CURRICULUM VITAE

Discipline : Médecine **Spécialité : Pédiatrie, Neurologie et épileptologie pédiatrique**

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1. Qualification

Diplômes :
Doctorat en Médecine obtenu le 23.10.90
(Faculté de Médecine de Strasbourg)
DES de Pédiatrie obtenu le 01.05.90
(Faculté de Médecine de Strasbourg)
DIU de Neurophysiologie Clinique obtenu le 10.09.93
(Faculté de médecine Pitié-Salpêtrière - Paris)
DIU de Neurologie Pédiatrique obtenu le 22.12.95
(Université R. Descartes- Hôpital Saint-Vincent-de-Paul)
DEA de Neuropsychologie Humaine obtenu le 16.07.01
(Université Paul Sabatier- Toulouse)

Titres :
Ancien Interne des Hôpitaux de Strasbourg, titre obtenu le 01.05.90
Ancien Chef de Clinique-Assistant des Hôpitaux, titre obtenu le 22.11.92
Attachée en Premier du 01.01.96 au 01.09.99

Membre de la Société Française de Neuropédiatrie
Membre de la Ligue Française Contre l'Epilepsie

Fonction :
Praticien hospitalier TP au service de Pédiatrie 1 (Hôpitaux Universitaires de Strasbourg)
Coordination du site constitutif du Centre de Référence des Epilepsies Rares
Coordination du Centre Référent des Troubles du Langage et des Apprentissages

Affiliation :
IGBMC-CNRS UMR 7104-Inserm U964 (J Chelly) (janvier 2015)

Recherche
-investigateur principal PHRC-I 2017
-protocoles essais cliniques avec le CIC pédiatrique
-BPC actualisées en avril 2019

Enseignement
DES de pédiatrie
DIU de neurologie pédiatrique
Master de neuropsychologie (UDS), orthophonie
Encadrement DIU troubles du neurodéveloppement, DU troubles des apprentissages

2.Travaux :

Encadrement de thèses de médecine, mémoires DES

Communications, posters congrès annuels SFNP, JFE, AES, EPNS

Articles (2012-2020)

102. Exome sequencing in 57 patients with self-limited focal epilepsies of childhood with typical or atypical presentations suggests novel candidate genes.
Rudolf G, de Bellescize J, **de Saint Martin A**, Arzimanoglou A, Valenti Hirsch MP, Labalme A, Boulay C, Simonet T, Boland A, Deleuze JF, Nitschké P, Ollivier E, Sanlaville D, Hirsch E, Chelly J, Lesca G. Eur J Paediatr Neurol. 2020 Jul;27:104-110. doi: 10.1016/j.ejpn.2020.05.003. Epub 2020 May 29.

101.Early-onset epileptic encephalopathy related to germline PIGA mutations: A series of 5 cases.
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99.Remarkable effect of transdermal nicotine in children with CHRNA4-related autosomal dominant sleep-related hypermotor epilepsy.
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98.Transition from ketogenic diet to triheptanoin in patients with GLUT1 deficiency syndrome.
Hainque E, Meneret A, Gras D, Atencio M, Luton MP, Barbier M, **De Saint Martin A**, Billette de Villemeur T, Ottolenghi C, Roze E, Mochel F.
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97.Expanding the genetic and phenotypic relevance of KCNB1 variants in developmental and epileptic encephalopathies: 27 new patients and overview of the literature.
Bar C, Barcia G, Jennesson M, Le Guyader G, Schneider A, Mignot C, Lesca G, Breuillard D, Montomoli M, Keren B, Doummar D, Billette de Villemeur T, Afenjar A, Marey I, Gerard M, Isnard H, Poisson A, Dupont S, Berquin P, Meyer P, Genevieve D, **De Saint Martin A**, El Chehadeh S, Chelly J, Guët A, Scalais E, Dorison N, Myers CT, Mefford HC, Howell KB,

Marini C, Freeman JL, Nica A, Terrone G, Sekhara T, Lebre AS, Odent S, Sadleir LG, Munnich A, Guerrini R, Scheffer IE, Kabashi E, Nabbout R.
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96.KCNT1 epilepsy with migrating focal seizures shows a temporal sequence with poor outcome, high mortality and SUDEP.
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94. Cognitive impairment in children with CACNA1A mutations.
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92. Quantitative analysis and EEG markers of KCNT1 epilepsy of infancy with migrating focal seizures.
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91. Correction: The landscape of epilepsy-related GATOR1 variants.
Baldassari S, Picard F, Verbeek NE, van Kempen M, Brilstra EH, Lesca G, Conti V, Guerrini R, Bisulli F, Licchetta L, Pippucci T, Tinuper P, Hirsch E, **de Saint Martin A**, Chelly J, Rudolf G, Chipaux M, Ferrand-Sorbets S, Dorfmüller G, Sisodiya S, Balestrini S, Schoeler N, Hernandez-Hernandez L, Krithika S, Oegema R, Hagebeuk E, Gunning B, Deckers C, Berghuis B, Wegner I, Niks EH, Jansen FE, Braun K, de Jong D, Rubboli G, Talvik I, Sander V, Uldall P, Jacquemont ML, Nava C, Leguern E, Julia S, Gambardella A, d'Orsi G, Crichiutti G, Faivre L, Darmency V, Benova B, Krsek P, Biraben A, Lebre AS, Jennesson M, Sattar S, Marchal C, NordliJr DR, Lindstrom K, Striano P, Lomax LB, Kiss C, Bartolomei F, Lepine AF, Schoonjans AS, Stouffs K, Jansen A, Panagiotakaki E, Ricard-Mousnier B,

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Baldassari S, Picard F, Verbeek NE, van Kempen M, Brilstra EH, Lesca G, Conti V, Guerrini R, Bisulli F, Licchetta L, Pippucci T, Tinuper P, Hirsch E, **de Saint Martin A**, Chelly J, Rudolf G, Chipaux M, Ferrand-Sorbets S, Dorfmüller G, Sisodiya S, Balestrini S, Schoeler N, Hernandez-Hernandez L, Krithika S, Oegema R, Hagebeuk E, Gunning B, Deckers C, Berghuis B, Wegner I, Niks E, Jansen FE, Braun K, de Jong D, Rubboli G, Talvik I, Sander V, Uldall P, Jacquemont ML, Nava C, Leguern E, Julia S, Gambardella A, d'Orsi G, Crichiutti G, Faivre L, Darmency V, Benova B, Krsek P, Biraben A, Lebre AS, Jennesson M, Sattar S, Marchal C, Nordli DR Jr, Lindstrom K, Striano P, Lomax LB, Kiss C, Bartolomei F, Lepine AF, Schoonjans AS, Stouffs K, Jansen A, Panagiotakaki E, Ricard-Mousnier B, Thevenon J, de Bellescize J, Catenoix H, Dorn T, Zenker M, Müller-Schlüter K, Brandt C, Krey I, Polster T, Wolff M, Balci M, Rostasy K, Achaz G, Zacher P, Becher T, Cloppenborg T, Yuskaitis CJ, Weckhuysen S, Poduri A, Lemke JR, Møller RS, Baulac S.
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88. Benign paroxysmal torticollis, benign paroxysmal vertigo, and benign tonic upward gaze are not benign disorders.
Humbertclaude V, Krams B, Nogue E, Nagot N, Annequin D, Tourniaire B, Tournier-Lasserve E, Riant F, Roubertie A; Episodic Syndromes Consortium.
Dev Med Child Neurol. 2018 Dec;60(12):1256-1263. doi: 10.1111/dmcn.13935. Epub 2018 Jun 21.

87. Defining the phenotypic spectrum of SLC6A1 mutations.
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Hum Genet. 2017 Apr;136(4):463-479.

81 Efficacy of a ketogenic diet in resistant myoclono-astatic epilepsy: A French multicenter retrospective study.
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MT, de Sain-van der Velde MG, Jans JJ, Amiel J, Avsec Z, Mertes C, Haack TB, Strom T, Meitinger T, Bonnen PE, Taylor RW, Gagneur J, van Hasselt PM, Rötig A, Delahodde A, Prokisch H, Fuchs SA, Paquis-Flucklinger V.Am J Hum Genet. 2017 Jan 5;100(1):151-159.

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