

## COMPLETE LIST OF PUBLICATIONS

### Original articles :

- 1- Cayuela J.M, **Gardie B**, Sigaux F. "Disruption of the Multiple Tumor Suppressor Gene MTS1/p16INK4a /CDKN2 by illegitimate V(D)J recombinase activity in T-cell Acute Lymphoblastic Leukemias". *Blood*, 1997, nov1, vol90, 3720-3726. (IF: 17,5).
- 2- **Gardie B**, Cayuela J.M, Martini S, Sigaux F. "Genomic alteration of the p19ARF encoding exons in T-cell Acute Lymphoblastic Leukemia ". *Blood*, 1998, feb1, 91,1016-1020. (IF: 17,5).
- 3- and Lagresle C<sup>1</sup>, **Gardie B**<sup>1</sup>, Fasseu M, Vieville J.C, Pla M, Sigaux F and Boris J.C. "Transgenic expression of the p16INK4a cyclin-dependant kinase inhibitor arrest CD4-CD8-thymocytes differentiation by inducing pre-TCR/CD3 mediated apoptosis." <sup>1</sup>Equal contribution to the work. *Journal of Immunology*, 2002, Mar 1, 168(5):2325-31. (IF: 4,8).
- 4- Olsen O, **Gardie B**, Yaswen P and Stampfer M. "Raf-induced growth arrest in human mammary epithelial cells is p16INK4a independant and is overcome in immortal cells during conversion". *Oncogene*, 2002 ,sept 12, 21(41) :6328-39. (IF: 7,9).
- 5- Al-Sheikh M, Mazurier E, **Gardie B**, Casadevall N, Galactéros F, Goossens M, Wajcman H, Préhu C, Ugo V. «A study of 36 unrelated cases with pure erythrocytosis revealed three new mutations in the erythropoietin receptor gene». *Haematologica*, 2008, Jul;93(7):1072-5. (IF: 7).
- 6- Ladroue C, Carcenac R, Leporrier M, Gad S, Le Hello C, Galateau-Salle F, Feunteun J, Pouyssegur J, Richard S, **Gardie B**. "A novel mutation in the PHD2 gene associated with congenital polycythemia and recurrent paraganglioma: evidences for a potential tumor-suppressor effect". *New England Journal of Medicine*, 2008, Dec 18, 359(25):2685-92. (IF: 74,6).
- 7- Vahteristo P, Koski TA, Näätsaari L, Kiuru M, Karhu A, Herva R, Sallinen SL, Vierimaa O, Björck E, Richard S, **Gardie B**, Bessis D, Van Glabeke E, Blanco I, Houlston R, Senter L, Hietala M, Aittomäki K, Aaltonen LA, Launonen V, Lehtonen R. "No evidence for a genetic modifier for renal cell cancer risk in HLRCC syndrome". *Familial Cancer*, 2010, Jun, 9(2):245-51. (IF: 1,7).
- 8- Marque M, **Gardie B**, Bressac de Paillerets B, Rustin P, Guillot B, Richard S, Bessis D. "Novel FH mutation in a patient with cutaneous leiomyomatosis associated with cutis verticis gyrata, eruptive collagenoma and Charcot-Marie-Tooth's disease". *British Journal of Dermatology*, 2010, Dec;163(6):1337-9. (FI:7).
- 9- **Gardie B.**, A. Remenieras, D. Kattygnarath, J. Bombled, S. Lefevre, V. Perrier-Trudova, P. Rustin, M. Barrois, A. Slama, M.F. Avril, D. Bessis, O. Caron, F. Caux, P. Collignon, I. Coupier, C. Cremin, H. Dollfus, C. Dugast, B. Escudier, L. Faivre, M. Field, B. Gilbert-Dussardier, N. Janin, Y. Leport, D. Leroux, D. Lipsker, F. Malthieu, B. McGillivray, C. Maugard, A. Mejean, I. Mortemousque, G. Plessis, B. Poppe, C. Pruvost-Balland, S. Rooker, J. Roume, N. Soufir, M. Steinraths, M.H. Tan, C. Theodore, L. Thomas, P. Vabres, E. Van Glabeke, J.B. Meric, V. Verkarre, G. Lenoir, V. Joulin, S. Deveaux, V. Cusin, J. Feunteun, B.T. Teh, B. Bressac-de Paillerets, and S. Richard. "Novel FH mutations in families with hereditary leiomyomatosis and renal cell cancer (HLRCC) and patients with isolated type 2 papillary renal cell carcinoma". *Journal of Medical Genetics*, 2011, Apr, 48(4):226-234. (IF: 4,9).
- 10- Ooi A, Wong JC, Petillo D, Roossien D, Perrier-Trudova V, Whitten D, Min BW, Tan MH, Zhang Z, Yang XJ, Zhou M, **Gardie B**, Molinie V, Richard S, Tan PH, Teh BT, Furge KA. An antioxidant response phenotype shared between hereditary and sporadic type 2 papillary renal cell carcinoma. *Cancer Cell*, 2011, 20(4):511-523. (IF: 26,6).
- 11- Bertolotto, C., F. Lesueur, S. Giuliano, T. Strub, M. de Lichy, K. Bille, P. Dessen, B. d'Hayer, H. Mohamdi, A. Remenieras, E. Maubec, A. de la Fouchardiere, V. Molinie, P. Vabres, S. Dalle, N. Poulalhon, T. Martin-Denavit, L. Thomas, P. Andry-Benzaquen, N. Dupin, F. Boitier, A. Rossi, J.L. Perrot, B. Labeille, C. Robert, B. Escudier, O. Caron, L. Brugieres, S. Saule, **B. Gardie**, S.

Gad, S. Richard, J. Couturier, B.T. Teh, P. Ghiorzo, L. Pastorino, S. Puig, C. Badenas, H. Olsson, C. Ingvar, E. Rouleau, R. Lidereau, P. Bahadoran, P. Vielh, E. Corda, H. Blanche, D. Zelenika, P. Galan, F. Aubin, B. Bachollet, C. Becuwe, P. Berthet, Y.J. Bignon, V. Bonadona, J.L. Bonafe, M.N. Bonnet-Dupeyron, F. Cambazard, J. Chevrant-Breton, I. Coupier, S. Dalac, L. Demange, M. d'Incan, C. Dugast, L. Faivre, L. Vincent-Fetita, M. Gauthier-Villars, B. Gilbert, F. Grange, J.J. Grob, P. Humbert, N. Janin, P. Joly, D. Kerob, C. Lasset, D. Leroux, J. Levang, J.M. Limacher, C. Livideanu, M. Longy, A. Lortholary, D. Stoppa-Lyonnet, S. Mansard, L. Mansuy, K. Marrou, C. Mateus, C. Maugard, N. Meyer, C. Nogues, P. Souteyrand, L. Venat-Bouvet, H. Zattara, V. Chaudru, G.M. Lenoir, M. Lathrop, I. Davidson, M.F. Avril, F. Demenais, R. Ballotti, B. Bressac-de Paillerets. "A SUMOylation-defective MITF germline mutation predisposes to melanoma and renal carcinoma". *Nature*, 2011, Oct 19, 480:94-8. (IF: 42,7).

**12-** Ladroue C, Hoogewijs D, Gad S, Carcenac R, Storti F, Barrois M, Gimenez-Roqueplo AP, Leparrier M, Casadevall N, Hermine O, Kiladjian JJ, Baruchel A, Fakhoury F, Bressac-de Paillerets B, Feunteun J, Mazure N, Pouyssegur J, Wenger RH, Richard S, **Gardie B**. "Distinct deregulation of the hypoxia inducible factor by PHD2 mutants identified in germline DNA of patients with polycythemia". *Haematologica*. 2012;97(1):9-14 (IF: 7).

**13-** Cartron PF, Nadaradjane A, Lepape F, Lalier L, **Gardie B**, Vallette FM. "Identification of TET1 Partners That Control Its DNA-Demethylating Function". *Genes Cancer*. 2013, May, 4(5-6):235-41. (IF: 2,4).

**14-** Couvé S, Ladroue C, Laine E, Mahtouk K, Guégan J, Gad S, Le Jeune H, Le Gentil M, Nuel G, Kim W, Lecomte B, Pagès JC, Collin C, Lasne F, Benusiglio P, Bressac-de Paillerets B, Feunteun J, Lazar V, Gimenez-Roqueplo, Mazure N, Dessen P, Tchertanov L, Mole D, Kaelin W, Ratcliffe P, Richard S, **Gardie B**. "Genetic evidence of a precisely tuned dysregulation in the hypoxia signaling pathway during oncogenesis". *Cancer Research*, 2014, Nov 15, 74(22):6554-64. (IF: 9,7).

**15-** Messai Y, Noman MZ, Hasmim M, Janji B, Tittrelli A, Boutet M, Baud V, Viry E, Billot K, Nanbakhsh A, Ben Safta T, Richon C, Ferlicot F, Donnadiou E, Couve S, **Gardie B**, Orlanducci F, Albiges L, Thiery J, Olive D, Escudier E, Chouaib S. "ITPR1 protects renal cancer cells against natural killer cells by inducing autophagy". *Cancer Research*, 2014, Dec 1, 74(23):6820-32. (IF: 9,7).

**16-** Perrier-Trudova V, Wong Huimin B, Kongpetch S, Huang D, g Rong Lui C, Ong P, Le Formal A, Poon SL, Sie EY, Myint SS, Gad S, **Gardie B**, Couvé S, Foong YM, Choudhury Y, Poh J, Ong CK, Toh CK, Ooi A, Richard S, Tan MH, Teh BT. Fumarate Hydratase-deficient Cell Line NCCFH1 as a New In Vitro Model of Hereditary Papillary Renal Cell Carcinoma Type 2. *Anticancer Research*. 2015, Dec, 35(12):6639-53. (IF: 1,9).

**17-** Muller M, Ferlicot S, Guillaud-Bataille M, Le Teuff G, Genestie C, Deveaux S, Slama A, Poulalhon N, Escudier B, Albiges L, Soufir N, Avril MF, **Gardie B**, Saldana C, Allory Y, Gimenez-Roqueplo AP, Bressac-de Paillerets B, Richard S, Benusiglio P. "Reassessing the clinical spectrum associated with Hereditary Leiomyomatosis and Renal Cell Carcinoma syndrome in French FH mutation carriers". *Clinical Genetics*. 2017 Dec;92(6):606-615. (IF: 3,6).

**18-** Girodon F, Airaud F, Garrec C, Bézieau S, **Gardie B**. "Gene panel sequencing in idiopathic erythrocytosis. *Haematologica*. 2017 Jan;102(1):e30. (comment). (FI:7).

**19-** Catherwood MA, Graham A, Cuthbert RJG, Garrec C, **Gardie B**, Girodon F, Laird S, Cross NCP, McMullin MF. Absence of CALR Mutations in Idiopathic Erythrocytosis Patients with Low Serum Erythropoietin Levels. *Acta Haematol*. 2018;139(4):217-219. (IF: 1,2).

**20-** Gattoliat JC, Couvé S, Meurice G, Orear C, Chiquet M, Ferlicot S, Verkarre V, Vasiliu V, Molinié V, Méjean A, Dessen P, Giraud S, Bressac-De-Paillerets B, **Gardie B**, Teh BT, Richard S, Gad S. Integrative Analysis of deregulated microRNAs and mRNAs in multiple recurrent synchronized renal tumors of von Hippel-Lindau patients. *Int. J. Oncol*. 2018 Oct;53(4):1455-1468. (IF: 3,9).

- 21-** Lenglet M, Robriquet F, Schwarz K, Camps C, Couturier A, Hoogewijs D, Buffet A, Knight SJL, Gad S, Couvé S, Chesnel F, Pacault M, Lindenbaum P, Job S, Dumont S, Besnard T, Cornec M, Dreau H, Pentony M, Kvikstad E, Deveaux S, Burnichon N, Ferlicot S, Vilaine M, Mazzella JM, Airaud F, Garrec C, Heidet L, Irtan S, Mantadakis E, Bouchireb K, Debatin KM, Redon R, Bezieau S, Bressac-de Paillerets B, Teh BT, Girodon F, Randi ML, Putti MC, Bours V, Van Wijk R, Göthert JR, Kattamis A, Janin N, Bento C, Taylor JC, Arlot-Bonnemains Y, Richard S, Gimenez-Roqueplo AP, Cario H, **Gardie B**. - Identification of a new VHL exon and complex splicing alterations in familial erythrocytosis or von Hippel-Lindau disease. *Blood*. 2018 Aug 2;132(5):469-483. Plenary paper. (IF: 17,5).
- 22-** Burette B, Bourgeois V, Buriller C, Aral B, Airaud F, **Gardie B**, Girodon F. High HFE mutation incidence in idiopathic erythrocytosis. *Br J Haematol*. 2018 Nov 8. (IF: 5,5).
- 23-** Besnard T, Sloboda N, Goldenberg A, Küry S, Cogné B, Breheret F, Trochu E, Conrad S, Vincent M, Deb W, Balguerie X, Barbarot S, Baujat G, Ben-Omran T, Bursztejn AC, Carmignac V, Datta AN, Delignières A, Faivre L, **Gardie B**, Guéant JL, Kuentz P, Lenglet M, Nassogne MC, Ramaekers V, Schnur RE, Si Y, Torti E, Thevenon J, Vabres P, Van Maldergem L, Wand D, Wiedemann A, Cariou B, Redon R, Lamazière A, Bézieau S, Feillet F, Isidor B. Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. *Genet Med*. 2019 Feb 6. (IF: 8, 9).
- 24-** Buffet A, Calsina B\*, Flores SK\*, S Giraud\*, Lenglet M\*, Romanet, Deflorenne E, Aller J, Bourdeau I, Bressac-de Paillerets B, Calatayud M, Dehais, de Mones E, AElenkova A, P Herman P, Kamenicky P, Lejeune S, JL Sadoul JL, Barlier A, Richard S, J Favier J, N Burnichon N, **Gardie B**, Dahia PLM, Robled M, Gimenez-Roqueplo AP. Germline mutations in the new E1' cryptic exon of the *VHL* gene in patients with tumors of von Hippel-Lindau disease spectrum or with paraganglioma. *Journal of Medical Genetics*. 2020 Jan 29. (IF: 4,9).
- 25-** Bonnin A, Gardie B, Girodon F, Airaud F, Garrec C, Bézieau S, Vignon G, Mottaz P, Labrousse J, Lellouche F. A new case of rare erythrocytosis due to *EGLN1* mutation with review of the literature. *Rev Med Interne*. 2020 Mar;41(3):196-199.
- 26-** Fabbri L, Dufies M, Lacas-Gervais S, **Gardie B**, Gad S, Parola J, Nottet N, Meyenberg Cunha-de Padua M, Contenti J, Borchiellini D, Ferrero JM, Rioux Leclercq N, Ambrosetti D, Mograbi B, Richard S, Viotti J, Chamorey E, Sadaghianloo N, Rouleau M, Craigen WJ, Mari B, Clavel S, Pagès G, Pouysségur J, Bost F and Mazure NM. Identification of a new aggressive axis driven by ciliogenesis and absence of VDAC1-ΔC in clear cell Renal Cell Carcinoma patients. *Theranostics*. 2020 Feb 3;10(6):2696-2713. (IF: 8,5).
- 27-** Filser M, Aral B, Airaud F, Chauveau A, Bruce A, Polfrit Y, Thiebaut Y, Gauthier M, Le Maréchal C, Lippert E, Béziau S, Garrec C, Gardie B, Girodon F. Low incidence of *EPOR* mutations in idiopathic erythrocytosis. *Haematologica*. 2020 Mar 12. 2019.244160. (IF: 7).
- 28-** Grenier M, Callegarin D, Nughe M, Gardie B, Riedinger JM, Girodon F. Can absolute polycythaemia be identified without measurement of the red cell mass? *Br J Haematol*. 2020 May 19. (IF: 5,5).
- 29-** Geay A, Aral B, Bourgeois V, Martin P, Airaud F, Garrec C, Bézieau S, **Gardie B**, Girodon F. Diagnosis of exon 12-positive polycythemia vera rescued by NGS. *Clin Case Rep*. 2020 Mar 21;8(5):790-792.
- 30-** Filser M, Giansily-Blaizot M, Grenier M, Monedero Alonso D, Bouyer G, Laurent Pérès, Egée S, Aral B, Airaud F, Da Costa L, Picard V, Cougoul P, Palach M, Béziau S, Garrec C, Patricia Aguilar-Martinez P, **Gardie B\***, Girodon F\*. Increased incidence of germline *PIEZO1* mutations in individuals with idiopathic erythrocytosis. Letter. *Blood*. 2020 Nov 12. (IF: 17,5).  
\* equally contribution to this work.
- 31-** Adolphe A, Ferlicot S, Verkarre V, Posseme K, Couvé S, Garnier P, Droin N, Deloger M, Job B, Giraud S, Bressac-de Paillerets B, **Gardie B**, Richard S, Renaud F, Sophie Gad S.

Germline mutation in the *NBR1* gene involved in autophagy detected in a family with renal tumors. **Cancer Genetics**, in press.

**32-** Gad S, Gwénaél Le Teuff G, Nguyen B, Verkarre V, Duchatelle V, Molinié V, Posseme K, Grandon B, Da Costa M, Bastien Job B, Meurice G, Droin N, Arnaud Méjean A, Couvé S, Renaud F, **Gardie B**, Tean The B, Richard S, Ferlicot S. Involvement of PBMR1 in VHL disease-associated clear cell renal cell carcinoma and its putative relationship with the HIF pathway. **Oncology Letters**, in press.

### **Reviews:**

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**2-** **Gardie B**, Cayuela JM, Sigaux F. "Anomalies de la phase G1 du cycle cellulaire dans les tumeurs humaines." **Hématologie**, 1997, vol3, juillet-août, 319-323.

**3-** Richard S, Joly D, J.M. Corréas JM, Y. Chretien, V. Vasiliu, G. Benoit, S. Ferlicot, D. Bessis, A. Mejean, N. Thiounn, E. Van Glabeke, L. Boccon-Gibod, J.M. Herve, M.F. Avril, B. Escudier, **Gardie B**, R. Lidereau, B. Bressac, D. Chauveau, S. Giraud. Prédispositions héréditaires au cancer rénal. Actualités Néphrologiques Jean Hamburger, **Flammarion Médecine-Sciences**, 2006, pp. 131-150.

**4-** Richard S, Ladroue C, Gad S, Giraud S, **Gardie B**. « Génétique et angiogenèse: l'exemple de la maladie de von Hippel Lindau ». **Bulletin du cancer**, 2007, Jul, 94 Spec No:S170-9.

**5-** Richard S, **Gardie B**, Couvé S, Gad S. "Von Hippel-Lindau: how a rare disease illuminates cancer biology". **Seminar in Cancer Biology**. 2012, may 30. (IF:11)

**6-** Bento C, Percy MJ, **Gardie B**, Maia TM, van Wijk R, Perrotta S, Della Ragione F, Almeida H, Rossi C, Girodon F, Aström M, Neumann D, Schnittger S, Landin B, Minkov M, Randi ML, Richard S, Casadevall N, Vainchenker W, Rives S, Hermouet S, Ribeiro ML, McMullin MF, Cario H. Genetic basis of congenital erythrocytosis: mutation update and online databases. **Human Mutation**, 2014, Jan;35(1):15-26. (IF: 4).

**7-** **Gardie B**, Percy M, Hoogewijs D, Chowdhury R, Bento C, Arsenault P, Richard S, Almeida H, Ewing J, Lambert F, McMullin MF, Schofield C and Lee F. The role of PHD2 mutations in the pathogenesis of erythrocytosis. **Hypoxia**, 2014, 2:71-90. Review and **Vidéo on line**. <http://dx.doi.org/10.2147/HP.S54455>. (IF: new peer reviewed open access journal).

**8-** Bento C, Cario H, **Gardie B**, Hermouet S, McMullin M.F. Co-coordination et écriture de deux chapitres du livre "Congenital erythrocytosis and hereditary thrombocytosis", **MPN&MPN-EuroNet COST final publication**, 2015.

**9-** Hermouet S, Bigot-Corbel E, and **Gardie B**. Pathogenesis of Myeloproliferative Neoplasms: Role and Mechanisms of Chronic Inflammation. **Mediators Inflamm.**, 2015:145293.(IF: 3,7).

**10-** Ratcliffe P, Koivunen P, Johanna Myllyharju J, Ragoussis J, Bovée J, Batinic-Haberle I, Vinatier C, Trichet V, Robriquet F, Oliver L, **Gardie B**. Update on hypoxia-inducible factors and hydroxylases in oxygen regulatory pathways: from physiology to therapeutics. **Hypoxia** (Auckl). 2017 Mar 15; 5:11-20. (IF: new peer reviewed open access journal).

**11-** **Gardie B**, Hermouet S. Chapitre du livre "Erythrocytoses héréditaires". **Traité de Médecine**, 5e édition, Médecine Sciences-Publications/Lavoisier, 2020.

**12-** **Gardie B**, Richard S. "Physiopathology of the Oxygen Sensing". **Revue du Praticien**. 2020 Jan;70(1):17-19.

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