

## CURRICULUM VITAE

### ELIF UZ



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#### EDUCATION

PhD (2008) Bilkent University, Department of Molecular Biology and Genetics

MSc (2002) Middle East Technical University, Department of Biotechnology

BSc (1999) Middle East Technical University, Department of Biology

#### WORK EXPERIENCE

2013-today Assistant professor at the Department of Molecular Biology and Genetics, Uludag University

2010-2013 Assistant professor at the Department of Biology, Duzce University

2008-2010 Post-doctoral fellow in Gene Mapping Laboratory, Hacettepe University

2002-2008 Teaching Assistant at Department of Molecular Biology and Genetics in Bilkent University

1999-2001 part-time assistant at "Development of Science Education Project" in TED Ankara High School.

## MEMBERSHIPS

1. European Society for Human Genetics

## PROJECTS

Dr. Uz was participated as researcher in the projects below.

2016-2019	TÜBİTAK-SBAG (215S620)- The investigation of the mediation of cylooxygenase and lypooxygenase pathways on centrally injected nesfatin-1 evoked cardiovascular effects
2015-2017	TÜBİTAK-TOVAG (114O809)- A comparative Study on Some Biological, Histological and Genetic Properties of Marine and Freshwater Populations of the Sand Smelt, <i>Atherina boyeri</i> (Risso, 1810)
2014-2017	TÜBİTAK-SBAG (114S354)- Use of genetically modified tolerant dendritic cells in mouse arthritis models
2009-2011	TÜBİTAK-SBAG(108S420)-E-RARE consortium- A Clinical and Scientific Approach to Craniofacial Malformations
2007-2008	TÜBİTAK-SBAG-HD-230-Mapping of disease locus in Unertan Syndrome
2006-2009	TÜBİTAK-SBAG-3334- X-chromosome inactivation and autoimmune disorders
2004-2007	ICGEB-CRP/TUR04-01- Identification of the molecular mechanisms in Rett syndrome
1999-2002	International Atomic Energy Agency- IAEA/ 10767/R0/Regular Budget Fund- Application of Microsatellite and AFLP markers in wheat for selecting and breeding desired varieties

## PUBLICATIONS

1. Cetinkaya A, Xiong JR, Vargel İ, Kösemehmetoğlu K, Canter Hİ, Gerdan ÖF, Longo N, Alzahrani A, Camps MP, Taskiran EZ, Laupheimer S, Botto LD, Paramalingam E, Gormez Z, **Uz E**, Yuksel B, Ruacan Ş, Sağıroğlu MŞ, Takahashi T, Reversade B, Akarsu NA. Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling. (2016) *Am J Hum Genet.* 99 :299-317.
2. Halacli SO, Ayvaz DC, Sun-Tan C, Erman B, **Uz E**, Yilmaz DY, Ozgul K, Tezcan İ, Sanal O. STK4 (MST1) deficiency in two siblings with autoimmune cytopenias: A novel mutation. (2015) *Clin Immunol.* 161: 316-323.
3. Alanay Y, Ergüner B, Utine E, Haçarız O, Kiper PO, Taşkiran EZ, Perçin F, **Uz E**, Sağıroğlu MŞ, Yuksel B, Boduroglu K, Akarsu NA. TCMO1 deficiency causes autosomal recessive cerebropathic dysplasia. (2014) *Am J Med Genet A.* 164: 291-304.
4. Keupp K, Li Y, Vargel I, Hoischen A, Richardson R, Neveling K, Alanay Y, **Uz E**, Elcioğlu N, Rachwalski M, Kamaci S, Tunçbilek G, Akin B, Grötzinger J, Konas E, Mavili E, Müller-Newen G, Collmann H, Roscioli T, Buckley MF, Yigit G, Gilissen C, Kress W, Veltman J, Hammerschmidt M, Akarsu NA, Wollnik B. Mutations in the interleukin receptor IL11RA cause autosomal recessive Crouzon-like craniosynostosis. (2013) *Mol Genet Genomic Med.* 1: 223-37.
5. Bonnard C, Strobl AC, Shboul M, Lee H, Merriman B, Nelson SF, Ababneh OH, **Uz E**, Güran T, Kayserili H, Hamamy H, Reversade B. Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. (2012) *Nat Genet.* 44: 709-13.
6. Putoux A, Thomas S, Coene KL, Davis EE, Alanay Y, Ogur G, **Uz E**, Buzas D, Gomes C, Patrier S, Bennett CL, Elkhartoufi N, Frison MH, Rigonnot L, Joyé N, Pruvost S, Utine GE, Boduroglu K, Nitschke P, Fertitta L, Thauvin-Robinet C, Munnich A, Cormier-Daire V, Hennekam R, Colin E, Akarsu NA, Bole-Feysot C, Cagnard N, Schmitt A, Goudin N, Lyonnet S, Encha-Razavi F, Siffroi JP, Winey M, Katsanis N, Gonzales M, Vekemans M, Beales PL, Attié-Bitach T. KIF7 mutations cause fetal hydroletharus and acrocallosal syndromes. (2011) *Nat Genet.* 43: 601-6.

7. Yağcıoğlu AE, İlhan BÇ, Göktaş MT, Babaoğlu MO, **Uz E**, Yazıcı MK. Agranulocytosis related to clozapine in monozygotic twins and association with allelic variants of multidrug resistance gene MDR1. (2011) *J Clin Psychopharmacol*. 31: 247-9.
8. **Uz E**, Alanay Y, Aktas D, Vargel I, Gucer S, Tuncbilek G, von Eggeling F, Yilmaz E, Deren O, Posorski N, Ozdag H, Liehr T, Balci S, Alikasifoglu M, Wollnik B, Akarsu AN. Disruption of Paired-Tail Homeobox Gene ALX1 (Cart1) Causes Extreme Microphthalmia and Severe Facial Clefting: Expanding the Spectrum of Autosomal Recessive "ALX-Related Frontonasal Dysplasia". (2010) *Am J Hum Genet*. 86: 789-96.
9. Kayserili H, **Uz E**, Niessen C, Vargel I, Alanay Y, Tuncbilek G, Yigit G, Uyguner O, Candan S, Okur H, Kaygin S, Balci S, Mavili E, Alikasifoglu M, Haase I, Wollnik B, Akarsu NA. ALX4 dysfunction disrupts craniofacial and epidermal development. (2009) *Hum Mol Genet*. 18: 4357-66.
10. **Uz E**, Yildirim-Ersoy F, Hakki EE, Akaya MS. Genetic relationship of wild einkorn based on geographical distribution in Anatolia and Thrace using AFLP markers. (2009) *Journal of Applied Biological Sciences* 3: 20-5.
11. **Uz E**, Mustafa C, Topaloglu R, Bilginer Y, Dursun A, Kasapcopur O, Ozen S, Bakkaloglu A, Ozcelik T. Extremely skewed X-chromosome inactivation is increased in juvenile idiopathic arthritis. (2009) *Arthritis Rheum*. 60: 3410-2.
12. Chabchoub G, **Uz E**, Maalej A, Mustafa CA, Rebai A, Mnif M, Bahloul Z, Farid NR, Ozcelik T, Ayadi H. Analysis of skewed X-chromosome inactivation in females with rheumatoid arthritis and autoimmune thyroid diseases. (2009) *Arthritis Res Ther*. 11: R106.
13. Plagnol V, **Uz E**, Wallace C, Stevens H, Clayton D, Ozcelik T, Todd JA. Extreme clonality in lymphoblastoid cell lines with implications for allele specific expression analyses. (2008) *PLoS ONE* 3: e2966.
14. Ozcelik T, Akarsu N, **Uz E**, Caglayan S, Gulsuner S, Onat OE, Tan M, Tan U. Reply to Herz *et al.* and Humphrey *et al.*: Genetic heterogeneity of cerebellar hypoplasia with quadrupedal locomotion. (2008) *Proc Natl Acad Sci U S A*. 105: E32-3.

15. Ozcelik T, Akarsu N, **Uz E**, Caglayan S, Gulsuner S, Onat OE, Tan M, Tan U. Mutations in the very low density lipoprotein receptor (*VLDLR*) cause cerebellar hypoplasia and quadrupedal locomotion in humans. (2008) *Proc Natl Acad Sci U S A*. 105: 4232-6.
16. **Uz E**, Loubiere LS, Gadi VK, Ozbalkan Z, Stewart J, Nelson JL, Ozcelik T. Skewed X chromosome inactivation in scleroderma. (2007) *Clin Rev Allergy&Immun* 34: 352-5.
17. Kaplan Y, Vargel I, Kansu T, Akin B, Rohmann E, Kamaci S, **Uz E**, Ozcelik T, Wollnik B, Akarsu NA. Skewed X-inactivation in an X-linked Nystagmus Family Resulted From a Novel, p.R229G, Missense Mutation in the *FRMD7* Gene. (2008) *Br J Ophthalmol*. 92: 135-41.
18. **Uz E**, Dolen I, Al AR, Ozcelik T, Extremely skewed X-chromosome inactivation is increased in pre-eclampsia. (2007) *Hum Genet*. 121:101-5.
19. Ozcelik T, **Uz E**, Akyerli CB, Bagislar S, Mustafa CA, Gursoy A, Akarsu N, Toruner G, Kamel N, Gullu S. Evidence from autoimmune thyroiditis of skewed X-chromosome inactivation in female predisposition to autoimmunity. (2006) *Eur J Hum Genet*. 14:791-7.