- Sinem Firtina, Yuk Yin Ng, Ozden Hatirnaz Ng, Sule Haskologlu, Ayça Kiykim, Elif Aydiner, Selda Hançerli Torun, Manolya Kara, Ayper Somer, Ugur Ozbek, Muge Sayitoglu, BTK gene variations in Primary Antibody Deficiencies. XXIV. National Immunology Congress, 2017, Istanbul
- Sinem Firtina, Yuk Yin Ng, Ozden Hatirnaz Ng, Ayça Kiykim, Elif Aydiner, Serdar Nepesov, Yildiz Camcioglu, Funda Çipe, Aysenur Kaya, Mujde Tuba Çogurlu, Ugur Ozbek, Muge Sayitoglu, Identification of Genetic Variations by NGS in SCID Patients, 2. Clinical Immunology Congress, 2016
- Sinem Firtina, Yucel Erbilgin, Ozden Hatirnaz Ng, Ismail Can, Zeynep Karakas, Tiraje Celkan, Gonul Aydogan, Çetin Timur, Nazan Sarper, Yildiz Yildirmak, Ugur Ozbek, Muge Sayitoglu. Deep sequencing Analysis in *TP53* gene in pediatric ALL, 39.National Hematology Congress, 2013, Antalya
- Sinem Firtina, Yucel Erbilgin, Ugur Ozbek, Muge Sayitoglu, Role of WNT signalling Pathway in Pediatric B-ALL, Medical Genetic Congress, 2012, Bursa

- Sinem Firtina, Yucel Erbilgin, Ozden Hatirnaz, Leyla Agaoglu Ulgur, Tiraje Celkan, Aykan Ozguven, Gonul Aydogan, Arzu Akçay, Emine Turkkan, Didem Atay, Çetin Timur, Asim Yoruk, Zeynep Yildiz Yildirmak, Sebnem Yilmaz, Serap Karaman, Ugur Ozbek, Muge Sayitoglu, Studying of WNT Signalling Pathway in Pediatric B-ALL, National Hematology Congress, 2012, Antalya
- Sinem Ozdemirli, Ozden Hatirnaz, Muge Sayitoglu, Ugur Ozbek 'Methylation of *DKK1* gene in acute leukemia, National Hematology Congress, 2008, Izmir

Selected Projects

- IRON II Study (International Robustness of Next Generation Sequencing) project-European Leukemia Network –European Union Project scholarship student
- Correction Of B-Cell Defficiency In X Linked Agammaglobulinemia (XLA) Mouse Model, No:111S476, 2013-2016
- Genom Wide Expression Analysis Of T-Cell Acute Lymphoblastic Leukemia, No:109S395, 2010-2013
- Intracellular interactions and functional analysis of WNT genes in acute leukemias No: SBAG106S112, 2006-2010
- Detection of *LEF1* gene variants in relapsed pediatric acute leukemia patients by whole genome sequencing, 2012, Turkish Society of Hematology.
- Analysis of *PAX5* gene in B-Lineage Acute Lymphoblastic Leukemia, 2009, Turkish Society of Hematology
- Metagenomic Analysis of Human Gut Microbiota, I.U. BAP, Researcher, 2017
- Identification of Genetic Background in Primary Immunodeficiencies with Next Generation Sequencing, Researcher,2017, No:52575
- Screening Genetic Variations by Next Generation Sequencing in CVID and Correlating the Variants with Prognosis and Clinic, 2015, No: 21237
- Whole Genome Analysis of SCIDs with Next Generation Technologies, 2012, Priority projects of I.U. No: 20499
- Whole Genome Analysis of High Risk Acute Leukemias, 2011, Outsourced financial Project in I.U. No:20520
- Analysis of *CRLF2* and *JAK2* hot spot mutations in acute lymphoblastic leukemias by Amplicon sequencing,2010 No:48096
- Investigation of *IKZF1* mutations by Next Generation Sequencing in B-cell acute lymphoblastic leukemia, 2010, no:35624
- Sorting of healthy B cell subgorups from bone marrow samples by flow cytometry, 2009, No: 3817

National Invited Talks

- Sinem Firtina, Applications of Next Generation Sequencing technics in Rare Diseases, 5th Course in Next Generation Sequencing – Hybrid course*,* 2016, Istanbul
- Sinem Firtina, Identification of Genetic Background in Primary Immunodeficiencies with Next Generation Sequencing, DETAE Young Researcher Meeting, 2015, Istanbul
- Sinem Firtina, Analysis and Clinical Applications of Amplicon Sequencing, 2th Course in Next Generation Sequencing - Hybrid course, 2013, Istanbul

Courses/workshops

- Methylome Workshop, 2013, Istanbul
- COST, European cooperation in science and technology Meeting, 2012, Istanbul,
- Leica Confocal Workshop, Innovations in Confocal Microscopy, 2012, Istanbul
- 2nd Turkish-Us Cytometry Workshop, 2009, Istanbul
- European society of human genetics (ESHG) course ' Medical Genetics and Genomics analysis in isolated and consanguineous populations', 2009, Ankara
- 2nd course in integration of cytogenetics, microarrays and massive sequencing in biomedical and clinical research hybrid course, 2009, Istanbul

Selected Awards

- XII. National Medical Genetics Congress, Determination of Disease Causing Variants in PID, *Third prize for best oral presentations*, 2016
- Clinical Immunology Congress, Identification of Genetic Variants in SCID, *Third prize for best presentations,* 2016
- Turkish Society of Hematology Young Research Awards, Studying of *WNT* Signalling Pathway in Pediatric B-ALL 2012
- Turkish Society of Hematology Young Research Awards, Analysis of *PAX5* gene in B-ALL, 2008
- Turkish Society of Hematology Young Research Awards, Methylation of *DKK1* gene in acute leukemia, 2008