

Publications scientifiques qui font date pour la STB et la LAM

1993-1997 Identification des gènes qui provoquent la sclérose tubéreuse de Bourneville

- *European Chromosome 16 Tuberous Sclerosis Consortium. Identification and characterization of the tuberous sclerosis gene on chromosome 16.* Cell. 1993 Dec 31;75(7):1305-15. PubMed PMID: 8269512.
- van Slechtenhorst M, de Hoogt R, Hermans C, Nellist M, Janssen B, Verhoef S, Lindhout D, van den Ouweland A, Halley D, Young J, Burley M, Jeremiah S, Woodward K, Nahmias J, Fox M, Ekong R, Osborne J, Wolfe J, Povey S, Snell RG, Cheadle JP, Jones AC, Tachataki M, Ravine D, Sampson JR, Reeve MP, Richardson P, Wilmer F, Munro C, Hawkins TL, Sepp T, Ali JB, Ward S, Green AJ, Yates JR, Kwiatkowska J, Henske EP, Short MP, Haines JH, Jozwiak S, Kwiatkowski DJ. *Identification of the tuberous sclerosis gene TSC1 on chromosome 9q34.* Science. 1997 Aug 8;277(5327):805-8. PubMed PMID: 9242607.

1999 Découverte du rôle des gènes régulant le cycle cellulaire

- Ito N, Rubin GM. *gigas, a Drosophila homolog of tuberous sclerosis gene product-2, regulates the cell cycle.* Cell. 1999 Feb 19;96(4):529-39. Erratum in: Cell 2001 May 4;105(3):415. PubMed PMID: 10052455.

1998-2002 Découverte que des mutations du gène TSC2 provoquent la LAM

- Smolarek TA, Wessner LL, McCormack FX, Mylet JC, Menon AG, Henske EP. *Evidence that lymphangiomyomatosis is caused by TSC2 mutations: chromosome 16p13 loss of heterozygosity in angiomyolipomas and lymph nodes from women with lymphangiomyomatosis.* Am J Hum Genet. 1998 Apr;62(4):810-5. PubMed PMID: 9529362; PubMed Central PMCID: PMC1377043.
- Carsillo T, Astrinidis A, Henske EP. *Mutations in the tuberous sclerosis complex gene TSC2 are a cause of sporadic pulmonary lymphangiomyomatosis.* Proc Natl Acad Sci U S A. 2000 May 23;97(11):6085-90. PubMed PMID: 10823953; PubMed Central PMCID: PMC18562. Discovery that mTOR is activated in LAM tissues
- Goncharova EA, Goncharov DA, Eszterhas A, Hunter DS, Glassberg MK, Yeung RS, Walker CL, Noonan D, Kwiatkowski DJ, Chou MM, Panettieri RA Jr, Krymskaya VP. *Tuberin regulates p70 S6 kinase activation and ribosomal protein S6 phosphorylation. A role for the TSC2 tumor suppressor gene in pulmonary lymphangiomyomatosis (LAM).* J Biol Chem. 2002 Aug 23;277(34):30958-67. Epub 2002 Jun 3. PubMed PMID: 12045200.

2005 Démonstration que les inhibiteurs mTOR ont un avenir prometteur pour la TSC

- Kenerson H, Dundon TA, Yeung RS. *Effects of rapamycin in the Eker rat model of tuberous sclerosis complex.* Pediatr Res. 2005 Jan;57(1):67-75. Epub 2004 Nov 19. PubMed PMID: 15557109.

2011 Identification du sirolimus comme étant une thérapie effective pour la LAM

- McCormack FX, Inoue Y, Moss J, Singer LG, Strange C, Nakata K, Barker AF, Chapman JT, Brantly ML, Stocks JM, Brown KK, Lynch JP 3rd, Goldberg HJ, Young LR, Kinder BW, Downey GP, Sullivan EJ, Colby TV, McKay RT, Cohen MM, Korbee L, Taveira-DaSilva AM, Lee HS, Krischer JP, Trapnell BC; National Institutes of Health Rare Lung Diseases Consortium; *MILES Trial Group*. *Efficacy and safety of sirolimus in lymphangiomyomatosis*. N Engl J Med. 2011 Apr 28;364(17):1595-606. doi: 10.1056/NEJMoa1100391. Epub 2011 Mar 16. PubMed PMID: 21410393; PubMed Central PMCID: PMC3118601.