

## Publications :

1. The GRIA3 c.2477G > A Variant Causes an Exaggerated Startle Reflex, Chorea, and Multifocal Myoclonus. Piard J, Béreau M, XiangWei W, Wirth T, Amsallem D, Buisson L, Richard P, Liu N, Xu Y, Myers SJ, Traynelis SF, Chelly J, Anheim M, Raynaud M, Maldergem LV, Yuan H. *Mov Disord.* 2020 Jul;35(7):1224-1232. doi: 10.1002/mds.28058. Epub 2020 May 5. PMID: 32369665
2. Diagnostic approach to neurotransmitter monoamine disorders: experience from clinical, biochemical, and genetic profiles. Kuster A, Arnoux JB, Barth M, Lamireau D, Houcinat N, Goizet C, Doray B, Gobin S, Schiff M, Cano A, Amsallem D, Barnerias C, Chaumette B, Plaze M, Slama A, Ios C, Desguerre I, Lebre AS, de Lonlay P, Christa L; Individual contributors who contributed to this work. *J Inherit Metab Dis.* 2018 Jan;41(1):129-139. doi: 10.1007/s10545-017-0079-6. Epub 2017 Sep 18. PMID: 28924877
3. Type 3 Gaucher disease, diagnostic in adulthood. Detollenaire C, Bengherbia M, Brassier A, de Villemeur TB, Amsallem D, Berger M, Stirnemann J, Belmatoug N, Rose C. *Mol Genet Metab Rep.* 2017 Jul 11;13:1-2. doi: 10.1016/j.ymgmr.2017.07.002. eCollection 2017 Dec. PMID: 28736718 Free PMC article. No abstract available.
4. DYRK1A mutations in two unrelated patients. Ruaud L, Mignot C, Guét A, Ohl C, Nava C, Héron D, Keren B, Depienne C, Benoit V, Maystadt I, Lederer D, Amsallem D, Piard J. *Eur J Med Genet.* 2015 Mar;58(3):168-74. doi: 10.1016/j.ejmg.2014.12.014. Epub 2015 Jan 30. PMID: 25641759
5. 14q12 and severe Rett-like phenotypes: new clinical insights and physical mapping of FOXG1-regulatory elements. Allou L, Lambert L, Amsallem D, Bieth E, Edery P, Destrée A, Rivier F, Amor D, Thompson E, Nicholl J, Harbord M, Nemos C, Saunier A, Moustaine A, Vigouroux A, Jonveaux P, Philippe C. *Eur J Hum Genet.* 2012 Dec;20(12):1216-23. doi: 10.1038/ejhg.2012.127. Epub 2012 Jun 27. PMID: 22739344 Free PMC article.
6. Diagnostic investigations for an unexplained developmental disability. Verloes A, Héron D, Billette de Villemeur T, Afenjar A, Baumann C, Bahi-Buisson N, Charles P, Faudet A, Jacquette A, Mignot C, Moutard ML, Passemard S, Rio M, Robel L, Rougeot C, Ville D, Burglen L, des Portes V; réseau DéfiScience. *Arch Pediatr.* 2012 Feb;19(2):194-207. Epub 2012 Jan 14. French. PMID: 22245660
7. Age-dependent Mendelian predisposition to herpes simplex virus type 1 encephalitis in childhood. Abel L, Plancoulaine S, Jouanguy E, Zhang SY, Mahfoufi N, Nicolas N, Sancho-Shimizu V, Alcaïs A, Guo Y, Cardon A, Boucherit S, Obach D, Clozel T, Lorenzo L, Amsallem D, Berquin P, Blanc T, Bost-Bru C, Chabrier S, Chabrol B, Cheuret E, Dulac O, Evrard P, Héron B, Lazaro L, Mancini J, Pedespan JM, Rivier F, Vallée L, Lebon P, Rozenberg F, Casanova JL, Tardieu M. *J Pediatr.* 2010 Oct;157(4):623-9. PMID: 20553844
8. Phenotypic variability in Rett syndrome associated with FOXG1 mutations in females. Philippe C, Amsallem D, Francannet C, Lambert L, Saunier A, Verneau F, Jonveaux P. *J Med Genet.* 2010 Jan;47(1):59-65. PMID: 19564653
9. Clinically observed chickenpox and the risk of childhood-onset multiple sclerosis. Mikaeloff Y, Caridade G, Suissa S, Tardieu M; KIDSEP Study Group. *Am J Epidemiol.* 2009 May 15;169(10):1260-6. PMID: 19329530

10. Effectiveness of early beta interferon on the first attack after confirmed multiple sclerosis : a comparative cohort study.  
Mikaeloff Y, Caridade G, Tardieu M, Suissa S; KIDSEP study group of the French Neuropediatric Society.  
Eur J Paediatr Neurol. 2008 May;12(3):205-9. PMID: 17881262
11. Maladie de Gaucher liée à un déficit en saposine C avec traitement par miglustat  
D Amsallem  
Presse Med 2006 : 35 2S35-36
12. Troubles neurologiques associés aux pathologies oculaires en Franche-Comté  
D.Amsallem, N.Khayat, K.Jamal-Bey et al..  
Rev Neurol 2005 : 2S 148
13. Third case of gaucher disease with SAP-C deficiency an evaluation of a twelve months therapy by miglustat  
D.Amsallem, Rodriguez, D., Vanier, MT., Khayat, N., Millat, G., Campello, M., Guillaume, C., Billette de Villemeur, T.  
SSIEM 42 st Annual Symposium 2005.., 6-9 septembre 2005, Paris.
14. Abcès cérébraux néonataux à Citrobacter Koseri  
N.Khayat, D.Amsallem, A.Berger et al.  
Rev Neurol 2005 : 2S 148.
15. Etude de 6 ans des affections démyélinisantes du système nerveux central chez l'enfant au CHU de Besançon.  
D.Amsallem, S.Robin, N.Khayat et al.  
Rev Neurol 2004 :3S 159.
16. Thrombose veineuse cérébrale et leucémie chez l'enfant.  
A.Burger, D.Amsallem, N.Khayat et al.  
Rev Neurol 2004 :3S 73.
17. Intérêt et analyse critique des EEG pédiatriques au CHU de Besançon : étude 2002 et 2003.  
N.Khayat, D.Amsallem, F.Coquet et al.  
Rev Neurol 2004 : 3S 154.
18. A detailed analysis of the MECP2 gene: prevalence of recurrent mutations and gross DNA rearrangements in Rett syndrome patients.  
Bourdon V, Philippe C, Labrune O, Amsallem D, Arnould C, Jonveaux P.  
Hum Genet. 2001 Jan;108(1):43-50. PMID: 11214906
19. Early onset neuronal intranuclear inclusion disease : a cause of childhood pontocerebellar atrophy.  
A.Gelot, G.Ponsot, D.Amsallem, D.Rodriguez.  
Brain pathology 2000 : Vol 10, n°4, C54-02.
20. Empirical treatment of fever in neutropenic children: the role of the carbapenems. International Antimicrobial Therapy Cooperative Group of the European Organisation for Research and Treatment of Cancer and the Gimema Infection Program.  
Cometta A, Viscoli C, Castagnola E, Massimo L, Giacchino R, Gibson B, Giacchino M, Balbo L, Engelhard D, Shapiro M, Amsallem D, Estavoyer JM, Ferster A, Glauser MP.  
Pediatr Infect Dis J. 1996 Aug;15(8):744-8. PMID: 8858693
21. Emery-Dreifuss disease with dominant autosomal transmission. A new family.  
Bensaid J, Vallat JM, Virot P, Amsallem D, Rauscher M.  
Presse Med. 1995 Sep 30;24(28):1317. PMID: 7501631
22. Total permanent auricular paralysis. Review of the literature apropos of 109 cases.

Bensaid J, Vallat JM, Amsallem D, Bernard Y, Rauscher M, Borsotti JP.  
Ann Cardiol Angeiol (Paris). 1995 Mar;44(3):139-45. PMID: 7793852

23. Vertebral-artery dissection following a judo session: a case report.  
Lannuzel A, Moulin T, Amsallem D, Galmiche J, Rumbach L.  
Neuropediatrics. 1994 Apr;25(2):106-8. PMID: 8072674