

Curriculum Vitae

Karen Brøndum-Nielsen, born August 7, 1947

Education and Employment

Graduated from University of Copenhagen, Medical school 1974.

Thesis, (Dr. med.) University of Copenhagen, 1985. Title: X-linked mental retardation with the fragile site at Xq28.

Specialist (speciallæge) in Pediatrics 1987.

Specialist /Swedish) (speciallæge) in Clinical Genetics 1988. Danish specialist aut. 1998

From 1976 research assistant at the John F. Kennedy Institute, from 1977-82 as clinical assistant (klinisk assistent).

Clinical training at departments in hospitals in Copenhagen 1974-76 and 1982-88.

Consultant (bitr. Överlakare) at the Department of Clinical Genetics, Karolinska Hospital, Stockholm, Sweden 1988-91. (Head of Department (klinikchef) from 1990-91).

Associate professor (lektor) at Department of Medical Genetics, Odense University 1991-93.

Professor (adjunct) (adjungeret professor) in Clinical Genetics, University of Copenhagen 1998-2013.

Professor in clinical genetics, Department of Clinical Medicine, University of Copenhagen, from 2013.

Consultant (overlæge) and Head of chromosome laboratory at the John F Kennedy Institute 1993, from 2001 Head of Institute. From 2003-13 Head and Director of the Kennedy institute-National Eye Clinic, now Kennedy Center).

In July 2012, the Kennedy Center was fused with the Capital region (region Hovedstaden) as part of University Hospital of Copenhagen, JulianeMarieCentret, Rigshospitalet.

Present employment

Clinical professor and consultant, Kennedy Center, JMC, Rigshospitalet, University Hospital of Copenhagen

Boards, Academic duties, etc.

Member of the board of Danish Society for research in Mental Retardation (oligofreni) 1982-88.

Member of the Board of the Danish Society of Medical Genetics 1992-96. President 1994-96.

Member of EU concerted action IMGSAC (International Molecular Genetic Study of Autism Consortium) 1998-

Official Opponent at several thesis and Ph.D. dissertations.

Member and/or chairperson of evaluation committees for several academic positions in Denmark and Sweden (professorships, associate professors)

Member of Danish Medical Research Council 1999-2005

Member of the Board of European Society of Human Genetics 2001-2007

Member of assessment committee for the Swedish Medical Research Council 2006-2008

Danish coordinator for Orphanet (European webbased information network on rare diseases) 2003-2010

Member of editorial board of Journal of Medical Genetics 2007-2010

Member of the editorial board of Intractable & Rare diseases Research 2011-

Reviewer for several journals, Am J Hum Genet, Eur J Hum Genet, Am J Med Genet, Clin Genet, Orphanet J Rare Dis, BMJ, Acta paediatrica and others

Member of various working groups at National Board of Health, on rare diseases, prenatal diagnosis and screening, disease risk and consanguinity, Specialty plan for Clinical Genetics, Current member of working group on National strategy for rare diseases, 2012-

Scientific Supervision

Supervision of two diploma students.

Supervision (Principal/faculty supervisor or project supervisor) of 11 graduated PhD students, 2 currently ongoing.

Publications and oral papers, posters

213 published papers in peer-reviewed journals. Published under Nielsen KB (till approx 1995) or Brondum-Nielsen K.

More than 150 oral papers, posters, etc.

Teaching experience

From 1977 -1986: Lectures at various professional educational institutions: Børnehave- og Socialpædagogiske seminarier, Hvidovre sygeplejeskole.

Clinical Lecturer (klinisk lektor) in Pediatrics 1986-89, Gentofte hospital and University of Copenhagen.

External examiner (Censor) in Genetics at the University of Copenhagen 1985-88

External examiner (Censor) in Clinical Genetics at University of Århus 1989-92

Lectures at Stockholm University pregraduate course in genetics 1989-90

Lectures in clinical genetics at specialist education in ophthalmology, pediatric pathology, Stockholm 1989-90

Lectures for undergraduate medical students, in general genetics and clinical genetics 1991-93, University of Southern Denmark

Organizer of exams and examination papers in Medical Genetics, 1992, University of Southern Denmark

Lectures in clinical genetics at courses for various specialist training (pediatrics, neurology, ophthalmology, psychiatry, oncology) since 1991

Regular lectures at courses for specialist training in clinical genetics (A kursus, U Kursus) since 1991. Topics clinical and molecular cytogenetics, syndromes and etiology of mental retardation,

Evaluator of master program in health sciences, study direction genetic counselling, University of Bergen, Norway, 2004.

Areas of scientific interests, scientific profile

Clinical genetics, Genotype-phenotype correlations, genetic aberrations associated with dysmorphic development, syndromes with intellectual disability and/or visual impairment.

Chromosome aberrations; prenatal diagnosis

Fragile X syndrome; Autism spectrum disorders