CURRICULUM VITAE

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Gilbert and Rose-Marie Chagoury School Of Medicine

Byblos – Lebanon

EDUCATION

2008: PhD from "Université de Versailles, Saint Quentin en Yvelines, France".
2002-2003: Diplôme d'Etudes Approfondies (M2) from « Université de Versailles Saint Quentin en Yvelines », France.
2000: Certificate of Genetics and Molecular Biology, St Joseph University, Lebanon.
1999: Masters of Sciences in Laboratory Medical Analysis. St Joseph University, Lebanon.
1998: BS in Sciences in Laboratory Medical Analysis. St Joseph University, Lebanon.
1996: Participation at the Workshops organized by the Lebanese School of Molecular Biology Method of Southern Blot, Polymorphisms Length of Restriction Fragments, Flow Cytometry.
1994: Lebanese Baccalaureat in secondary education, second part, science series. Maison Provinciale des filles de la charité, Lebanon.
French Baccalaureat in secondary education, Mathematics and sciences of nature (section D). Maison Provinciale des filles de la charité, Lebanon.

PROFESSIONAL EXPERIENCES

September 2020-ongoing : Associate Professor of Human Genetics, Department of Human Genetics, Gilbert and Rose-Marie Chagoury School Of Medicine, Byblos – Lebanon
1998-August 2019: Director of the Molecular Biology section at the Medical Genetics Unit, Faculty of Medicine, St Joseph University. Molecular diagnosis of inherited diseases by means of different techniques (Sanger sequencing, Next Generation sequencing-WES, Oligo SNP arrays- CGH arrays, MLPA, Real Time PCR, Digital PCR...)
Human Identification. Preimplantation Genetic diagnosis.
h-index: 32, Research gate score : 39.89
2008-2020: in charge of the molecular biology diagnosis at the Genetics Research Laboratory at Chronic Care center.
2017-31/08/2020: Associate Professor at the faculty of Medicine of Saint Joseph University. Teaching molecular Biology and basics of genetics.
2018: One month training (December) at Institut de Pathologie et de Genetique Humaine (IPG) – Belgium.
2009-2017: Assistant Professor at the faculty of Medicine of Saint Joseph University. Teaching molecular Biology and basics of genetics.
2002-2009: Instructor at the faculty of Medicine of Saint Joseph University. Teaching molecular Biology and basics of genetics.
2015: 2nd Prize « Prix d'excellence franco-libanais pour la recherche scientifique »
2012: Participation at the European Society of human Genetics conference. Nürnberg, Germany, June 23-26, 2012**.**
2012: Participation at the European Society of human Genetics conference. Nürnberg, Germany, June 23-26, 2012**.**

2011: Participation at the European Society of human Genetics conference. Amsterdam RAI, The Netherlands, May 28 - 31, 2011.
2011: Participation to the "4th course in "The integration of cytogenetics, microarrays and massive sequencing in biomedical and clinical research", "EuroMediterranean University Center" of Ronzano, Bologna, October 25-28, 2011.
2010: Participation at the European Society of human Genetics conference. Gothenburg, Sweden - June 12 - 15, 2010
2009: Participation to the "2nd course in "The integration of cytogenetics, microarrays and massive sequencing in biomedical and clinical research", "EuroMediterranean University Center" of Ronzano, Bologna, October 18-22, 2009.
2009: Participation at the European Society of human Genetics conference. Barcelona, Espagne, 31 mai-3 juin 2008.
2008: Participation at the European human Genetics conference. Barcelona, Spain, 31 may-3 june 2008.
2007: Participation at the European human Genetics conference. Nice, France, June 2007.
2006: Participation at the European human Genetics conference. Amsterdam, The Netherlands, 6-9 may 2006.
2005: Participation at the European human Genetics conference. Prague, CZECH Republic, 7-10 may 2005.
2003-2006: Teaching the Molecular Biology courses to the 2nd year of medicine students (PCEM2)-Faculty of Medicine Saint Joseph University, Lebanon
2005: Participation at the workshop of Quantitative Real Time PCR at Pasteur institute of Tunis, Tunisia December 6-11, 2004.
2004: Three weeks training (15 the Mars at April 2) at the Forensic laboratory of imprinting at the service of biochemistry and molecular biology, Hospital Raymond Poincaré.
2004: Participation at the second Human and Medical Genetics conference "assises de Génétique Humaine et Médicale". Angers-France-30, January 31 and February 1,
2001: Six weeks training (of November 15 to December 24) at the Center of Studies of antenatal Biology, at the University of Versailles, St Quentin in Yvelines.
2000-2001: Two months training (December and January) at the Pasteur Institute, unit of Genetics of sensory deficits.
1998: Two months training (November and December) at the Laboratory of Human Molecular Genetics. Faculty of Pharmacy. Claude Bernard University Lyon.

PUBLICATIONS

1-Medlej-Hashim M, Rawashdeh M, Chouery E, Mansour I, Delague V, Lefranc G, Naman R, Loiselet J, Megarbane A.
Genetic screening of fourteen mutations in Jordanian familial Mediterranean fever patients.
Hum Mutat. 2000 Apr;15(4):384.

2- Delague V, Bareil C, Tuffery S, Bouvagnet P, Chouery E, Koussa S, Maisonobe T, Loiselet J, Megarbane A, Claustres M.
Mapping of a new locus for autosomal recessive demyelinating Charcot-Marie-Tooth disease to 19q13.1-13.3 in a large consanguineous Lebanese family: exclusion of MAG as a candidate gene.
Am J Hum Genet. 2000 Jul;67(1):236-43.

3- Delague V, Souraty N, Khallouf E, Tardy V, Chouery E, Halaby G, Loiselet J, Morel Y, Megarbane A.

Mutational analysis in Lebanese patients with congenital adrenal hyperplasia due to a deficit in 21-hydroxylase.
Horm Res. 2000;53(2):77-82.

4- Megarbane A, Waked N, Chouery E, Moglabey YB, Saliba N, Mornet E, Serre JL, Slim R.
Microcephaly, cutis verticis gyrata of the scalp, retinitis pigmentosa, cataracts, sensorineural deafness, and mental retardation in two brothers.
Am J Med Genet. 2001 Jan 22;98(3):244-9.

5- Mansour I, Delague V, Cazeneuve C, Dode C, Chouery E, Pecheux C, Medlej-Hashim M, Salem N, El Zein L, Levan-Petit I, Lefranc G, Goossens M, Delpech M, Amselem S, Loiselet J, Grateau G, Megarbane A, Naman R.
Familial Mediterranean fever in Lebanon: mutation spectrum, evidence for cases in Maronites, Greek orthodoxes, Greek catholics, Syriacs and Chiites and for an association between amyloidosis and M694V and M694I mutations.
Eur J Hum Genet. 2001 Jan;9(1):51-5.

6- Delague V, Bareil C, Bouvagnet P, Salem N, Chouery E, Loiselet J, Megarbane A, Claustres M.
Nonprogressive autosomal recessive ataxia maps to chromosome 9q34-9qter in a large consanguineous Lebanese family.
Ann Neurol. 2001 Aug;50(2):250-3.

7- Mustapha M, Salem N, Delague V, Chouery E, Ghassibeh M, Rai M, Loiselet J, Petit C, Megarbane A.
Autosomal recessive non-syndromic hearing loss in the Lebanese population: prevalence of the 30delG mutation and report of two novel mutations in the connexin 26 (GJB2) gene.
J Med Genet. 2001 Oct;38(10):E36.

8- Medlej-Hashim M, Petit I, Adib S, Chouery E, Salem N, Delague V, Rawashdeh M, Mansour I, Lefranc G, Naman R, Loiselet J, Lecron JC, Serre JL, Megarbane A.
Familial Mediterranean Fever: association of elevated IgD plasma levels with specific MEFV mutations.
Eur J Hum Genet. 2001 Nov;9(11):849-54.

9- Medlej-Hashim M, Salem N, Chouery E, Rawashdeh M, Delague V, Haffar M, Mansour I, Naman R, Lefranc G, Loiselet J, Megarbane A.
Familial Mediterranean fever: the potential for misdiagnosis of E148V using the E148Q usual RFLP Detection method.
Clin Genet. 2002 Jan;61(1):71-3.

10- Mustapha M, Chouery E, Chardenoux S, Naboulsi M, Paronnaud J, Lemainque A, Megarbane A, Loiselet J, Weil D, Lathrop M, Petit C.
DFNB31, a recessive form of sensorineural hearing loss, maps to chromosome 9q32-34.
Eur J Hum Genet. 2002 Mar;10(3):210-2.

11- Mustapha M, Chouery E, Torchard-Pagnez D, Nouaille S, Khrais A, Sayegh FN, Megarbane A, Loiselet J, Lathrop M, Petit C, Weil D.
A novel locus for Usher syndrome type I, USH1G, maps to chromosome 17q24-25.

Hum Genet. 2002 Apr;110(4):348-50.

12- Megarbane A, Hersh JH, Chouery E, Fabre M.
Craniosynostosis, telecanthus, scalp hair abnormalities, and sensorineural deafness in two sibs.
Am J Med Genet. 2002 May 15;109(4):323-7.

13- Megarbane A, Sanders A, Chouery E, Delague V, Medlej-Hashim M, Torbey PH.
An unknown autoinflammatory syndrome associated with short stature and dysmorphic features in a young boy.
J Rheumatol. 2002 May;29(5):1084-7.

14- Delague V, Bareil C, Bouvagnet P, Salem N, Chouery E, Loiselet J, Megarbane A, Claustres M.
A new autosomal recessive non-progressive congenital cerebellar ataxia associated with mental retardation, optic atrophy, and skin abnormalities (CAMOS) maps to chromosome 15q24-q26 in a large consanguineous Lebanese Druze Family.
Neurogenetics.2002 Mar;4(1):23-7.

15- Medlej-Hashim M, Mustapha M, Chouery E, Weil D, Parronaud J, Salem N, Delague V, Loiselet J, Lathrop M, Petit C, Megarbane A.
Non-syndromic recessive deafness in Jordan: mapping of a new locus to chromosome 9q34.3 and prevalence of DFNB1 mutations.
Eur J Hum Genet. 2002 Jun;10(6):391-4.

16- Megarbane A, Rassi S, Chouery E, Delague V, Perez de Nanclares Leal G, Tabet M, Castano L, Loiselet J.
A new dominant branchiogenic-deafness syndrome with internal auditory canal hypoplasia and abnormal extremities.
Am J Med Genet. 2003 Jul 15;120A(2):276-82.

17- Abifadel M, Jambart S, Allard D, Rabes JP, Varret M, Derre A, Chouery E, Salem N, Junien C, Aydenian H, Boileau C.
Identification of the first Lebanese mutation in the LPL gene and description of a rapid detection method.
Clin Genet. 2004 Feb;65(2):158-61

18-Medlej-Hashim M, Delague V, Chouery E, Salem N, Rawashdeh M, Lefranc G, Loiselet J, Megarbane A.
Amyloidosis in familial Mediterranean fever patients: correlation with MEFV genotype and SAA1 and MICA polymorphisms effects.
BMC Med Genet. 2004 Feb 10;5(1):4.

19- Megarbane A, Daou L, Megarbane H, Cave H, Chouery E, Verloes A.
New autosomal recessive syndrome with short stature and facio-auriculo-thoracic malformations.
Am J Med Genet. 2004 Aug 1;128A(4):414-7.

20- Ged C, Megarbane H, Chouery E, Lalanne M, Megarbane A, de Verneuil H.
Congenital erythropoietic porphyria: report of a novel mutation with absence of clinical manifestations in a homozygous mutant sibling.

J Invest Dermatol. 2004 Sep;123(3):589-91.

21 - Medlej-Hashim M, Serre JL, Corbani S, Saab O, Jalkh N, Delague V, Chouery E, Salem N, Loiselet J, Lefranc G, Mégarbané A (2005).
Familial Mediterranean fever (FMF) in Lebanon and Jordan: a population genetics study and report of three novel mutations. Eur J Med Genet. 48(4):412-20.

22 - Delague V, Souaid M, Chouery E, Depetris D, Sanlaville D, Mattei MG, Mégarbané A. Screening for subtelomeric rearrangements using automated fluorescent genotyping of microsatellite markers: a Lebanese study. Eur J Med Genet. 2006 Mar-Apr;49(2):117-26.

23- Stoetzel C, Laurier V, Davis EE, Muller J, Rix S, Badano JL, Leitch CC, Salem N, Chouery E, Corbani S, Jalk N, Vicaire S, Sarda P, Hamel C, Lacombe D, Holder M, Odent S, Holder S, Brooks AS, Elcioglu NH, Da Silva E, Rossillion B, Sigaudy S, de Ravel TJ, Alan Lewis R, Leheup B, Verloes A, Amati-Bonneau P, Mégarbané A, Poch O, Bonneau D, Beales PL, Mandel JL, Katsanis N, Dollfus H.
BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus.
Nat Genet 2006 38(5):521-4

24- Hoffmann K, Muller JS, Stricker S, Megarbane A, Rajab A, Lindner TH, Cohen M, Chouery E, Adaimy L, Ghanem I, Delague V, Boltshauser E, Talim B, Horvath R, Robinson PN, Lochmuller H, Hubner C, Mundlos S.
Escobar syndrome is a prenatal myasthenia caused by disruption of the acetylcholine receptor fetal gamma subunit.
Am J Hum Genet. 2006 Aug;79(2):303-12.

25- Megarbane H, Boehm N, Chouery E, Bernard R, Salem N, Halaby E, Levy N, Megarbane A.
X-linked reticulate pigmentary layer. Report of a new patient and demonstration of a skewed X-inactivation.
Genet Couns. 2005;16(1):85-9.

26- Megarbane A, Chouery E, Rassi S, Delague V.
A new autosomal recessive oto-facial syndrome with midline malformations. Am J Med Genet A. 2005 Feb 1;132(4):398-401.

27- De Sandre-Giovannoli A, Delague V, Hamadouche T, Chaouch M, Krahn M, Boccaccio I, Maisonobe T, Chouery E, Jabbour R, Atweh S, Grid D, Megarbane A, Levy N.
Homozygosity mapping of autosomal recessive demyelinating Charcot-Marie-Tooth neuropathy (CMT4H) to a novel locus on chromosome 12p11.21-q13.11.
J Med Genet. 2005 Mar;42(3):260-5.

28-Gannage-Yared MH, Dode C, Ghanem I, Chouery E, Jalkh N, Hardelin JP, Megarbane A.
Coexistence of Kallmann syndrome and complete androgen insensitivity in the same patient. Eur J Endocrinol. 2005 Jun;152(6):813-7.

29- Belguith H, Hajji S, Salem N, Charfeddine I, Lahmar I, Amor MB, Ouldim K, Chouery E, Driss N, Drira M, Megarbane A, Rebai A, Sefiani A, Masmoudi S, Ayadi H.

Analysis of GJB2 mutation: evidence for a Mediterranean ancestor for the 35delG mutation. Clin Genet. 2005 Aug;68(2):188-9.

30-Delague V, Chouery E, Corbani S, Ghanem I, Aamar S, Fischer J, Levy-Lahad E, Urtizberea JA, Megarbane A.
Molecular study of WISP3 in nine families originating from the Middle-East and presenting with progressive pseudorheumatoid dysplasia: identification of two novel mutations, and description of a founder effect.
Am J Med Genet A. 2005 Oct 1;138(2):118-26.

31- Van Maldergem L, Siitonen HA, Jalkh N, Chouery E, De Roy M, Delague V, Muenke M, Jabs EW, Cai J, Wang LL, Plon SE, Fourneau C, Kestila M, Gillerot Y, Megarbane A, Verloes A.
Revisiting the craniosynostosis-radial ray hypoplasia association: Baller-Gerold syndrome caused by mutations in the RECQL4 gene.
J Med Genet. 2006 Feb;43(2):148-52. Epub 2005 Jun 17.

32- Laurier V, Stoetzel C, Muller J, Thibault C, Corbani S, Jalkh N, Salem N, Chouery E, Poch O, Licaire S, Danse JM, Amati-Bonneau P, Bonneau D, Megarbane A, Mandel JL, Dollfus H.
Pitfalls of homozygosity mapping: an extended consanguineous Bardet-Biedl syndrome family with two mutant genes (BBS2, BBS10), three mutations, but no triallelism.
Eur J Hum Genet. 2006 Jul 5

33- Hoffmann K, Muller JS, Stricker S, Megarbane A, Rajab A, Lindner TH, Cohen M, Chouery E, Adaimy L, Ghanem I, Delague V, Boltshauser E, Talim B, Horvath R,Robinson PN, Lochmüller H, Hübner C, Mundlos S.
Escobar syndrome is a prenatal myasthenia caused by disruption of the acetylcholine receptor fetal gamma subunit. Am J Hum Genet. 2006 Aug;79(2):303-12.

34- Souraty N, Noun P, Djambas-Khayat C, Chouery E, Pangrazio A, Villa A, Lefranc G, Frattini A, Mégarbané A.
Molecular study of six families originating from the Middle-East and presenting with autosomal recessive osteopetrosis. Eur J Med Genet. 2007 May-Jun;50(3):188-99. Epub 2007 Feb 21.

35- Delague V, Jacquier A, Hamadouche T, Poitelon Y, Baudot C, Boccaccio I, Chouery E, Chaouch M, Kassouri N, Jabbour R, Grid D, Mégarbané A, Haase G, Lévy N.
Mutations in FGD4 encoding the Rho GDP/GTP exchange factor FRABIN cause autosomal recessive Charcot-Marie-Tooth type 4H. Am J Hum Genet. 2007 Jul;81(1):1-16.

36- Jalkh N, Génin E, Chouery E, Delague V, Medlej-Hashim M, Idrac CA, Mégarbané A, Serre JL.
Familial Mediterranean Fever in Lebanon: founder effects for different MEFV mutations. Ann Hum Genet. 2008 Jan;72(Pt 1):41-7.

37- Adaimy L, Chouery E, Megarbane H, Mroueh S, Delague V, Nicolas E, Belguith H, de Mazancourt P, Megarbane A.
Mutation in WNT10A is associated with an autosomal recessive ectodermal dysplasia: the odonto-onycho-dermal dysplasia. Am J Hum Genet. 2007 Oct;81(4):821-8.

38- Djambas Khayat C, Salem N, Chouery E, Corbani S, Moix I, Nicolas E, Morris MA, DE Moerloose P, Mégarbané A.
Molecular analysis of F8 in Lebanese haemophilia A patients: novel mutations and phenotype-genotype correlation. Haemophilia. 2008 May 12.

39- Chouery E, Delague V, Bergougnoux A, Koussa S, Serre JL, Mégarbané A.
Mutations in TREM2 lead to pure early-onset dementia without bone cysts. Hum Mutat. 2008 Jun 10;29(9):E194-E204.

40- Mégarbané A, Chouery E, Ghanem I.
A multiplex family with possible metaphyseal Spahr-type dysplasia and exclusion of RMRP and COL10A1 as candidate genes. Am J Med Genet A. 2008 Jul 15;146A(14):1865-70.

41- Chouery E, Kfoury J, Delague V, Jalkh N, Bejjani P, Serre JL, Mégarbané A.
A novel locus for autosomal recessive primary torsion dystonia (DYT17) maps to 20p11.22-q13.12. Neurogenetics. 2008 Oct;9(4):287-93.

42- Belguith H, Masmoudi S, Medlej-Hashim M, Chouery E, Weil D, Ayadi H, Petit C, Mégarbané A.
Re-assigning the DFNB33 locus to chromosome 10p11.23-q21.1.
Eur J Hum Genet. 2008 Sep 10.

43- Polok B, Escher P, Ambresin A, Chouery E, Bolay S, Meunier I, Nan F, Hamel C, Munier FL, Thilo B, Mégarbané A, Schorderet DF.
Mutations in CNNM4 cause recessive cone-rod dystrophy with amelogenesis imperfecta. Am J Hum Genet. 2009 Feb;84(2):259-65.

44- Nader L, Lahoud L, Chouery E, Aftimos G, Bois P, Farès NA. B-type natriuretic peptide receptors in hypertrophied adult rat cardiomyocytes. Ann Cardiol Angeiol (Paris). 2010 Feb;59(1):20-4. Epub 2009 Oct 17. PubMed PMID: 19969282.

45- Stora S, Conte M, Chouery E, Richa S, Jalkh N, Gillart AC, Joannis AL, Mégarbané A. A 56-year-old female patient with facio-oculo-acoustico-renal syndrome (FOAR) syndrome. Report on the natural history and of a novel mutation. Eur J Med Genet. 2009 Sep-Oct;52(5):341-3. Epub 2009 Jul 3. PubMed PMID: 19577669.

46- Chouery E, Pangrazio A, Frattini A, Villa A, Van Wesenbeeck L, Piters E, Van Hul W, Coxon FP, Schouten T, Helfrich M, Lefranc G, Mégarbané A. A new familial sclerosing bone dysplasia. J Bone Miner Res. 2010 Mar;25(3):676-80. PubMed PMID: 20422625.

47- Nicolas E, Poitelon Y, Chouery E, Salem N, Levy N, Mégarbané A, Delague V. CAMOS, a nonprogressive, autosomal recessive, congenital cerebellar ataxia, is caused by a mutant zinc-finger protein, ZNF592. Eur J Hum Genet. 2010 Oct;18(10):1107-13. Epub 2010 Jun 9. PubMed PMID: 20531441.

48- Saliba Y, Chouery E, Mégarbané A, Jabbour H, Fares N. Microalbuminuria versus brain natriuretic peptide in cardiac hypertrophy of hypertensive rats. Physiol Res. 2010 Jun 9. [Epub ahead of print] PubMed PMID: 20533867.

49- Medlej-Hashim M, Nehme N, Chouery E, Jalkh N, Megarbane A. 1Novel MEFV transcripts in Familial Mediterranean fever patients and controls. BMC Med Genet. 2010 Jun 9;11:87. PubMed PMID: 20534143; PubMed Central PMCID: PMC2894788.

50- Chouery E, Delague V, Jalkh N, Salem N, Kfoury J, Rodriguez D, Chabrol B, Boespflug-Tanguy O, Lévy N, Serre JL, Mégarbané A. A whole-genome scan in a large family with leukodystrophy and oligodontia reveals linkage to 10q22. Neurogenetics. 2010 Aug 19. [Epub ahead of print] PubMed PMID: 20721593.

51- Chouery E, Coble MD, Strouss KM, Saunier JL, Jalkh N, Medlej-Hashim M, Ayoub F, Mégarbané A. Population genetic data for 17 STR markers from Lebanon. Leg Med (Tokyo). 2010 Nov;12(6):324-6. Epub 2010 Sep 21. PubMed PMID: 20863737.

52- Medlej-Hashim M, Chouery E, Salem N, Delague V, Lefranc G, Loiselet J, Mégarbané A. Familial Mediterranean fever in a large Lebanese family: Multiple MEFV mutations and evidence for a Founder effect of the p.[M694I] mutation. Eur J Med Genet. 2010 Oct 15.

53- Okada I, Hamanoue H, Terada K, Tohma T, Megarbane A, Chouery E, Abou-Ghoch J, Jalkh N, Cogulu O, Ozkinay F, Horie K, Takeda J, Furuichi T, Ikegawa S, Nishiyama K, Miyatake S, Nishimura A, Mizuguchi T, Niikawa N, Hirahara F, Kaname T, Yoshiura K, Tsurusaki Y, Doi H, Miyake N, Furukawa T, Matsumoto N, Saitsu H.
SMOC1 is essential for ocular and limb development in humans and mice. Am J Hum Genet. 2011 Jan 7;88(1):30-41.

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55- Corbani S, Chouery E, Eid B, Jalkh N, Ghoch JA, Mégarbané A.
Mild Campomelic Dysplasia: Report on a Case and Review. Mol Syndromol. 2011 Jan;1(4):163-168.

56- Haddad NM, Ente D, Chouery E, Jalkh N, Mehawej C, Khoueir Z, Pingault V, Mégarbané A.
Molecular Study of Three Lebanese and Syrian Patients with Waardenburg Syndrome and Report of Novel Mutations in the EDNRB and MITF Genes. Mol Syndromol. 2011 Jan;1(4):169-175.

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How many entities exist for the spectrum of disorders associated with brachydactyly, syndactyly, short stature, microcephaly, and intellectual disability? Am J Med Genet A.

58- Mégarbané A, Chouery E, Mignon-Ravix C, El Sabbagh S, Corbani S, Ghoch JA, Jalkh N, Mehawej C, Lévy N, Villard L.
Ambiguous genitalia, microcephaly, seizures, bone malformations, and early death: a distinct MCA/MR syndrome. Am J Med Genet A. 2011 May;155A(5):1147-51.

59- Bernard G, Chouery E, Putorti ML, Tétreault M, Takanohashi A, Carosso G, Clément I, Boespflug-Tanguy O, Rodriguez D, Delague V, Abou Ghoch J, Jalkh N,Dorboz I, Fribourg S, Teichmann M, Megarbane A, Schiffmann R, Vanderver A, Brais B.
Mutations of POLR3A Encoding a Catalytic Subunit of RNA Polymerase Pol III Cause a Recessive Hypomyelinating Leukodystrophy. Am J Hum Genet. 2011 Sep 9;89(3):415-23

60- Chouery E, Abou-Ghoch J, Corbani S, El Ali N, Korban R, Salem N, Castro C, Klayme S, Azoury-Abou Rjeily M, Khoury-Matar R, Debo G, Germanos-Haddad M, Delague V, Lefranc G, Mégarbané A.
A novel deletion in ZBTB24 in a Lebanese family with immunodeficiency, centromeric instability, and facial anomalies syndrome type 2. Clin Genet. 2011 Sep 9. doi: 10.1111/j.1399-0004.2011.01783.x.

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Molecular screening of MECP2 gene in a cohort of Lebanese patients suspected with Rett syndrome: report on a mild case with a novel indel mutation. J Intellect Disabil Res. 2011 Sep 29. doi: 10.1111/j.1365-2788.2011.01479.x.

62- Maalouf D, Mégarbané H, Chouery E, Nasr J, Badens C, Lacoste C, Grzeschik KH, Mégarbané A. A novel mutation in the PORCN gene underlying a case of almost unilateral focal dermal hypoplasia. Arch Dermatol. 2012 Jan;148(1):85-8. PubMed PMID: 22250236.

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Cardiorenal syndrome in hypertensive rats: microalbuminuria, inflammation and ventricular hypertrophy. Physiol Res. 2012 Mar 6;61(1):13-24. Epub 2011 Dec 20.PubMed PMID: 22188107.

64- Mehawej C, Chouery E, Maalouf D, Baujat G, Le Merrer M, Cormier-Daire V, Mégarbané A.
Identification of a novel causative mutation in the ROR2 gene in a Lebanese family with a mild form of recessive Robinow syndrome. Eur J Med Genet. 2012 Feb;55(2):103-8. Epub 2011 Nov 27. PubMed PMID: 22178368.

65- Mégarbané A, Hanna N, Chouery E, Jalkh N, Mehawej C, Boileau C.
Marfanoid habitus, inguinal hernia, advanced bone age, and distinctive facial features: a new collagenopathy? Am J Med Genet A. 2012 May;158A(5):1185-9.

66- Haddad NM, Waked N, Bejjani R, Khoueir Z, Chouery E, Corbani S, Mégarbané A.
Clinical and molecular findings in three Lebanese families with Bietti crystalline dystrophy: report on a novel mutation. Mol Vis. 2012;18:1182-8. Epub 2012 May 5.

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Prevalance of BRCA1 and BRCA2 mutations in familial breast cancer patients in Lebanon. Hered Cancer Clin Pract. 2012 Jun 19;10(1):7.

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Prices and distinctions

2019 : Premier prix du RARD (Recent Advances in Rare Diseases) pour ma présentation orale: The added value of WES reanalysis in the field of genetic diagnosis: Lessons learned from 200 exomes in the Lebanese population.
2017 : prix d'excellence scientifique du LAAS (Lebanese Association for advancement in Sciences)
2015 : 2ème prix de jeunes chercheurs de « La Société des membres de la Légion d'honneur » (SMLH), en partenariat avec l'Institut français du Liban avec une mention honorable.