Curriculum Vitae

Katharina Wimmer, PhD
Associate Professor (Ao.Univ.-Prof.)
Division Human Genetics
Medical University Innsbruck, Austria

Education/Degrees/Awards

10/2002	**Habilitation for Medical Biology and Human Genetics**, Medical University Vienna, Austria; Title: *Identification of genetic and epigenetic alterations in solid tumors using Restriction Landmark Genomic Scanning: considering as example neuroblastoma.* Habilitation Award of the *Verein zur Förderung von Wissenschaft und Forschung in den neuen Universitätskliniken am Allgemeinen Krankenhaus der Stadt Wien*
1/1994	**"Doctor rerum naturalium technicarum" (PhD)** from BOKU University of Natural Resources and Applied Life Sciences, Vienna, Austria
6/1990 - 1/1994	Doctoral thesis at Department of Applied Microbiology; title: *Characterization of cell culture supernatants of a human melanoma cell and a recombinant CHO cell (two-dimensional electrophoresis applied to control production systems with animal cells and detection of proliferation stimulating factors in melanoma cells)*
6/1990	**"Diplom Ingenieur für Lebensmittel und Biotechnologie " (Dipl.Ing.)** from BOKU University
3 - 11/1989	Diploma thesis at the Department of Applied Microbiology, title: *Isolation and characterization of sulfide and carbon disulfide* metabolizing bacteria.
10/1984 - 6/1990	Study of biotechnology at BOKU University
10/1983 - 9/1984	Study of agriculture at BOKU University

Scientific postdoctoral training

9/97 - 9/98	Research fellow at the University of Vienna, Medical faculty, Department of Medical Biology
1/95 - 7/97	Research fellow at the University of Michigan, Comprehensive Cancer Center, Department of Pediatrics, Ann Arbor, MI, USA (laboratory/ Prof. S.M. Hanash, MD PhD)
6 - 8/1994	Continuation training at the Hôpital Cantonal Universitaire de Génève, Laboratoire Central de Chimie Clinique, Geneva, Switzerland (Department Science & Development/ Prof. Dr. med. D. Hochstrasser)
1/1994 -12/94	Research fellow at the BOKU University of Natural Resources and Applied Life Sciences, Vienna, Austria (Department of Applied Microbiology)

Academic positions

Since 4/2014	**Deputy head** Division of Human Genetics, Medical University Innsbruck
Since 12/2008	**Associate professor** (Ao. Univ.-Prof.) at the Medical University Innsbruck, Division of Human Genetics
1 – 6/2005	**Visiting professor** at the Department of Genetics, Medical Genomics Laboratory (head Prof. L. Messiaen), University of Alabama at Birmingham, AL, USA (sabbatical leave from the Medical University Vienna)
3/2003 – 11/2008	**Associate professor** (Ao. Univ.-Prof.) at the Medical University of Vienna, Department of Medical Genetics
10/1998 - 2/2003	**Scientific assistant** (Universitäts Assistent) at the Medical University of Vienna, Department of Medical Biology

Field of activities at the Division of Human Genetics, Medical University Innsbruck

Diagnostic work

I established and supervise the onco-genetic diagnostic laboratory offering the molecular genetic analysis for a broad spectrum hereditary cancer susceptibility syndromes (>600 cases/year) with specific focus on the neurofibromatoses, Lynch syndrome, constitutional mismatch repair deficiency syndrome and polyposis syndromes.

Research interests

In general my research is strongly associated with the onco-genetic diagnostic laboratory. Hence, one of the major aims of my research lab is it to develop and improve diagnostic tools for the identification and classification of mutations.

Topic A: Evaluation of 'atypical' splice mutations

The broad application of RNA-based assays developed in our laboratory proved to substantially increase mutation detection rates in several tumor suppressor genes by effectively uncovering splice alterations caused by mutations that either fully escape the detection of gDNA based assays or cannot readily be classified as deleterious from the analysis of gDNA only. Evaluation of these splice mutations allows to elucidating basic mechanism of splice site definition and inactivation. My laboratory has a long-standing interest in the characterization of the splice mutations outside the canonical GT-AG dinucleotides in the *NF1* and other tumor suppressor genes with the aim to elucidate molecular mechanisms of splice site disruption and to improve tools to predict splicing effects of genomic variants.

Topic B: Constitutional mismatch repair deficiency (CMMRD) syndrome

CMMRD is a rare autosomal recessively inherited cancer susceptibility syndrome caused by biallelic mutations in one of the four DNA mismatch repair (MMR) genes, *MLH1*, *MSH2*, *MSH6* and *PMS2*. CMMRD shows clinical overlap with other cancer susceptibility syndromes, notably neurofibromatosis type 1 (NF1) and polyposis syndromes. Being recognized only recently as a distinct childhood cancer susceptibility syndrome, there is still a lack of knowledge on the natural history and the clinical expression of this syndrome. Approximately 60% of all CMMRD patients carry mutations in the notoriously difficult to analyze *PMS2* gene for which we developed effective and reliable RNA-based assays and improved MLPA analysis which proved to be pivotal to circumvent diagnostic obstacles that are caused by the presence of pseudogenes of this gene. By stating a molecular diagnosis in a number of patients the lab substantially contributed to the delineation of the tumor spectrum and non-neoplastic features of the syndrome. This led under my lead to the development of clinical diagnostic criteria proposed by the European consortium care for CMMRD (C4CMMRD. Future goals are to evaluate in close collaboration with C4CMMRD, which I cofounded, clinical data on CMMRD patients, both with the ultimate goal to improve the management of CMMRD patients as well as to delineate differential diagnoses to CMMRD and assess the prevalence of CMMRD among specific cancer entities.

Teaching

- Lectures and practical courses in human genetics and cancer genetics for medical students and students of molecular medicine
- Supervision of Diploma/Master(6) and PhD (1) students

Grants

2018 ongoing	Austrian Science Fund: KLI 734-B26 *Prevalence of CMMRD in NF1/SPRED1 Mutation Negatives*
2009 - 2011	Austrian Science Fund: P21172 *Analysis of PMS2 hybrid alleles*
2003 - 2005	Marie Curie fellowship of the European Commission: QLK6-CT-2002 51681 *Identification of genes with prognostic and therapeutic relevance in hepatoblatoma* (submitted with the postdoc Dr. A. Zatkova)
2004	Hochschuljubiläumsstiftung der Stadt Wien: *Improvement of the diagnostic methods for NF1 and NF2*
2001 - 2004	Austrian Science Fund: P14944-GEN *Characterization of a new 8q12-13 amplicon in hepatoblastom*
2002	Hochschuljubiläumsstiftung der Stadt Wien: H1170/2002 *The role of the NF1 gene in juvenile myelomonocytic leukemia*
1998 - 2000	Austrian Science Fund: P12942-GEN *Identification of novel genes in hepatoblastoma* (together with and submitted by Univ.-Prof. Dr. C. Fonatsch)
1999 - 2000	Kommission Onkologie der Medizinischen Fakultät der Universität Wien: *Mutation of the tumor suppressor gene NF1 in NF 1-associated tumours*
1999	Jubiläumsfonds der Österreichischen Nationalbank: Projekt Nr. 7519 *Development of a yeast-fusion-protein assay for mutation analysis in the neurofibromatosis type 1 gene*
1995 - 1997	Erwin Schrödinger fellowship or the Austrian Science Fund: J01089 *Characterisation of a proliferation-regulated cytosolic phospho-protein*
1991 -1993	Austrian Science Fund: P08810-MOB (PhD fellowship) (together with and submitted by Dr. M. Reiter)

Memberships

- Austrian Society of Human Genetics (ÖGH)
- European Society of Human Genetics (ESHG)
- Interantional Society for Gastrointestinal Hereditary Tumours (InSiGHT)
- Scientific consultant of the *Austrian von Recklinghausen-Neurofibromatosis Society since 2002* (referred expertise see also *Medical Tribune*, 2002, 9:14)
- Member of the ‚Wissenschaftlicher Ausschuss für Genanalyse und Gentherapie am Menschen' (WAGG) of the Bundesministerium für Frauen und Gesundheit (Austrian Ministry for Women and Health) term 11/2013-10/2018

Publications

Total number of publications: peer reviewed research articles (79), peer reviewed review articles (13), letters to editor/commentaries (3), educational (7), book chapters (6)

H-Index: 30 (excluding self-citations)

Link to all Publications in MedLine