

## Jean-Louis Mandel

Chaire Génétique humaine (2003-2016)

# BIBLIOGRAPHIE

## PRINCIPALES PUBLICATIONS

- Oberlé I., Rousseau F., Heitz D., Kretz C., Devys D., Hanauer A., Boué J., Bertheas M. F. et Mandel J. L., « Instability of a 550bp DNA segment and abnormal methylation in Fragile X syndrome », *Science*, vol. 252, n0 5009, 1991, p. 1097-1102.
- Rousseau F., Heitz D., Biancalana V., Blumenfeld S., Kretz C., Boué J., Tommerup N., Van Der Hagen C., DeLozier Blanchet C., Croquette M. F., Gilgenkrantz S., Jalbert P., Voelckel M. A., Oberlé I. et Mandel J.-L., « Efficient and reliable direct diagnosis of the Fragile X mental retardation syndrome », *New England Journal of Medicine*, vol. 325, 1991, p. 1673-1681.
- Ben Hamida C., Doerflinger N., Belal S., Linder C., Reutenauer L., Dib C., Gyapay G., Vignal A., Le Paslier D., Cohen D., Pandolfo M., Mokini V., Novelli G., Hentati F., Ben Hamida M., Mandel J. L. et Koenig M., « Localization of Friedreich ataxia phenotype with selective vitamin E deficiency to chromosome 8q by homozygosity mapping », *Nature Genetics*, vol. 5, 1993, p. 195-200.
- Devys D., Lutz Y., Rouyer N., Bellocq J. P. et Mandel J. L., « FMR-1 protein is cytoplasmic, is most abundant in neurons and appears normal in carriers of fragile X premutation », *Nature Genetics*, vol. 4, 1993, p. 335-340.
- Imbert G., Kretz C., Johnson K. et Mandel J. L., « Origin of the expansion mutation in myotonic dystrophy », *Nature Genetics*, vol. 4, 1993, p. 72-76.
- Mosser J., Douar A. M., Sarde C. O., Kioschis P., Feil R., Moser H., Poustka A. M., Mandel J. L. et Aubourg P., « Putative X-linked adrenoleukodystrophy gene shares unexpected homology with ABC transporters », *Nature*, vol. 361, 1993, p. 726-730.
- Mosser J., Lutz Y., Stoeckel M. E., Sarde C. O., Kretz C., Douar A. M., Aubourg P. et Mandel J. L., « The gene responsible for adrenoleukodystrophy encodes a peroxisomal membrane protein », *Human Mol. Genet.*, vol. 3, 1994, p. 265-271.

- Ouahchi K., Arita M., Kayden H., Hentati F., Ben Hamida M., Sokol R., Arai H., Inoue K., Mandel J. L. et Koenig M., « Ataxia with isolated vitamin E deficiency is caused by mutations in the -tocopherol transfer protein », *Nature Genet*, vol. 9, 1995, p. 141-145.
- Trottier Y., Lutz Y., Stevanin G., Imbert G., Devys D., Cancel G., Weber C., Saudou F., David G., Tora L., Agid Y., Brice A. et Mandel J. L., « Polyglutamine expansion as a common pathological epitope detected in Huntington's disease and in four dominant cerebellar ataxias », *Nature*, vol. 378, 1995, p. 403-406.
- Campuzano V., Montermini L., Moltò M. D., Pianese L., Cossée M., Cavalcanti F., Monros E., Rodius F., Duclos F., Monticelli A., Zara F., Cañizares J., Koutnikova H., Bidichandani S., Gellera C., Brice A., Trouillas P., De Michele G., Filla A., De Frutos R., Palau F., Patel P. I., Di Donato S., Mandel J. L., Coccozza S., Koenig M. et Pandolfo M., « Friedreich Ataxia. The first autosomal recessive disease caused by an intronic GAA triplet repeat expansion », *Science*, vol. 271, 1996, p. 1423-1427.
- Dürr A., Cossée M., Agid Y., Campuzano V., Mignard C., Penet C., Mandel J. L., Brice A. et Koenig M., « Clinical and genetic abnormalities in patients with Friedreich's Ataxia », *New Engl. J. Med.*, vol. 335, 1996, p. 1169-1175.
- Imbert G., Saudou F., Yvert G., Devys D., Trottier Y., Garnier J. M., Weber C., Mandel J. L., Cancel G., Abbas N., Dürr A., Didierjean O., Stevanin G., Agid Y. et Brice A., « Cloning of the gene for spinocerebellar ataxia 2 reveals a locus with high sensitivity to expanded CAG/glutamine repeats », *Nature Genet*, vol. 14, 1996, p. 285-291.
- Laporte J., Hu L. J., Kretz C., Mandel J. L., Kioschis P., Coy J. F., Klauck S. M., Poustka A. et Dahl N., « A gene mutated in X-linked myotubular myopathy defines a new putative tyrosine phosphatase family conserved in yeast », *Nature Genet*, vol. 13, 1996, p. 175-182.
- Sittler A., Devys D., Weber C. et Mandel J. L., « Alternative splicing of exon 14 determines nuclear or cytoplasmic localisation of FMR1 protein isoforms », *Hum. Mol. Genet.*, vol. 5, 1996, p. 95-102.
- Trivier E., De Cesare D., Jacquot S., Pannetier S., Zackai E., Young I., Mandel J. L., Sassone-Corsi P. et Hanauer A., « Mutations in the Kinase Rsk-2 associated with Coffin-Lowry syndrome », *Nature*, vol. 384, 1996, p. 567-570.
- David G., Abbas N., Stevanin G., Dürr A., Yvert G., Cancel G., Weber C., Imbert G., Saudou F., Antoniou E., Drabkin H., Gemmill R., Giunti P., Benomar A., Wood N., Ruberg M., Agid Y., Mandel J. L. et Brice A., « Cloning of the SCA7 gene reveals a highly unstable CAG repeat expansion », *Nature Genet.*, vol. 17, 1997, p. 65-70.
- Paulson H. L., Perez M. K., Trottier Y., Trojanowski J. Q., Subramony S. H., Das S. S., Vig P., Mandel J. L., Fischbeck K. H., Pittman R. N., « Intranuclear inclusions of expanded polyglutamine protein in spinocerebellar ataxia type 3 », *Neuron*, vol. 19, 1997, p. 333-344.
- Cossée M., Schmitt M., Campuzano V., Reutenauer L., Moutou C., Mandel J. L. et Koenig M., « Evolution of the Friedreich's ataxia trinucleotide repeat expansion: founder effect and premutations », *Proc. Natl. Acad. Sci.*, vol. 94, 1997, p. 7452-7457.
- Lunkes A. et Mandel J. L., « A cellular model that recapitulates major pathogenic steps of Huntington's disease », *Hum. Mol. Genet.*, vol. 7, 1998, p. 1355-1361.

- Mérienne K., Jacquot S., Pannetier S., Zeniou M., Bankier A., Gecz J., Mandel J. L., Mulley J., Sassone-Corsi P. et Hanauer A., « A missense mutation in RPS6KA3 (RSK2) responsible for non-specific mental retardation », *Nature Genet.*, vol. 22, 1999, p. 13-14.
- Blondeau F., Laporte J., Bodin S., Superti-Furga G., Payrastre B. et Mandel J. L., « Myotubularin, a phosphatase deficient in myotubular myopathy, acts on phosphatidylinositol 3-kinase and phosphatidylinositol 3-phosphate pathway », *Human Mol. Genet.*, vol. 9, 2000, p. 2223-2229.
- Schaeffer C., Bardoni B., Mandel J. L., Ehresmann B., Ehresmann C. et Moine H., « The fragile X mental retardation protein binds specifically to its mRNA via a purine quartet motif », *EMBO J.*, vol. 20, 2001, p. 4803-4813.
- Chelly J. et Mandel J. L., « Monogenic causes of X-linked mental retardation », *Nature Reviews Genet.*, vol. 2, 2001, p. 669-680.
- Laporte J., Blondeau F., Buj-Bello A. et Mandel J. L., « The myotubularin family: from genetic disease to phosphoinositide metabolism », *Trends in Genet.*, vol. 17, 2001, p. 221-228.
- Schenck A., Bardoni B., Moro A., Bagni C. et Mandel J. L., « A highly conserved protein family interacting with the fragile X mental retardation protein and displaying selective interactions with the related protein FXR1P and FXR2P », *Proc. Natl. Acad. Sci.*, vol. 98, 2001, p. 8844-8849.
- Buj-Bello A., Laugel V., Messaddeq N., Zahreddine H., Laporte J., Pellissier J. F. et Mandel J. L., « The lipid phosphatase myotubularin is essential for skeletal muscle maintenance but not for myogenesis in mice », *Proc. Natl. Acad. Sci. USA*, vol. 99, 2002, p.15060-15065.
- Lunkes A., Lindenberg K. S., Ben-Haiem L., Weber C., Devys D., Landwehrmeyer G. B., Mandel J. L. et Trottier Y., « Proteases acting on mutant huntingtin generate leaved products that differentially build up cytoplasmic and nuclear inclusions », *Mol. Cell.*, vol. 10, 2002, p. 259-269.
- Schenck A., Bardoni B., Langmann C., Harden N., Mandel J. L. et Giangrande A., « CYFIP/Sra-1 Controls Neuronal Connectivity in Drosophila and Links the Rac1 GTPase Pathway to the Fragile X Protein », *Neuron*, vol. 38, 2003, p. 887-898.