CURRICULUM VITAE for Zeynep Tümer

Degrees

2014 **DMSc** (Doktor of Medical Sciences, dr.med.), Faculty of Health Sciences, University of Copenhagen

1996 **PhD** (Doktor of Philosophy), Faculty of Health Sciences, University of Copenhagen

1985 **MD** (Medical Degree) Egean University Medical School, Izmir, Turkey.

Appointments*

2008-2016	**Research professor**, Kennedy Center, Department of Clinical Genetics, Copenhagen University Hospital, Rigshospitalet
2009-2014	**Adjunct professor**, ICMM, Faculty of Health Sciences, KU
1999-2008	**Associate professor**, ICMM, Faculty of Health Sciences, KU
1996-1999	**Assistant professor**, Department of Medical Genetics, IMBG, Faculty of Health Sciences, KU
1996-1999	**Scientific consultant**, John F. Kennedy Institute
1996	**Scientific consultant** in Molecular Genetics Department, Institute for Experimental Medicine, Istanbul University, Turkey, under UNESCO program TOKTEN (Transfer Of Knowledge Through Expatriate Nations)
1990-1996	Clinical assistant, Research fellow and Senior scientist, John F. Kennedy Institute.
1994-1996	**Senior scientist**, John F. Kennedy Institute.
1990-1993	**Research fellow** (PhD studies) John F. Kennedy Institute.
1990	**Clinical assistant**, Department of Medical Genetics, John F. Kennedy Institute.
1989-1990	**Project leader**: "Establishing selfhelping groups among immigrant families with handicapped children" and "Introducing prenatal diagnosis of hemoglobinopathies in Denmark," John F. Kennedy Institute.
1988-1989	**Voluntary**, Immune-haematology laboratory, States Serum Institute, Copenhagen
1989	**Medical practice,** Department for Multihandicapped, Vangedehuse, Copenhagen (two months)
1988-1989	**Research fellow,** Immune-hematology laboratorium, Statens Serum Institute, Copenhagen (voluntary)
1986-1987	**Part-time clinical assistant,** Biga Ultrasound Clinic, Çanakkale, Turkey (1.5.1986 - 1.11.1987): Examination of more than 1000 patients during this period.
1985-1987	**General practitioner**, Departments of Internal Medicine and Surgery in Biga State Hospital, Çanakkale, Turkey

** **KU,** University of Copenhagen; **ICMM**, Department of Cellular and Molecular Medicine; **IMBG**, Institute for Medical Biochemistry and Genetics, Faculty of Health Sciences; **Kennedy Center** (Previous **John F. Kennedy Institute**) was a national research institution for genetics and visual and mental handicap (Sektorforskningsinstitution) before being part of Copenhagen University Hospital, Rigshospitalet.*

Management and administrative experience

2008-2016	**Chief** of research and development of *Applied Human Molecular Genetics* section, Kennedy Center, Department of Clinical Genetics, Copenhagen University Hospital, Rigshospitalet
2001-2008	**Vice director**, Wilhelm Johannsen Centre for Functional Genome Research established by the Danish National Research Foundation, ICMM, Faculty of Health Sciences, KU
2004-2008	**Leader** of the Cytogenetic Diagnosis Lab., ICMM, Faculty of Health Sciences, KU
2006	**Head of Department**, Department of Medical Genetics, IMBG, Faculty of Health Sciences, KU
2005-2006	**Deputy Head of Department**, Department of Medical Genetics, IMBG, Faculty of Health Sciences, KU

Administrative education

- Research Management Course, Copenhagen Business School (CBS-SIMI Executive), Copenhagen, 2011
- Management of Employees, Leadership Development Program, Capital Region of Denmark, 2015

Publications

- **170 scientific manuscripts** (published or in press) in peer-reviewed journals with impact factor (abstracts, submitted manuscripts and manuscripts in preparation are excluded). **First author in 28 manuscripts; single author in 3 manuscripts; senior author in 61 manuscripts.**
- **13 book-chapters/proceedings**
- **8 miscellaneous publications** e.g. book translation etc

Research supervision

- Current supervisions: **1 PhD** student, 1 research assistant, 1 MSc
- Completed supervisions: **21 PhD, 20 MSc, 24 BSc**, and several international research fellows/Erasmus students

Scientific meetings:

>100 invited lectures/oral presentations; >150 other scientific presentations.

Assessment committees

Member or chairman of several national/international PhD and MSc assessment committees; professor assessment committee; several hiring committees, censorship in bachelor and candidate projects; EU projects

Referee-reviewer

Nature Rev Neurology, Am J Hum Genet, FASEB J, Am J Med Genet, Acta Paed, Chem Rev, Clin Genet, Gene, J Inher Met Dis, Eur J Hum Genet, Hum Genet, Brain Res, Brain Dev, Biochim Biophys Acta, Gen Med, Neuropaed, Biological Psychiatry, PlosOne, J Med Genet etc.

Board Membership

- **European Society of Human Genetics** (ESHG) board member (2015-2020)
- Ethical Advisory Board for **Genome Denmark** (a national platform for sequencing and bioinformatics (2013-2015)
- Editorial board member of American Journal of Neurodegenerative Diseases (2012-).

Scientific collaboration

National collaborations:
Several research institutions and clinical departments in Denmark, including Faculty of Health Sciences, KU; Aarhus University and Skejby Hospital; Center for Biological Sequences (CBS), Technical University of Denmark (DTU); Central Region (Region H) hospital clinics, including Herlev hospital; Department of Genomic Medicine, Rigshospitalet, Denmark.

International collaborations with contract:

- Pan-European collaboration on Tourette Syndrome (TS)
  - European Network for TS (**COST action BM0905**) – representative of Denmark, management committee (http://www.cost.eu/COST_Actions/bmbs/BM0905)
  - TS-EUROTRAIN (**Marie-Curie ITN**, PITN-GA-2012-316978)] – Workpackage leadership, co-leadership (http://ts-eurotrain.eu/)
- Pan-European collaboration on imprinting disorders (ID)
  - European Network for Human Congenital Imprinting Disorders (EUCID) **COST action BM1208** (http://www.imprinting-disorders.eu/)
- International collaboration with Japan, Armenia, Georgia and United Kingdom on mitochondria disorders.

International Science and Technology Center (ISTC) project – Molecular pathogenesis of mitochondrial oxphos diseases (#A-2151) (http://www.istc.int/en/project/53B1E92D8E7B7D2846257F1F00313FAB)

Fund-raising

Grant support as PI at the level of **16 Million DKK since 2008** during employment at the Kennedy Centre. Establishment of WJC, Danish National Research Foundation Center as vice director 2001-2008 (PI, Center leader Prof. Niels Tommerup). Co-applicant of several grants for continuance of our research team at WJC (> 30 M DKK) and at the Kennedy Center (> 13M DKK).

Grants at PI level:

- The Ministy of Children and Education: E-learning in genetics– 500.000 DKK (2013-2015)
- TS-EUROTRAIN (Marie Curie ITN): Tourette Syndrome – Total budget 3M ECU, Kennedy budget 2.4M DKK (2013-2015)
- Lundbeck Foundation: Tourette Project— 3M (2014-2016)
- Danish Strategic Research Council: Novel Treatments of Cognitive Dysfunction (COGNITO)–Total budget 17M DKK, KC budget 1.2M DKK (2012-2013)
- Dagmar Marshalls Fond – Tourette Syndrome and co-morbidity disorders – 60.000 DKK (2011)
- Signe og Peter Gregersens Mindefond: Neurodevelopmental Disorders and Microarray – 48.000 DKK (2011)
- PhD Grant (KU) -- Tourette Syndrome – 2.4M DKK (2011-2013)
- Danish Agency for Science, Technology and Innovation (FI), Joint PhD with DTU: Nanocytogenetics – 2.4M DKK (2010-2013)
- Lundbeck Foundation: Tourette Project – 3M DKK (2009-2011)
- Neurocluster: Age Related X-inactivation – 600.000 DKK (2009)

Clinical and diagnostic activities

2008- present (Kennedy Center, KC):

- Leader of the Microarray diagnostic facility:
  - Establishment of high resolution microarray technology (Affymetrix CytoScan HD).
  - Responsible for the method and reporting of the results to the clinical departments.
  - Data analysis and evaluation of the results and their relevance to the phenotype of the patients.
  - Education of young clinicians in microarray evaluation and reporting of the results.
- Establishment of the NGS gene panel for the diagnosis of Cornelia de Lange Syndrome (CdLS).
- Method responsibility for the diagnostic FISH (Flourescent in situ hybridization) analysis.
- External quality assessment (EQA) for chromosome microarray, NGS and FISH (CEQAS – www.ceqas.org)
- Imprinting disorders (as part of the EUCID Cost Action, FP7)
  - Member of the Silver Russell Syndrome clinical consensus study panel
  - Preparation of a public database on the (epi)genetic findings of Silver Russell Syndrome (in progress).
- Preparation of Gene Cards for Axenfeld Rieger Syndrome, Menkes disease and occipital horn syndrome (http://www.genecards.org/)
- Curation of a public database for Menkes disease as part of Leiden Open Variation Database (LOVD2). The database includes genotype and genotype data of all published and known cases (https://grenada.lumc.nl/LOVD2/MD/home.php?select_db=ATP7A)

- Acreditation
- Medunderskriver: Cornelia de Lange syndrome, Menkes disease, Wilson disease, qPCR deletions-/duplikationsanalyse (postnatal og prenatal)

Earlier diagnostic activities

2004-2008

Leader of the Cytogenetics Laboratory, Medical Genetics Clinic, ICMM, Faculty of Health Sciences, KU. Responsible for the diagnosis and reporting of the patients referred to the clinic.

1993-1997

Establishment of genetic diagnostic test for Menkes disease at KC as the first laboratory Worldwide: performance of the genetic test (pre- and postnatal diagnosis), evaluation of the results, reporting of the results. Investigated about 300 patients and their families for mutations and carried out post-/pre-/carrier diagnosis during this period.

Research areas ad interests

Main research areas: Molecular disease mechanisms in monogene and complex disorders; epigenetics and imprinting; cell biology including intracellular metal metabolism and homeostasis, miRNA.

Main research interest is to understand the underlying molecular disease mechanisms (genetic and epigenetic mechanisms, oxidative stress and secondary deficiencies of mitochondria including mitophagy) involved in various human disorders including neurodevelopmental and neuropsychiatric disorders, metabolic disorders, congenital disorders etc. The genetic mechanisms are elucidated through investigation of cytogenetic abnormalities (using high-throughput mapping of translocations), copy number variations (using high resolution chip technologies), and sequence variations (using next-generation sequencing, NGS, based technologies) followed by functional studies (quantitative methodologies, tissue in situ hybridization techniques). The epigenetic mechanisms are investigated through methylation analysis, and investigation of miRNAs and other ncRNAs. Main aim of my research is to apply the results to clinics usage (applied/translational genetics).

The current projects include, but not limited to: 1) Investigation of the role of epigenetic and genetic mechanisms in Tourette syndrome and co-morbidities (ADHD, OCD, Autism) ; 2) underlying (epi)genetic changes in intellectual disabilities; 3 imprinting disorders (including multi locus methylation deficiency disorders); 4) copy number variation related congenital disorders; 5) congenital developmental defects including the cohesin deficiency disorders, Cornelia de Lange syndrome; 6) involvement of oxidative stress and mitochondria deficiency in movement disorders (fx. ataxia, Tourette syndrome, Rett-syndrome).