

# Liste des publications

## Articles originaux, revues à comité de lecture (référencés dans les bases)

1 - Laugé A, Lefebvre C, Laurent-Puig P, Caux V, **Gad S**, Eng C, Longy M, Stoppa-Lyonnet D. No evidence for germline *PTEN* mutations in families with breast and brain tumours. *Int J Cancer*. 1999 Jun 21;84(3):216-9.

2 - **Gad S**, Aurias A, Puget N, Mairal A, Schurra C, Montagna M, Pages S, Caux V, Mazoyer S, Bensimon A, Stoppa-Lyonnet D. Color bar coding the *BRCA1* gene on combed DNA: a useful strategy for detecting large gene rearrangements. *Genes Chromosomes Cancer*. 2001 May;31(1):75-84.

3 - **Gad S**, Scheuner MT, Pages-Berhouet S, Caux-Moncoutier V, Bensimon A, Aurias A, Pinto M, Stoppa-Lyonnet D. Identification of a large rearrangement of the *BRCA1* gene using colour bar code on combed DNA in an American breast/ovarian cancer family previously studied by direct sequencing. *J Med Genet*. 2001 Jun;38(6):388-92.

4 - Baldeyron C, Jacquemin E, Smith J, Jacquemont C, De Oliveira I, **Gad S**, Feunteun J, Stoppa-Lyonnet D, Papadopoulo D. A single mutated *BRCA1* allele leads to impaired fidelity of double strand break end-joining. *Oncogene*. 2002 Feb 21;21(9):1401-10.

5 - Puget N, **Gad S**, Perrin-Vidoz L, Sinilnikova OM, Stoppa-Lyonnet D, Lenoir GM, Mazoyer S. Distinct *BRCA1* rearrangements involving the *BRCA1* pseudogene suggest the existence of a recombination hot spot. *Am J Hum Genet*. 2002 Apr;70(4):858-65.

6 - **Gad S**, Caux-Moncoutier V, Pagès-Berhouet S, Gauthier-Villars M, Coupier I, Pujol P, Frénay M, Gilbert B, Maugard C, Bignon YJ, Chevrier A, Rossi A, Fricker JP, Nguyen TD, Demange L, Aurias A, Bensimon A, Stoppa-Lyonnet D. Significant contribution of large *BRCA1* gene rearrangements in 120 French breast and ovarian cancer families. *Oncogene*. 2002 Oct 3;21(44):6841-7.

7 - Casilli F, Di Rocco ZC, **Gad S**, Tournier I, Stoppa-Lyonnet D, Frebourg T, Tosi M. Rapid detection of novel *BRCA1* rearrangements in high-risk breast-ovarian cancer families using multiplex PCR of short fluorescent fragments. *Hum Mutat*. 2002 Sep;20(3):218-26.

8 - **Gad S**, Klinger M, Caux-Moncoutier V, Pages-Berhouet S, Gauthier-Villars M, Coupier I, Bensimon A, Aurias A, Stoppa-Lyonnet D. Bar code screening on combed DNA for large rearrangements of the *BRCA1* and *BRCA2* genes in French breast cancer families. *J Med Genet*. 2002 Nov;39(11):817-21.

9 - Ginolhac SM, **Gad S**, Corbex M, Bressac-De-Paillerets B, Chompret A, Bignon YJ, Peyrat JP, Fournier J, Lasset C, Giraud S, Muller D, Fricker JP, Hardouin A, Berthet P, Maugard C, Nogues C, Lidereau R, Longy M, Olschwang S, Toulas C, Guimbaud R, Yannoukakos D, Szabo C, Durocher F, Moisan AM, Simard J, Mazoyer S, Lynch HT, Goldgar D, Stoppa-Lyonnet D, Lenoir GM, Sinilnikova OM. *BRCA1* wild-type allele modifies risk of ovarian cancer in carriers of *BRCA1* germ-line mutations. *Cancer Epidemiol Biomarkers Prev*. 2003 Feb;12(2):90-5.

10 - **Gad S**, Bièche I, Barrois M, Casilli F, Pages-Berhouet S, Dehainault C, Gauthier-Villars M, Bensimon A, Aurias A, Lidereau R, Bressac-de Paillerets B, Tosi M, Mazoyer S, Stoppa-Lyonnet D. Characterisation of a 161 kb deletion extending from the *NBR1* to the *BRCA1* genes in a French breast-ovarian cancer family. *Hum Mutat*. 2003 Jun;21(6):654.

11 - Blons H, **Gad S**, Zinzindohoué F, Manière I, Beauregard J, Tregouet D, Brasnu D, Beaune P, Laccourreye O, Laurent-Puig P. Matrix metalloproteinase 3 polymorphism: a predictive factor of response to neoadjuvant chemotherapy in head and neck squamous cell carcinoma. *Clin Cancer Res*. 2004 Apr 15;10(8):2594-9.

12 - **Gad S**, Teboul D, Lièvre A, Goasguen N, Berger A, Beaune P, Laurent-Puig P. Is the gene encoding Chibby implicated as a tumour suppressor in colorectal cancer ? *BMC Cancer*. 2004 Jul 9;4:31.

- 13 - Andreu P, Colnot S, Godard C, **Gad S**, Chafey P, Niwa-Kawakita M, Laurent-Puig P, Kahn A, Robine S, Perret C, Romagnolo B. Crypt-restricted proliferation and commitment to the Paneth cell lineage following *Apc* loss in the mouse intestine. *Development*. 2005 Mar;132(6):1443-51.
- 14 - **Gad S**, Lefèvre SH, Khoo SK, Giraud S, Vieillefond A, Vasiliu V, Ferlicot S, Molinié V, Denoux Y, Thiounn N, Chrétien Y, Méjean A, Zerbib M, Benoît G, Hervé JM, Allègre G, Bressac-de Paillerets B, Teh BT, Richard S. Mutations in *BHD* and *TP53* genes, but not in *HNF1beta* gene, in a large series of sporadic chromophobe renal cell carcinoma. *Br J Cancer*. 2007 Jan 29;96(2):336-40. Erratum in: *Br J Cancer*. 2007 Apr 23;96(8):1314.
- 15 - Charbotel B\*, **Gad S\***, Caiola D, Bérout C, Fevotte J, Bergeret A, Ferlicot S, Richard S. Trichloroethylene exposure and somatic mutations of the *VHL* gene in patients with Renal Cell Carcinoma. *J Occup Med Toxicol*. 2007 Nov 12;2:13.
- 16 - Rad FH\*, Ulusakarya A\*, **Gad S**, Sibony M, Juin F, Richard S, Machover D, Uzan G. Novel somatic mutations of the *VHL* gene in an erythropoietin-producing renal carcinoma associated with secondary polycythemia and elevated circulating endothelial progenitor cells. *Am J Hematol*. 2008 Feb;83(2):155-8.
- 17 - Woodward ER, Ricketts C, Killick P, **Gad S**, Morris MR, Kavalier F, Hodgson SV, Giraud S, Bressac-de Paillerets B, Chapman C, Escudier B, Latif F, Richard S, Maher ER. Familial non-VHL clear cell (conventional) renal cell carcinoma: clinical features, segregation analysis, and mutation analysis of *FLCN*. *Clin Cancer Res*. 2008 Sep 15;14(18):5925-30.
- 18 - Ladroue C, Carcenac R, Leporrier M, **Gad S**, Le Hello C, Galateau-Salle F, Feunteun J, Pouysségur J, Richard S, Gardie B. *PHD2* mutation and congenital erythrocytosis with paraganglioma. *N Engl J Med*. 2008 Dec 18;359(25):2685-92.
- 19 - Lorenz P, Dietmann S, Wilhelm T, Koczan D, Autran S, **Gad S**, Wen G, Ding G, Li Y, Rousseau-Merck MF, Thiesen HJ. The ancient mammalian KRAB zinc finger gene cluster on human chromosome 8q24.3 illustrates principles of C2H2 zinc finger evolution associated with unique expression profiles in human tissues. *BMC Genomics*. 2010 Mar 26;11:206.
- 20 - Klomp JA, Petillo D, Niemi NM, Dykema KJ, Chen J, Yang XJ, Sääf A, Zickert P, Aly M, Bergerheim U, Nordenskjöld M, **Gad S**, Giraud S, Denoux Y, Yonneau L, Méjean A, Vasiliu V, Richard S, MacKeigan JP, Teh BT, Furge KA. Birt-Hogg-Dubé renal tumors are genetically distinct from other renal neoplasias and are associated with up-regulation of mitochondrial gene expression. *BMC Med Genomics*. 2010 Dec 16;3:59.
- 21 - Perier A, Fregni G, Wittnebel S, **Gad S**, Allard M, Gervois N, Escudier B, Azzarone B, Caignard A. Mutations of the von Hippel-Lindau gene confer increased susceptibility to natural killer cells of clear-cell renal cell carcinoma. *Oncogene*. 2011 Jun 9;30(23):2622-32.
- 22 - Ladroue C, Hoogewijs D, **Gad S**, Carcenac R, Storti F, Barrois M, Gimenez-Roqueplo AP, Leporrier M, Casadevall N, Hermine O, Kiladjian JJ, Baruchel A, Fakhoury F, Bressac-de Paillerets B, Feunteun J, Mazure N, Pouysségur 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 Jan;97(1):9-14.
- 23 - Bertolotto C, Lesueur F, Giuliano S, Strub T, de Lichy M, Bille K, Dessen P, d'Hayer B, Mohamdi H, Remenieras A, Maubec E, de la Fouchardière A, Molinié V, Vabres P, Dalle S, Poulalhon N, Martin-Denavit T, Thomas L, Andry-Benzaquen P, Dupin N, Boitier F, Rossi A, Perrot JL, Labeille B, Robert C, Escudier B, Caron O, Brugières L, Saule S, Gardie B, **Gad S**, Richard S, Couturier J, Teh BT, Ghiorzo P, Pastorino L, Puig S, Badenas C, Olsson H, Ingvar C, Rouleau E, Lidereau R, Bahadoran P, Vielh P, Corda E, Blanché H, Zelenika D, Galan P; French Familial Melanoma Study Group, Aubin F, Bachollet B, Becuwe C, Berthet P, Bignon YJ, Bonadona V, Bonafe JL, Bonnet-Dupeyron MN, Cambazard F, Chevrant-Breton J, Coupier I, Dalac S, Demange L, d'Incan M, Dugast C, Faivre L, Vincent-Fétita L, Gauthier-Villars M, Gilbert B, Grange F, Grob JJ, Humbert P, Janin N, Joly P, Kerob D, Lasset C, Leroux D, Levang J, Limacher JM, Livideanu C, Longy M, Lortholary A, Stoppa-Lyonnet D, Mansard S, Mansuy L, Marrou K, Matéus C, Maugard C, Meyer N, Nogues C, Souteyrand P, Venat-Bouvet L, Zattara H, Chaudru V, Lenoir GM, Lathrop M, Davidson I, Avril MF, Demenais F, Ballotti R, Bressac-de Paillerets B. A SUMOylation-defective MITF germline mutation predisposes to melanoma and renal carcinoma. *Nature*. 2011 Oct 19;480(7375):94-8. Erratum in: *Nature*. 2016 Mar 3;531(7592):126.

24 - Castro-Vega LJ, Jouravleva K, Liu WY, Martinez C, Gestraud P, Hupé P, Servant N, Albaud B, Gentien D, **Gad S**, Richard S, Bacchetti S, Londoño-Vallejo A. Telomere crisis in kidney epithelial cells promotes the acquisition of a microRNA signature retrieved in aggressive renal cell carcinomas. *Carcinogenesis*. 2013 May;34(5):1173-80.

25 - Popova T, Hebert L, Jacquemin V, **Gad S**, Caux-Moncoutier V, Dubois-d'Enghien C, Richaudeau B, Renaudin X, Sellers J, Nicolas A, Sastre-Garau X, Desjardins L, Gyapay G, Raynal V, Sinilnikova OM, Andrieu N, Manié E, de Pauw A, Gesta P, Bonadona V, Maugard CM, Penet C, Avril MF, Barillot E, Cabaret O, Delattre O, Richard S, Caron O, Benfodda M, Hu HH, Soufir N, Bressac-de Paillerets B, Stoppa-Lyonnet D, Stern MH. Germline *BAP1* mutations predispose to renal cell carcinomas. *Am J Hum Genet*. 2013 Jun 6;92(6):974-80.

26 - 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; ECE-Consortium, Chauveau A, Gimenez-Roqueplo AP, Bressac-de-Paillerets B, Altindirek D, Lorenzo F, Lambert F, Dan H, **Gad-Lapiteau S**, Catarina Oliveira A, Rossi C, Fraga C, Taradin G, Martin-Nuñez G, Vitória H, Diaz Aguado H, Palmblad J, Vidán J, Relvas L, Ribeiro ML, Luigi Larocca M, Luigia Randi M, Pedro Silveira M, Percy M, Gross M, Marques da Costa R, Beshara S, Ben-Ami T, Ugo V; ECE-Consortium. Genetic basis of congenital erythrocytosis: mutation update and online databases. *Hum Mutat*. 2014 Jan;35(1):15-26.

27 - Albiges L, Guegan J, Le Formal A, Verkarre V, Rioux-Leclercq N, Sibony M, Bernhard JC, Camparo P, Merabet Z, Molinie V, Allory Y, Orear C, Couvé S, **Gad S**, Patard JJ, Escudier B. *MET* is a potential target across all papillary renal cell carcinomas: result from a large molecular study of pRCC with CGH array and matching gene expression array. *Clin Cancer Res*. 2014 Jul 1;20(13):3411-21.

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

29 - Benusiglio PR\*, Couvé S\*, Gilbert-Dussardier B, Deveaux S, Le Jeune H, Da Costa M, Fromont G, Memeteau F, Yacoub M, Coupier I, Leroux D, Méjean A, Escudier B, Giraud S, Gimenez-Roqueplo AP, Blondel C, Frouin E, Teh BT, Ferlicot S, Bressac-de Paillerets B, Richard S, **Gad S**. A germline mutation in *PBRM1* predisposes to renal cell carcinoma. *J Med Genet*. 2015 Jun;52(6):426-30.

30 - Hasmim M, Bruno S, Azzi S, Gallerne C, Michel JG, Chiabotto G, Lecoz V, Romei C, Spaggiari GM, Pezzolo A, Pistoia V, Angevin E, **Gad S**, Ferlicot S, Messai Y, Kieda C, Clay D, Sabatini F, Escudier B, Camussi G, Eid P, Azzarone B, Chouaib S. Isolation and characterization of renal cancer stem cells from patient-derived xenografts. *Oncotarget*. 2016 Mar 29;7(13):15507-24.

31 - Perrier-Trudova V, Huimin BW, Kongpetch S, Huang D, Ong P, Le Formal A, Poon SL, Siew 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 Res*. 2015 Dec;35(12):6639-53.

32 - Messai Y, **Gad S\***, Noman MZ\*, Le Teuff G, Couve S, Janji B, Kammerer SF, Rioux-Leclercq N, Hasmim M, Ferlicot S, Baud V, Mejean A, Mole DR, Richard S, Eggermont AM, Albiges L, Mami-Chouaib F, Escudier B, Chouaib S. Renal Cell Carcinoma Programmed Death-ligand 1, a New Direct Target of Hypoxia-inducible Factor-2 Alpha, is Regulated by von Hippel-Lindau Gene Mutation Status. *Eur Urol*. 2016 Oct;70(4):623-632.

33 - Kammerer-Jacquet SF, Crouzet L, Brunot A, Dagher J, Pladys A, Edeline J, Laguerre B, Peyronnet B, Mathieu R, Verhoest G, Patard JJ, Lespagnol A, Mosser J, Denis M, Messai Y, **Gad-Lapiteau S**, Chouaib S, Belaud-Rotureau MA, Bensalah K, Rioux-Leclercq N. Independent association of PD-L1 expression with noninactivated VHL clear cell renal cell carcinoma-A finding with therapeutic potential. *Int J Cancer*. 2017 Jan 1;140(1):142-148.

34 - 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.

35 - Gattolliat CH, Couvé S, Meurice G, Oréar C, Droin N, Chiquet M, Ferlicot S, Verkarre V, Vasiliu V, Molinié V, Méjean A, Dessen P, Giraud S, Bressac-De-Paillerets B, Gardie B, Tean Teh B, Richard S, **Gad S**. Integrative analysis of dysregulated microRNAs and mRNAs in multiple recurrent synchronized renal tumors from patients with von Hippel-Lindau disease. *Int J Oncol*. 2018 Oct;53(4):1455-1468.

### **Autres articles originaux**

36 - **Gad S**, Sultan-Amar V, Meric JB, Izzedine H, Khayat D, Richard S, Rixe O. Somatic von Hippel-Lindau (VHL) gene analysis and clinical outcome under antiangiogenic treatment in metastatic renal cell carcinoma: preliminary results. *Targ Oncol*. 2007 Jan 9;2:3-6.

37 - Benusiglio PR, **Gad S**, Massard C, Carton E, Longchamp E, Faudot T, Lamoril J, Ferlicot S. Case Report: Expanding the tumour spectrum associated with the Birt-Hogg-Dubé cancer susceptibility syndrome. *F1000Res*. 2014 Jul 11;3:159.

### **Articles de synthèse**

38 - Gauthier-Villars M, **Gad S**, Caux V, Pagès S, Blandy C, Stoppa-Lyonnet D. Genetic testing for breast cancer predisposition. *Surg Clin North Am*. 1999 Oct;79(5):1171-87, xxi. Review.

39 - Coupier I, **Gad S**, Gauthier-Villars M, Stoppa-Lyonnet D. Prédilection génétique et cancer de l'ovaire. *Oncologie* 2001;3:130-137. Review. French.

40 - **Gad S**, Aurias A, Bensimon A, Stoppa-Lyonnet D. Peignage d'ADN et grands réarrangements du gène BRCA1, ou comment dénoncer le monopole de Myriad Genetics sur les tests de prédilection au cancer du sein. *Medecine/Sciences* 2001 Oct;17(10):1072-5. Review. French.

41- Richard S, Ladroue C, **Gad S**, Giraud S, Gardie B; Réseau national maladie de VHL et prédispositions héréditaires au cancer du rein de l'Institut national du cancer (INCa). [Genetics and angiogenesis: the example of von Hippel-Lindau disease]. *Bull Cancer*. 2007 Jul;94 Spec No:S170-9. Review. French.

42 - Richard S, Gardie B, Couvé S, **Gad S**. Von Hippel-Lindau: how a rare disease illuminates cancer biology. *Semin Cancer Biol*. 2013 Feb;23(1):26-37. Review.