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GENETIQUE ET BIOTHERAPIES DES MALADIES DEGENERATIVES
ET PROLIFERATIVES DU SYSTEME NERVEUX
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Curriculum vitae

Date of birth : 20 July 1953 in Paris , maried, five children, french

Medical Studies

1970-1976: Medical School of Paris-Descartes University
1975: nomination at the "Concours d'Internat in Médecine" (residency) of Paris
1981: Medical thesis, University of Paris V (Paris-Descartes University)
1981: certified on Pediatric board

Medical Fonctions

1977-1981: Residency in Hospitals of Paris (Adult Neurology, Pediatrics, Pediatric Neurology, Genetics et Metabolic diseases, Pediatric ICU)
1982-1985: Chef de Clinique (post residency) in Pediatric Neurology and in Pediatric ICU
1995- : Professor of Pediatrics, Hôpital Saint-Vincent de Paul and Medical School of Paris-Descartes University

Research Functions

1981-1985: Research fellow at Unité 188 of INSERM
1985-1987: Fellowship in the laboratory of Pr. H Moser (Kennedy Institute and Johns Hopkins University, Baltimore, Maryland, USA)
1987-1990 : « In charge of Research » at INSERM
1990-1995 : Director of Research at INSERM
1995- 2005 : Senior scientist as Professor of Pediatrics at INSERM U342 and then INSERM U561
2006- : Director of INSERM research unit UMR745

Teaching at the University Paris-Descartes

1995-2004 : Director of a DEA (Diplome d'Etudes Approfondies) which is a pre-PhD teaching program in Molecular Genetics of Developmental and Oncogenetic Diseases (University of Paris V)
2004- : member of the Ecole Doctorale (Gc2id) board
1995- : Teaching at the Medical School of Medecine of Paris-Descartes University: Genetics, Neurology and Pediatrics

1995- : Teaching in different PhD program trainings

2003-2006 : « Chargé de Mission » at the Ministery of Research and Technology

Member of :Faculty board of Faculty of Medecine, University Paris-Descartes ; Scientific board of « Vaincre les Maladies Lysosomales » Association, Scientific board of Association Française Ataxie de Friedreich

President of the Scientific Board of European Leukodystrophy Foundation

Member of: American Society for Biochemistry and Molecular Biology ; American Society of Gene Therapy ; Society for the Study of Inborn Errors of Metabolism ; European Pediatric Neurology Society ; French Pediatric Society ; French Genetic Society ; French Neuropediatric Society

Reviewer in : Nature Genetics, EMBO Journal, American Journal of Human Genetics, Journal of Clinical Investigation, Annals of Neurology, Human Molecular Genetics, European Journal of Genetics, Journal of Lipid Research, Pediatrics Research, Human Genetics, Journal of Lipid Research, Lancet, Neurology, Brain.

Clinical Activities

- Chief of a Neuropediatrics Department at Hôpital Saint-Vincent de Paul : this department is particularly devoited to patients with leukodystrophies (inherited diseases of white matter), white matter diseases of the neonate, neurofibromatosis type I, and neuro-metabolic diseases.

Main activities of research

At INSERM UMR745 research unit (Hôpital Saint-Vincent de Paul, Paris), I am the director of 22 person team working in the field of :

1/ X-linked adrenoleukodystrophy (ALD) : physiopathology, gene therapy, identification of novel drug targets, identification of modifier genes, hematopoietic and mesenchymal stem cell transplantation, clinical trials

2/ Metachromatic Leukodystrophy Disease (MLD) : gene therapy

3/ Neurofibromatosis type I : physiopathology, identification of modifier genes, identification of novel drug targets

4/ Alzheimer disease : physiopathology, identification of novel drug targets

MAJOR PUBLICATIONS

Aubourg P., Sack G.H., Meyers D.A., Lease J.J., and Moser H.W. Linkage of adrenoleucodystrophy to a polymorphic DNA probe. **Ann. Neurol.** 21:349-352, 1987

Aubourg P., Sack G.H., Moser H.W. Frequent alterations of visual pigment genes in adreneulokodystrophy. **Am. J. Hum. Genet.** 42:408-413, 1988

Aubourg P., Blanche S., Jambaqué I., Rocchiccioli F., Naud-Saudreau C., Kalifa G., Rolland M.O., Debré M., Chaussain J.L., Griscelli C., Fisher A., Bougnères P.F. Reversal of early neurologic and neuroradiologic manifestations of X-linked adrenoleukodystrophy by bone marrow transplantation. **N. Engl. J. Med.**, 322:1860-1866, 1990.

Mosser J, Douar AM, Sarde CO, Kioschis P, Feil R, Moser H, Poustka AM, Mandel JL, Aubourg P. Putative X-linked adrenoleukodystrophy gene shares unexpected homology with ABC transporters. **Nature** 361: 726-730, 1993.

Aubourg P, Adamsbaum C, Lavallard-Rousseau, F. Rocchiccioli, N. Cartier, I. Jambaqué, C. Jakobezak, A. Lemaitre, F. Boureau, C. Wolf, P-F. Bougnères. A two-year trial of oleic ans erucic acids (Lorenzo' oil) as treatment for adrenomyeloneuropathy. **N. Engl. J. Med.** 329: 745-753, 1993.

Fanen P, Guidoux S, Sarde C, Mandel J-L, Goossens M, Aubourg P. Identification of mutations in the putative ATP-binding domain of the adrenoleukodystrophy gene. **J. Clin. Invest** 94: 516-520, 1994.

Cartier N, Lopez J, Moullier P, Rocchiccioli F, Rolland M-O, Jorge P, Mandel J-L, Bougnères P-F, Danos O, Aubourg P. Retroviral-mediated gene transfer corrects very-long-chain fatty acid metabolism in adrenoleukodystrophy fibroblasts. **Proc. Natl. Acad. Sci. USA.** 92: 1674-1678, 1995.

Feigenbaum V, Lombard-Platet G, Guidoux S, Sarde C, Mandel J-L, Aubourg P. Mutational and protein analysis of patients and heterozygous women with X-linked adrenoleukodystrophy. **Am. J. Hum. Genet**.

58: 1135-1144, 1996.
Fouquet F, Min Zhou J-M, Ralston E, Murray K, Troalen F, Magal E, Robain O, Dubois-Dalcq M, Aubourg P. Expression of the Adrenoleukodystrophy protein in the human and mouse central nervous system**. Neurobiology. Dis**. 3: 271-285 1997.
Doerflinger N, Micléa J.M., Lopez J., Chomienne C., Bougnères P., Aubourg P., Cartier N. Retroviral transfer and long-term expression of the adrenoleukopdystrophy gene in human CD34+ cells. **Hum. Gene Ther**. 9: 1025-1036, 1998.
Liu L, Janvier K, Berteaux-Lecellier V, Cartier N, Benarous R, Aubourg P. Homo- and heterodimerization of peroxisomal ABC half transporters. **J. Biol. Chem**. 274: 32738-32743, 1999.
Dubois-Dalcq M, Feigenbaum V, Aubourg P. The neurobiology of X-linked adrenoleukodystrophy (ALD), a demyelinating peroxisomal disorder. **Trends Neurosci.** 22: 4-12, 1999.
Shapiro E, Krivit W, Lockman L, Jambaqué I, Peters C, Cowan M, Harris R, Bordigoni P, Loes D, Moser H, Fischer A, Aubourg P. Long-term beneficial effect of bone marrow transplantation for childhood onset cerebral X-linked adrenoleukodystrophy. **Lancet**, 356: 713-718, 2000
Benhamida S, Pflumio F, Dubart-Kupperschmitt A, Zhao-Emonet J-C, Cavazzana-Calvo M, Rocchiccioli F, Fichelson S, Aubourg P, Charneau P, Cartier N. Transduction of mobilized peripheral blood CD34+ cells with HIV vector encoding adrenoleukodystrophy gene mediates long-term engraftment of NOD/SCID mice. **Mol. Ther.** 7 : 317-324, 2003.
Asheuer M, Pflumio F, Benhamida S, Fouquet F, Dubart-Kupperschmitt, Aubourg P, Cartier N. Genetically modified human CD34+ can differentiate into brain microglia and express therapeutic protein. **Proc. Natl. Acad. Sci. USA**. 101 : 3557-3562, 2004.
Moreira M-C. Klur S, Watanabe M, Nemeth AH, Le Ber I, Moniz J-C, Tranchant C, Aubourg P, M Tazir, L Schöls, M Pandolfo, J B Schulz, J Pouget, P Calvas, M Shizuka-Ikeda, M Shoji, M Tanaka, L Izatt, C E Shaw, A M'Zahem, E Dunne, P Bomont, T Benhassine, N Bouslam, G Stevanin, A Brice, J Guimarães, P Mendonça, C Barbot, P Coutinho, J Sequeiros, A Dürr, J -M Warter & M Koenig. Senataxin, the ortholog of a yeast RNA helicase, is mutant in ataxia-ocular apraxia 2. **Nature Genet**. 36: 225-227, 2004.
Peters C, Charnasi LR, Tan Y, Ziegler R, Shapiro E, DeForz T, Grewal SS, Orchad PJ, Abel SL, Goldman AI, Ramsay NKC, Dusenbery KE, Loe DJ, Lockman LA, Kato S, Aubourg P, Moser HW, Krivit W. Cerebral X-linked adrenoleukodystrophy: the international hematopoietic cell transplantation experience from 1982 to 1999. **Blood** 104:881-888, 2004.
Asheuer M, Bièche I, Laurendeau I, Moser A, Hainque B, Vidaud M, Aubourg P. Molecular and biochemical mechanisms for distinct neurological phenotypes in X-linked adrenoleukodystrophy. **Hum. Mol Genet**, 14:1293-303, 2005.
Sevin C, Benraiss A, Van Dam D, Bonnin D, Nagels G, Verot L, Laurendeau I, Vidaud M, Gieselmann V, Vanier M, De Deyn PP, Aubourg P, Cartier N. Intracerebral Adeno-Associated Virus-mediated gene transfer in rapidly progressive forms of metachromatic leukodystrophy. **Hum. Mol. Genet**. 15: 53-64, 2006.
Sevin C, Verot L, Benraiss A, Van Dam D, Bonnin D, Nagels G, Fouquet F, Gieselmann V, Vanier MT, De Deyn PP, Aubourg P, Cartier N. Partial cure of established disease in an animal model of metachromatic leukodystrophy after intracerebral adeno-associated virus-mediated gene transfer **Gene Ther**. 2007, 14 (5) :405-414.
Sevin C, Aubourg P, Cartier N. Enzyme, cell and gene-based therapies for metachromatic leukodystrophy. **J Inherit Metab Dis**. 2007 Apr;30(2):175-83.
Aubourg P. Axons need glial peroxisomes. **Nat Genet.** 2007 Aug;39(8):936-8.