

Name: ABRAMOWICZ
First name: Marc
Date of birth: December 27, 1962
Nationality: Belgian

Education:
MD - 1986, Free University of Brussels (ULB)
PhD - 1992, Free University of Brussels (ULB)

Professional positions:
Chef de Clinique (Head, Clinic of Medical Genetics), Hopital Erasme - ULB
Associate Professor, School of Medicine, ULB

Research interest:
Medical genetics: chromosomal mapping, and gene identification, of rare genetic diseases like primary microcephaly and congenital hypothyroidism.

Main publications:

- Van Bogaert P, Azizieh R, Desir J, Aeby A, De Meirleir L, Laes JF, Christiaens F, **Abramowicz MJ** (2007) Mutation of a potassium channel-related gene in progressive myoclonic epilepsy. *Ann Neurol.* 2007 Jun;61(6):579-86
- Desir J, Moya G, Reish O, Van Regemorter N, Deconinck H, David KL, Meire FM, **Abramowicz M** (2007) Borate transporter SLC4A11 mutations cause both Harboyan syndrome and non-syndromic corneal endothelial dystrophy. *J Med Genet.* 2007 May;44(5):322-6.
- Thomée C, Schubert SW, Parma J, Lê PQ, Hashemolhosseini S, Wegner M, **Abramowicz MJ** (2005) GCMB mutation in familial isolated hypoparathyroidism with residual secretion of parathyroid hormone. *J Clin Endocrinol Metab* 90: 2487-92
- Jamieson CR, Fryns JP, Jacobs J, Matthijs G, **Abramowicz MJ** (2000) Primary autosomal recessive microcephaly: MCPH5 maps to 1q25-q32. *Am J Hum Genet* 67: 1575-7.
- **Abramowicz MJ**, Duprez L, Parma J, Vassart G, Heinrichs C (1997) Familial congenital hypothyroidism due to inactivating mutation of the thyrotropin receptor causing profound hypoplasia of the thyroid gland. *J Clin Invest* 99: 3018-3024.