Axe de recherche génétique et maladies rares

Au GHE : Hôpital Femme Mère Enfant & CBPE

GHN : pavillon T et maternité

GHS : maternité et oncologie

Les publications majeures de l'axe depuis 5 ans

1. Gokce-Samar Z, Vetro A, De Bellescize J, Pisano T, Monteiro L, Penaud N, Korff CM, Fluss J, Marini C, Cesaroni E, Alvarez BM, Sanlaville D, Chatron N, Arzimanoglou AA, Labalme A, Cuddapah VA, Ruggiero SM, Lecoquierre F, Nicolas G, Marie GA, Lebas A, Testard HO, Helbig KL, Ruiz A, Ngoh A, Kurian MA, Reid K, Spaull R, Joset P, Ramantani G, Steindl K, Krenn M, Gerstl L, Vieker S, Craiu D, Pendziwiat M, Haldeman-Englert C, Kanivets I, Romanova I, Rajan DS, Rosenfeld JA, Au M, Grand K, Graham M Jr, Isapof A, Villeneuve N, Smol T, Caumes R, Zacher P, Neuser S, Tinschert S, Platzer K, Bartolomaeus T, Mohnke I, Radtke M, Jamra RA, Helbig I, Jansen FE, Koop K, Rudolf G, Küry S, Courchet J, Guerrini R, **Lesca G**. Molecular and Phenotypic Characterization of the RORB-Related Disorder. Neurology. 2024 Jan 23;102(2):e207945. doi: 10.1212

2. Kvacskay P, El Jammal T, Lorenz HM, Pacheco Y, **Calender A**. Whole exome sequencing of a German sarcoidosis family with four affected and one spontaneous remission case. Rheumatology (Oxford). 2023 Jul 21:kead349. doi: 10.1093/rheumatology/kead349. Epub ahead of print. PMID: 37478346.

3. **Chatron N,** Cabet S, Alix E, Buenerd A, Cox P, Guibaud L, Labalme A, Marks P, Osio D, Putoux A, Sanlaville D, Lesca G, Vasiljevic A. A novel lethal recognizable polymicrogyric syndrome caused by ATP1A2 homozygous truncating variants. Brain. 2019 Nov 1;142(11):3367-3374. doi: 10.1093/brain/awz272

4. Masson J, Pebrel-Richard C, Egloff M, Frétigny M, Beaumont M, Uguen K, Rollat-Farnier PA, Diguet F, Perthus I, Le Gudayer G, Haye D, Dupeyron MB, Putoux A, Raskin-Champion F, Till M, Chatron N, Doray B, Bardel C, Vinciguerra C, **Sanlaville D**, Schluth-Bolard C. Familial transmission of chromoanagenesis leads to unpredictable unbalanced rearrangements through meiotic recombination. Clin Genet. 2023 Apr;103(4):401-412. doi: 10.1111

5. Giannuzzi G, Logsdon GA, Chatron N, Miller DE, Reversat J, Munson KM, Hoekzema K, Bonnet-Dupeyron MN, Rollat-Farnier PA, Baker CA, **Sanlaville D**, Eichler EE, Schluth-Bolard C, Reymond A. Alpha Satellite Insertion Close to an Ancestral Centromeric Region. Mol Biol Evol. 2021 Dec 9;38(12):5576-5587

6. Almentina Ramos Shidi F, Cologne A, Delous M, Besson A, Putoux A, Leutenegger AL, Lacroix V, **Edery P**, Mazoyer S, Bordonné R. Mutations in the non-coding RNU4ATAC gene affect the homeostasis and function of the Integrator complex. Nucleic Acids Res. 2023 Jan 25;51(2):712-727. doi: 10.1093/

7. Morel G, Duhamel C, Boussion S, Frénois F, Lesca G, Chatron N, Labalme A, Sanlaville D, Edery P, Thevenon J, Faivre L, Fassier A, Prodhomme O, Escande F, Manouvrier S, Petit F, Geneviève D, **Rossi M**. Mandibular-pelvic-patellar syndrome is a novel PITX1-related disorder due to alteration of PITX1 transactivation ability. Hum Mutat. 2020 Sep;41(9):1499-1506. doi: 10.1002

8. **Dupuis-Girod S**, Rivière S, Lavigne C, Fargeton AE, Gilbert-Dussardier B, Grobost V, Leguy-Seguin V, Maillard H, Mohamed S, Decullier E, Roux A, Bernard L, Saurin JC, Saroul N, Faure F, Cartier C, Altwegg R, Laccourreye L, Oberti F, Beaudoin M, Dhelens C, Desvignes C, Azzopardi N, Paintaud G, Hermann R, Chinet T. Efficacy and safety of intravenous bevacizumab on severe bleeding associated with hemorrhagic hereditary telangiectasia: A national, randomized multicenter trial.. J Intern Med. 2023 Dec;294(6):761-774. doi: 10.1111

9. **Dupuis-Girod S**, Pitiot V, Bergerot C, Fargeton AE, Beaudoin M, Decullier E, Bréant V, Colombet B, Philouze P, Faure F, Letievant. Efficacy and safety of intravenous bevacizumab on severe bleeding associated with hemorrhagic hereditary telangiectasia: A national, randomized multicenter trial. JC.Sci Rep. 2019 Aug 19;9(1):11986. doi: 10.10

10. **Guilhem A, Dupuis-Girod S**, Espitia O, Rivière S, Seguier J, Kerjouan M, Lavigne C, Maillard H, Magro P, Alric L, Lipsker D, Parrot A, Leguy V, Vanlemmens C, Guibaud L, Vikkula M, Eyries M, Valette PJ, Giraud S. Seven cases of hereditary haemorrhagic telangiectasia-like hepatic vascular abnormalities associated with EPHB4 pathogenic variants. J Med Genet. 2023 Sep;60(9):905-909. doi: 10.1136

11. Kvacskay P, El Jammal T, Lorenz HM, Pacheco Y, **Calender A.** Whole exome sequencing of a German sarcoidosis family with four affected and one spontaneous remission case. Rheumatology (Oxford). 2023 Jul 21:kead349. doi: 10.1093/rheumatology/kead349. Epub ahead of print. PMID: 37478346.

12. Cottin V, Blanchard E, Kerjouan M, Lazor R, Reynaud-Gaubert M, Taille C, Uzunhan Y, Wemeau L, Andrejak C, Baud D, Bonniaud P, Brillet PY, **Calender A**, Chalabreysse L, Court-Fortune I, Desbaillets NP, Ferretti G, Guillemot A, Hardelin L, Kambouchner M, Leclerc V, Lederlin M, Malinge MC, Mancel A,

Marchand-Adam S, Maury JM, Naccache JM, Nasser M, Nunes H, Pagnoux G, Prévot G, Rousset-Jablonski C, Rouviere O, Si-Mohamed S, Touraine R, Traclet J, Turquier S, Vagnarelli S, Ahmad K; OrphaLung network. French recommendations for the diagnosis and management of lymphangioleiomyomatosis. Respir Med Res. 2023 Jun;83:101010. doi: 10.1016/j.resmer.2023.101010. Epub 2023 Mar 24. PMID: 37087906.

13. Pacheco Y, Valeyre D, El Jammal T, Vallee M, Chevalier F, Lamartine J, Sigaudo-Roussel D, Verrier B, Israel-Biet D, Freymond N, Cottin V**, Calender A**. Autophagy and Mitophagy-Related Pathways at the Crossroads of Genetic Pathways Involved in Familial Sarcoidosis and Host-Pathogen Interactions Induced by Coronaviruses. Cells. 2021 Aug 5;10(8):1995. doi: 10.3390/cells10081995. PMID: 34440765; PMCID: PMC8393644.

14. Sève P, Pacheco Y, Durupt F, Jamilloux Y, Gerfaud-Valentin M, Isaac S, Boussel L, **Calender A,** Androdias G, Valeyre D, El Jammal T. Sarcoidosis: A Clinical Overview from Symptoms to Diagnosis. Cells. 2021 Mar 31;10(4):766. doi: 10.3390/cells10040766. PMID: 33807303; PMCID: PMC8066110.

15. **Calender A,** Israel-Biet D, Valeyre D, Pacheco Y. Modeling Potential Autophagy Pathways in COVID-19 and Sarcoidosis. Trends Immunol. 2020 Oct;41(10):856-859. doi: 10.1016/j.it.2020.08.001. Epub 2020 Aug 10. PMID: 32863134; PMCID: PMC7416769.

16. **Calender A**, Weichhart T, Valeyre D, Pacheco Y. Current Insights in Genetics of Sarcoidosis: Functional and Clinical Impacts. J Clin Med. 2020 Aug 13;9(8):2633. doi: 10.3390/jcm9082633. PMID: 32823753; PMCID: PMC7465171.

17. Pacheco Y, Lim CX, Weichhart T, Valeyre D, Bentaher A, **Calender A.** Sarcoidosis and the mTOR, Rac1, and Autophagy Triad. Trends Immunol. 2020 Apr;41(4):286-299. doi: 10.1016/j.it.2020.01.007. Epub 2020 Feb 28. PMID: 32122794.

18. **Calender A**, Lim CX, Weichhart T, Buisson A, Besnard V, Rollat-Farnier PA, Bardel C, Roy P, Cottin V, Devouassoux G, Finat A, Pinson S, Lebecque S, Nunes H, Israel-Biet D, Bentaher A, Valeyre D, Pacheco Y; in the frame of GSF (Group Sarcoidosis France). Exome sequencing and pathogenicity-network analysis of five French families implicate mTOR signalling and autophagy in familial sarcoidosis. Eur Respir J. 2019 Aug 1;54(2):1900430. doi: 10.1183/13993003.00430-2019. PMID: 31023854.

19. Nathan N, Sileo C**, Calender A**, Pacheco Y, Rosental PA, Cavalin C, Macchi O, Valeyre D, Clement A; French Sarcoidosis Group (GSF); Silicosis Research Group. Paediatric sarcoidosis. Paediatr Respir Rev. 2019 Feb;29:53-59. doi: 10.1016/j.prrv.2018.05.003. Epub 2018 May 19. PMID: 30917882.