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NBIA - Neurodegeneration w/ Brain Iron Accumulation, Updated: December 13, 2016 by John Anastasiadis, email: **anastasiadis77@gmail.com**, tel: +1 908 200 5063

Clinical Centers & Research Organizations

Europe

United Kingdom

University of Newcastle upon Tyne
The Medical School
Mitochondrial Research Group
Prof. Patrick Chinnery NE2 4HH, Newcastle upon Tyne, UK Phone: (+44) 0191 208 5101 Email: patrick.chinnery@ncl.ac.uk Website: http://www.ncl.ac.uk/biomedicine/ Patrick's new email: pfc25@medschl.cam.ac.uk
International NBIA Patient Registry Clinical Center

⚠ Patrick Chinnery is no longer working at the university hospital of Newcastle, UK. I'm currently not sure whether someone else from Newcastle stepped in there for NBIA research and/or if Dr. Chinnery continues with his NBIA investigations in his new job. http://www.mrc-mbu.cam.ac.uk/people/patrick-chinnery This would require further investigation.

- **Neuroferritinopathy** research

University College London, Dr Manju Kurian Email: manju.kurian@ucl.ac.uk; Dr Apostolos Papandreou Email: apostolos.papandreou@ucl.ac.uk

- **BPAN** cell research
- **PLAN** gene therapy
- **PLAN** natural history study

Italy

The Foundation of the Carlo Besta Neurological Institute, IRCCS
Nardo Nardocci Via Celoria 11 20133 Milano, Italy Phone: (+39) 02 2394 2210 Email: nardo.nardocci@istituto-besta.it
Website: http://www.istituto-besta.com/
International NBIA Patient Registry Clinical Center

- **PKAN** research

San Raffaele Scientific Institute, Milano Dr. Sonia Levi, Email: levi.sonia@hsr.it

- **PKAN** research

Germany

International NBIA Biobank; Dr. Arcangela Iuso; Email: arcangela.iuso@helmholtz-muenchen.de Tel: +49 89 3187-2495

Institute of Human Genetics at the Technical University Munich (TUM) , Dr. Holger Prokisch, Email: prokisch@helmholtz-muenchen.de Tel: +49 89 3187-2890

- **MPAN** research
- **BPAN** mouse model

Helmholtz Zentrum München, Dr. Ana Messias, Email: ana.messias@helmholtz-muenchen.de Tel. +49 89 3187-4706

- **MPAN** research

Klinikum der Universität München
Friedrich-Baur-Institute at the Department of Neurology
Prof. Thomas Klopstock & Dr. Ivan Karin Ziemssenstr. 1a 80336 Munich, Germany Phone: (+49) 089-44005-7421 Email: tircon@med.uni-muenchen.de
Website: http://www.baur-institut.de/
International NBIA Patient Registry Clinical Center

International NBIA Patient Registry; Dr. Thomas Klopstock, Email: thomas.klopstock@med.uni-muenchen.de; Dr. Doriana Buechner, Tel: 089/4400-57421 Email: boriana.buechner@med.uni-muenchen.de

Hungary

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Dr. Peter Acs, MD, PhD Neurologist Rét u. 2., 7623. Pécs, Hungary Phone: (+36) 072-536-000 / 37624 Email: acs.peter@pte.hu
International NBIA Patient Registry Associated Clinical Center

University of Pecs and Markusovszky University Teaching Hospital
Prof. Dr. Bernadette Kalman, MD, PhD, DSc. Neurologist, Head of Molecular Pathology Markusovszky u. 5. 9700. Szombathely Phone(+ 36) 094-515-624 Email: bernadett.kalman@etk.pte.hu; kalman.bernadett@markusovszky.hu
International NBIA Patient Registry Associated Clinical Center

Semmelweis University
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Prof. Dr. Maria Judit Molnar Tömö u. 25-29 1083 Budapest, Hungary Phone: (+36) 01459 1492 E-mail: molnarmj@gmail.com
International NBIA Patient Registry Associated Clinical Center

Spain

Hospital Sant Joan de Déu, Universitat de Barcelona
Servei de Neurologia
Dr. Belen Perez Duenas & Dr. Alejandra Darling Passeig de Sant Joan de Deu 2 08950 Esplugues de Llobregat, Barcelona, Spain Phone: (+34) 0932804000 E-mail: bperez@hsjdbcn.org; adarling@hsjdbcn.org
International NBIA Patient Registry Associated Clinical Center

Universidad Pablo de Olavide
Centro Andaluz De Biología del Desarrollo-CSIC
Grupo de Fisiopatología Celular en la Enfermedad y el DesarrolloLaboratorio 210

Dr. José A. Sánchez Alcázar jasanalc@upo.es

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954978019
954349381

- **PKAN**, experimental treatment on 3 PKAN patients
- **PLAN**, "treatment for PLAN patients coming soon."
- **MPAN**
- **Aceruloplasminemia**

IBiS /HOSPITAL UNIVERSITARIO VIRGEN DEL ROCIO
Dr. Pablo Mir and Dr. Marcos Madruga pmir@us.es and mmadruga@us.es
Avda. Manuel Siurot, s/n.
41013 - Sevilla
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They are neurologists and researchers in very narrow cooperation with the Universidad Pablo de Olavide. This institution is in charge of transferring the knowledge from the laboratory to the patients

Switzerland

CHUV (Cantonal hôpital universitaire Vaud)
Dr. Diana Ballhausen, neuropediatrie département (has three PKAN patients) diana.ballhausen@chuv.ch
Professor Dr. Marie-Claude Addor, CHUV, génétique département marie-claude.addor@chuv.ch

Poland

Instytut Pomnik Centrum Zdrowia Dziecka
Klinika Neurologii i Epileptologii
Dr. Tomasz Kmiec Aleja Dzieci Polskich 20 04730 Warsaw, Poland Phone: (+48) 22 815 7797 Email: t.kmiec@czd.pl
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Czech Republic

Neurologická klinika Všeobecné fakultní nemocnice a 1.lékařské fakulty Univerzity Karlovy
MU Dr. Petr Dušek Ph.D. Kateřinská 30 120 00 Praha 2, Czech Republic Phone: (+42)0 22496 5528 Email: pdusek@gmail.com
International NBIA Patient Registry Associated Clinical Center

Netherlands

University of Groningen Medical Center
Ody Sibon, Ph.D. Email: o.c.m.sibon@umcg.nl

North America

UCSF Benioff Children's Hospital Oakland
Elliott Vichinsky, MD 747 52nd Street Oakland, CA 94609, USA Phone: (+1) 510 612-6202 Email: evichinsky@mail.cho.org
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International NBIA Patient Registry Clinical Center

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St. Jude Children's Research Hospital
Suzanne Jackowski, PhD Infectious Diseases MS 320, Room E8051 St. Jude Children's Research Hospital 262 Danny Thomas Place Memphis, TN 38105-3678
Email: suzanne.jackowski@stjude.org
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Oregon Health & Science University
Susan Hayflick, MD & Penny Hogarth, MD 3181 SW Sam Jackson Park Road Portland, OR 97239, USA Phone: (+1) 503 494 7703 Email: hayflick@ohsu.edu, hogarthp@ohsu.edu
Website: www.nbiacure.org
International NBIA Patient Registry Clinical Center

- **PKAN, Best Practices**, guidelines on recommendations and instructions for taking care of someone with PKAN
- **BPAN-RETT Project**, study to translate basic science discovery gained through laboratory experiments into useful medical practices"
- **PKAN READY**, 5 year study to better understand the natural history of PKAN, how symptoms appear and change over time
- **PLAN READY,** study to better understand the natural history of PLAN, how symptoms appear and change over time
- **NBIA RATING SCALES,** NBIA specific rating scale to measure disease progression
- **MOUSE MODELS**, "knock-out" and "knock-in" are the two main types of transgenic mouse models
  - **PKAN Knock-Out Mice**, ✕
  - **PKAN Knock-In Mice** :Funded by NBIA Disorders Association and developed by OzGene
  - **BPAN**, tissue samples taken June 2016
  - Other NBIA Transgenic Mice
- **NBIA Gene Discovery**
- **Brain Iron Metabolism**

Middle East

Iran

University of Teheran, Department of Molecular Biology and Genetics, Professor Dr. Elahe Elahi, elahe.elahi@gmail.com

University Faculty of Medicine
Dr. Mohammad Rohani
mohammadroohani@gmail.com

Prof. Gholamali Shahidi,
gholamali.shahidi@gmail.com

Asia

China

Peking University First Hospital Beijing
Ye Wu M.D.
Department of Pediatrics

Institute of Biophysics, Chinese Academy of Sciences
Hong Zhang, Ph.D, Prof., Principal Investigator National Laboratory of Biomacromolecules, IBP Mechanism and regulation of autophagy in multicellular organisms, email: hongzhang@sun5.ibp.ac.cn
Tel：010-64848238

- **BPAN** Wdr45 knockout mice

Pharmaceutical Industry

- **ApoPharma Inc,** USA (involved in deferiprone trial)
- **AciesBio**, Slovenia
- **Retrophin, Inc.**, USA

Disease States

- **PKAN** (*pantothenate kinase-associated neurodegeneration*)
- **PLAN** (*PLA2G6-associated neurodegeneration*)
- **MPAN** (*mitochondrial membrane protein-associated neurodegeneration*)
- **FAHN** (*Fatty Acid Hydroxylase-associated Neurodegeneration*)
- **BPAN** (*Beta-propeller Protein-associated Neurodegeneration*)
- **CoPAN** (*COASY Protein-Associated Neurodegeneration*)
- **Woodhouse-Sakati syndrome**
- **Neuroferritinopathy**
- **Aceruloplasminemia**
- **Kufor-Rakeb syndrome**

Communities & Organizations

North America

NBIA Disorders Association, www.nbiadisorders.org

- Events
  - **6th International Symposium on NBIA & Related Disorders**, April 7-8, 2017 at Skamania Lodge , Stevenson, Washington
  - Biennial **Ninth International NBIA Disorders Association Family Conference,** June 1-4, 2017 in Deerfield, IL
- **NBIA Canada**, www.nbiacanada.com

Virtual / International

- **Facebook**
  - **NBIA Families and Supporters** 384 Members
  - **NBIA Disorders Association**
  - **PKAN Families**
  - **BPAN**, 123 Members
  - **NBIA-PKAN, PLAN, NKAN, MPAN** 14 members
  - **NBIA Families,** UK
  - **NBIA Families**, Australia
  - **NBIA Suisse**
  - **ENACH Asociación**
- **RareConnect**, www.rareconnect.org
- **NBIA Alliance**, www.nbiaalliance.org

Europe

- **Post TIRCON** (Treat Iron-Related Childhood-Onset Neurodegeneration)
- **NBIA Suisse** (Swiss), Fatemeh Mollet, Email: Nbiasuisse@gmail.com
- **ENACH Asociacion** (Spanish), www.enach.org
- **AISNAF** (Italian), http://aisnaf.org, *Norton Security thinks this site is a computer threat*
- **Stichting Ijzersterk** (Dutch),* www.stichtingijzersterk.nl
- **AIDNAI** (French), www.aidnai.org
- **Hoffnungsbaum** e.V. (German), http://www.hoffnungsbaum.de

Umbrella Groups for rare disorders

- **NORD** - National Organization for Rare Diseases, https://rarediseases.org
- **Eurordis**
- **Global Genes**
- **Genetic Alliance**