FONDATION maladies rares

PUBLICATIONS DES PROJETS SOUTENUS PAR LA FONDATION

2017

Title	Author(s)	Source
Sclérodermie systémique et difficultés professionnelles: résultats d'une enquête prospective	N.Peres, S.Morell-Dubois, E.Hachulla, P.Y.Hatron, A.Duhamel, D.Godard, A.S.Tellart-Mamert, M.Lambert, D.Launay, S.Fantoni-Quinton	LA REVUE DE MEDECINE INTERNE 2017 OCT. IN PRESS
Heterogeneity of FHF1 related phenotype: Novel case with early onset severe attacks of apnea, partial mitochondrial respiratory chain complex II deficiency, neonatal onset seizures without neurodegeneration	Villeneuve, N (Villeneuve, Nathalie); Abidi, A (Abidi, Affef); Cacciagli, P (Cacciagli, Pierre); Mignon-Ravix, C (Mignon-Ravix, Cecile); Chabrol, B (Chabrol, Brigitte); Villard, L (Villard, Laurent); Milh, M (Milh, Mathieu)	EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY Volume: 21 Issue: 5 Pages: 783-786 Published: SEP 2017
Molecular, clinical and neuropsychological study in 31 patients with Kabuki syndrome and KMT2D mutations	Lehman, N (Lehman, N.); Mazery, AC (Mazery, A. C.); Visier, A (Visier, A.); Baumann, C (Baumann, C.); Lachesnais, D (Lachesnais, D.); Capri, Y (Capri, Y.); Toutain, A (Toutain, A.); Odent, S (Odent, S.); Mikaty, M (Mikaty, M.); Goizet, C (Goizet, C.); Taupiac, E (Taupiac, E.); Jacquemont, ML (Jacquemont, M. L.); Sanchez, E (Sanchez, E.); Schaefer, E (Schaefer, E.); Gatinois, V (Gatinois, V.); Faivre, L (Faivre, L.); Minot, D (Minot, D.); Kayirangwa, H (Kayirangwa, H.); Sang, KHLQ (Sang, K. -H. L. Q.); Boddaert, N (Boddaert, N.); Bayard, S (Bayard, S.); Lacombe, D (Lacombe, D.); Moutton, S (Moutton, S.); Touitou, I (Touitou, I.); Rio, M (Rio, M.); Amiel, J (Amiel, J.); Lyonnet, S (Lyonnet, S.); Sanlaville, D (Sanlaville, D.); Picot, MC (Picot, M. C.); Genevieve, D (Genevieve, D.)	CLINICAL GENETICS Volume: 92 Issue: 3 Pages: 298-305 DOI: 10.1111/cge.13010 Published: SEP 2017
Blepharocheilodontic syndrome is a CDH1 pathway-related disorder due to mutations in CDH1 and CTNND1	Ghoumid, J (Ghoumid, Jamal); Stichelbout, M (Stichelbout, Morgane); Jourdain, AS (Jourdain, Anne-Sophie); Frenois, F (Frenois, Frederic); Lejeune-Dumoulin, S (Lejeune-Dumoulin, Sophie); Alex-Cordier, MP (Alex-Cordier, Marie-Pierre); Lebrun, M (Lebrun, Marine); Guerreschi, P (Guerreschi, Pierre); Duquennoy-Martinot, V (Duquennoy-Martinot, Veronique); Vinchon, M (Vinchon, Matthieu); Ferri, J (Ferri, Joel); Jung, M (Jung, Matthieu); Vicaire, S (Vicaire, Serge); Vanlerberghe, C (Vanlerberghe, Clemence); Escande, F (Escande, Fabienne); Petit, F (Petit, Florence); Manouvrier-Hanu, S (Manouvrier-Hanu, Sylvie)	GENETICS IN MEDICINE Volume: 19 Issue: 9 Pages: 1013-1021 DOI: 10.1038/gim.2017.11 Published: SEP 2017
A novel de novo mutation in MYT1, the unique OAVS gene identified so far	Berenguer, M (Berenguer, Marie); Tingaud-Sequeira, A (Tingaud-Sequeira, Angele); Colovati, M (Colovati, Mileny); Melaragno, MI (Melaragno, Maria I.); Bragagnolo, S (Bragagnolo, Silvia); Perez, ABA (Perez, Ana B. A.); Arveiler, B (Arveiler, Benoit); Lacombe, D (Lacombe, Didier); Rooryck, C (Rooryck, Caroline)	EUROPEAN JOURNAL OF HUMAN GENETICS Volume: 25 Issue: 9 Pages: 1083-1086 DOI: 10.1038/ejhg.2017.101 Published: SEP 2017

Title	Author(s)	Source
Homozygous Truncating Variants in TBC1D23 Cause Pontocerebellar Hypoplasia and Alter Cortical Development	Ivanova, EL (Ivanova, Ekaterina L.); Mau-Them, FT (Mau-Them, Frederic Tran); Riazuddin, S (Riazuddin, Saima); Kahrizi, K (Kahrizi, Kimia); Laugel, V (Laugel, Vincent); Schaefer, E (Schaefer, Elise); Martin, AD (Martin, Anne de Saint); Runge, K (Runge, Karen); Iqbal, Z (Iqbal, Zafar); Spitz, MA (Spitz, Marie-Aude); Laura, M (Laura, Mary); Drouot, N (Drouot, Nathalie); Gerard, B (Gerard, Benedicte); Deleuze, JF (Deleuze, Jean-Francois); de Brouwer, APM (de Brouwer, Arjan P. M.); Razzaq, A (Razzaq, Attia); Dollfus, H (Dollfus, Helene); Assir, MZ (Assir, Muhammad Zaman); Nitchke, P (Nitchke, Patrick); Hinckelmann, MV (Hinckelmann, Maria-Victoria); Ropers, H (Ropers, Hilger); Riazuddin, S (Riazuddin, Sheikh); Najmabadi, H (Najmabadi, Hossein); van Bokhoven, H (van Bokhoven, Hans); Chelly, J (Chelly, Jamel)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 101 Issue: 3 Pages: 428-440 DOI: 10.1016/j.ajhg.2017.07.010 Published: SEP 7 2017
CSNK2B splice site mutations in patients cause intellectual disability with or without myoclonic epilepsy	Poirier, K (Poirier, Karine); Hubert, L (Hubert, Laurence); Viot, G (Viot, Geraldine); Rio, M (Rio, Marlene); Billuart, P (Billuart, Pierre); Besmond, C (Besmond, Claude); Bienvenu, T (Bienvenu, Thierry)	HUMAN MUTATION Volume: 38 Issue: 8 Pages: 932-941 DOI: 10.1002/humu.23270 Published: AUG 2017
Implanted Dental Pulp Cells Fail to Induce Regeneration in Partial Pulpotomies	F. Mangione, M. EzEldeen, C. Bardet, J. Lesieur, M. Bonneau, F. Decup, B. Salmon, R. Jacobs, C. Chaussain, and S. Opsahl-Vital	JOURNAL OF DENTAL RESEARCH 2017 Aug 1:22034517725523. doi: 10.1177/0022034517725523
SPINK2 deficiency causes infertility by inducing sperm defects in heterozygotes and azoospermia inhomozygotes	Kherraf, ZE (Kherraf, Zine-Eddine); Christou-Kent, M (Christou-Kent, Marie); Karaouzene, T (Karaouzene, Thomas); Amiri-Yekta, A (Amiri-Yekta, Amir); Martinez, G (Martinez, Guillaume); Vargas, AS (Vargas, Alexandra S.); Lambert, E (Lambert, Emeline); Borel, C (Borel, Christelle); Dorphin, B (Dorphin, Beatrice); Aknin-Seifer, I (Aknin-Seifer, Isabelle); Mitchell, MJ (Mitchell, Michael J.); Metzler-Guillemain, C (Metzler-Guillemain, Catherine); Escoffier, J (Escoffier, Jessica); Nef, S (Nef, Serge); Grepillat, M (Grepillat, Mariane); Thierry-Mieg, N (Thierry-Mieg, Nicolas); Satre, V (Satre, Veronique); Bailly, M (Bailly, Marc); Boitrelle, F (Boitrelle, Florence); Pernet-Gallay, K (Pernet-Gallay, Karin); Hennebicq, S (Hennebicq, Sylviane); Faure, J (Faure, Julien); Bottari, SP (Bottari, Serge P.); Coutton, C (Coutton, Charles); Ray, PF (Ray, Pierre F.); Arnoult, C (Arnoult, Christophe)	EMBO MOLECULAR MEDICINE Volume: 9 Issue: 8 Pages: 1132-1149 DOI: 10.15252/emmm.201607461 Published: AUG 2017
HSPB8 haploinsufficiency causes dominant adult-onset axial and distal myopathy	Echaniz-Laguna, A (Echaniz-Laguna, Andoni); Lornage, X (Lornage, Xaviere); Lannes, B (Lannes, Beatrice); Schneider, R (Schneider, Raphael); Bierry, G (Bierry, Guillaume); Dondaine, N (Dondaine, Nicolas); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Bohm, J (Bohm, Johann); Thompson, J (Thompson, Julie); Laporte, J (Laporte, Jocelyn); Biancalana, V (Biancalana, Valerie)	ACTA NEUROPATHOLOGICA Volume: 134 Issue: 1 Pages: 163-165 DOI: 10.1007/s00401-017-1724-8 Published: JUL 2017
Macrothrombocytopenia and dense granule deficiency associated with FLI1 variants: ultrastructural and pathogenic features	Saultier, P (Saultier, Paul); Vidal, L (Vidal, Lea); Canault, M (Canault, Matthias); Bernot, D (Bernot, Denis); Falaise, C (Falaise, Celine); Pouymayou, C (Pouymayou, Catherine); Bordet, JC (Bordet, Jean-Claude); Saut, N (Saut, Noemie); Rostan, A (Rostan, Agathe); Baccini, V (Baccini, Veronique); Peiretti, F (Peiretti, Franck); Favier, M (Favier, Marie); Lucca, P (Lucca, Pauline); Deleuze, JF (Deleuze, Jean-Francois); Olaso, R (Olaso, Robert); Boland, A (Boland, Anne); Morange, PE (Morange, Pierre Emmanuel); Gachet, C (Gachet, Christian); Malergue, F (Malergue, Fabrice); Faure, S (Faure, Sixtine); Eckly, A (Eckly, Anita); Tregouet, DA (Tregouet, David-Alexandre); Poggi, M (Poggi, Marjorie); Alessi, MC (Alessi, Marie-Christine)	HAEMATOLOGICA Volume: 102 Issue: 6 Pages: 1006-1016 DOI: 10.3324/haematol.2016.153577 Published: JUN 2017
Fifteen years of research on oral-facial-digital syndromes: from 1 to 16 causal genes	Bruel, AL (Bruel, Ange-Line); Franco, B (Franco, Brunella); Duffourd, Y (Duffourd, Yannis); Thevenon, J (Thevenon, Julien); Jego, L (Jego, Laurence); Lopez, E (Lopez, Estelle); Deleuze, JF (Deleuze, Jean-Francois); Doummar, D (Doummar, Diane); Giles, RH (Giles, Rachel H.); Johnson, CA (Johnson, Colin A.); Huynen, MA (Huynen, Martijn A.); Chevrier, V (Chevrier, Veronique); Burglen, L (Burglen, Lydie); Morleo, M (Morleo, Manuela); Desguerres, I (Desguerres, Isabelle); Pierquin, G (Pierquin, Genevieve); Doray, B (Doray, Berenice); Gilbert-Dussardier, B (Gilbert-Dussardier, Brigitte); Reversade, B (Reversade, Bruno); Steichen-Gersdorf, E (Steichen-Gersdorf, Elisabeth); Baumann, C (Baumann, Clarisse); Panigrahi, I (Panigrahi, Inusha); Fargeot-Espaliat, A (Fargeot-Espaliat, Anne); Dieux, A (Dieux, Anne); David, A (David, Albert); Goldenberg, A (Goldenberg, Alice); Bongers, E (Bongers, Ernie); Gaillard, D (Gaillard, Dominique); Argente, J (Argente, Jesus); Aral, B (Aral, Bernard); Gigot, N (Gigot, Nadege); St-Onge, J (St-Onge, Judith); Birnbaum, D (Birnbaum, Daniel); Phadke, SR (Phadke, Shubha R.); Cormier-Daire, V (Cormier-Daire, Valerie); Eguether, T (Eguether, Thibaut); Pazour, GJ (Pazour, Gregory J.); Herranz-Perez, V (Herranz-Perez, Vicente); Goldstein, JS (Goldstein, Jaclyn S.); Pasquier, L (Pasquier, Laurent); Loget, P (Loget, Philippe); Saunier, S (Saunier, Sophie); Megarbane, A (Megarbane, Andre); Rosnet, O (Rosnet, Olivier); Leroux, MR (Leroux, Michel R.); Wallingford, JB (Wallingford, John B.); Blacque, OE (Blacque, Oliver E.); Nachury, MV (Nachury, Maxence V.); Attie-Bitach, T (Attie-Bitach, Tania); Riviere, JB (Riviere, Jean-Baptiste); Faivre, L (Faivre, Laurence); Thauvin-Robinet, C (Thauvin-Robinet, Christel)	JOURNAL OF MEDICAL GENETICS Volume: 54 Issue: 6 Pages: 371-380 DOI: 10.1136/jmedgenet-2016-104436 Published: JUN 2017

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Copy Number Variations Found in Patients with a Corpus Callosum Abnormality and Intellectual Disability	Heide, S (Heide, Solveig); Keren, B (Keren, Boris); de Villemeur, TB (de Villemeur, Thierry Billette); Chantot-Bastaraud, S (Chantot-Bastaraud, Sandra); Depienne, C (Depienne, Christel); Nava, C (Nava, Caroline); Mignot, C (Mignot, Cyril); Jacquette, A (Jacquette, Aurelia); Fonteneau, E (Fonteneau, Eric); Lejeune, E (Lejeune, Elodie); Mach, C (Mach, Corinne); Marey, I (Marey, Isabelle); Whalen, S (Whalen, Sandra); Lacombe, D (Lacombe, Didier); Naudion, S (Naudion, Sophie); Rooryck, C (Rooryck, Caroline); Toutain, A (Toutain, Annick); Le Caignec, C (Le Caignec, Cedric); Haye, D (Haye, Damien); Olivier-Faivre, L (Olivier-Faivre, Laurence); Masurel-Paulet, A (Masurel-Paulet, Alice); Thauvin-Robinet, C (Thauvin-Robinet, Christel); Lesne, F (Lesne, Fabien); Faudet, A (Faudet, Anne); Ville, D (Ville, Dorothee); des Portes, V (des Portes, Vincent); Sanlaville, D (Sanlaville, Damien); Siffroi, JP (Siffroi, Jean-Pierre); Moutard, ML (Moutard, Marie-Laure); Heron, D (Heron, Delphine)	JOURNAL OF PEDIATRICS Volume: 185 Pages: 160-+ DOI: 10.1016/j.jpeds.2017.02.023 Published: JUN 2017
NADPH Oxidase-1 Plays a Key Role in Keratinocyte Responses to UV Radiation and UVB-Induced Skin Carcinogenesis	Raad, H (Raad, Houssam); Serrano-Sanchez, M (Serrano-Sanchez, Martin); Harfouche, G (Harfouche, Ghida); Mahfouf, W (Mahfouf, Walid); Bortolotto, D (Bortolotto, Doriane); Bergeron, V (Bergeron, Vanessa); Kasraian, Z (Kasraian, Zeinab); Dousset, L (Dousset, Lea); Hosseini, M (Hosseini, Mohsen); Taieb, A (Taieb, Alain); Rezvani, HR (Rezvani, Hamid Reza)	JOURNAL OF INVESTIGATIVE DERMATOLOGY Volume: 137 Issue: 6 Pages: 1311-1321 DOI: 10.1016/j.jid.2016.12.027 Published: JUN 2017
Acquired TET2 mutation in one patient with familial platelet disorder with predisposition to AML led to the development of pre-leukaemic clone resulting in T2-ALL and AML-M0	Manchev, VT (Manchev, Vladimir T.); Bouzid, H (Bouzid, Hind); Antony-Debre, I (Antony-Debre, Ileana); Leite, B (Leite, Betty); Meurice, G (Meurice, Guillaume); Droin, N (Droin, Nathalie); Prebet, T (Prebet, Thomas); Costello, RT (Costello, Regis T.); Vainchenker, W (Vainchenker, William); Plo, I (Plo, Isabelle); Diop, M (Diop, M'boyba); Macintyre, E (Macintyre, Elizabeth); Asnafi, V (Asnafi, Vahid); Favier, R (Favier, Remi); Baccini, V (Baccini, Veronique); Raslova, H (Raslova, Hana)	JOURNAL OF CELLULAR AND MOLECULAR MEDICINE Volume: 21 Issue: 6 Pages: 1237-1242 DOI: 10.1111/jcmm.13051 Published: JUN 2017
The Subjective Experience of Patients Diagnosed with Hereditary Hemorrhagic Telangiectasia: a Qualitative Study	Geerts, L (Geerts, Laura); Fantini-Hauwel, C (Fantini-Hauwel, Carole); Brugalle, E (Brugalle, Elodie); Boute, O (Boute, Odile); Frenois, F (Frenois, Frederic); Defrance, L (Defrance, Lydie); Manouvrier-Hanu, S (Manouvrier-Hanu, Sylvie); Petit, F (Petit, Florence); Antoine, P (Antoine, Pascal)	JOURNAL OF GENETIC COUNSELING Volume: 26 Issue: 3 Pages: 612-619 DOI: 10.1007/s10897-016-0033-z Published: JUN 2017
A no-stop mutation in MAGEB4 is a possible cause of rare X-linked azoospermia and oligozoospermia in a consanguineous Turkish family	Okutman, O (Okutman, Ozlem); Muller, J (Muller, Jean); Skory, V (Skory, Valerie); Garnier, JM (Garnier, Jean Marie); Gaucherot, A (Gaucherot, Angeline); Baert, Y (Baert, Yoni); Lamour, V (Lamour, Valerie); Serdarogullari, M (Serdarogullari, Munevver); Gultomruk, M (Gultomruk, Meral); Ropke, A (Roepke, Albrecht); Kliesch, S (Kliesch, Sabine); Herbepin, V (Herbepin, Viviana); Aknin, I (Aknin, Isabelle); Benkhalifa, M (Benkhalifa, Moncef); Teletin, M (Teletin, Marius); Bakircioglu, E (Bakircioglu, Emre); Goossens, E (Goossens, Ellen); Charlet-Berguerand, N (Charlet-Berguerand, Nicolas); Bahceci, M (Bahceci, Mustafa); Tuttelmann, F (Tuettelmann, Frank); Viville, S (Viville, STephane)	JOURNAL OF ASSISTED REPRODUCTION AND GENETICS Volume: 34 Issue: 5 Pages: 683-694 DOI: 10.1007/s10815-017-0900-z Published: MAY 2017
A homozygous mutation of GNRHR in a familial case diagnosed with polycystic ovary syndrome	Caburet, S (Caburet, Sandrine); Fruchter, RB (Fruchter, Ronit Beck); Legois, B (Legois, Berangere); Fellous, M (Fellous, Marc); Shalev, S (Shalev, Stavit); Veitia, RA (Veitia, Reiner A.)	EUROPEAN JOURNAL OF ENDOCRINOLOGY Volume: 176 Issue: 5 Pages: K9-K14 DOI: 10.1530/EJE-16-0968 Published: MAY 2017
EDNRB mutations causeWaardenburg syndrome type II in the heterozygous state	Issa, S (Issa, Sarah); Bondurand, N (Bondurand, Nadege); Faubert, E (Faubert, Emmanuelle); Poisson, S (Poisson, Sylvain); Lecerf, L (Lecerf, Laure); Nitschke, P (Nitschke, Patrick); Deggouj, N (Deggouj, Naima); Loundon, N (Loundon, Natalie); Jonard, L (Jonard, Laurence); David, A (David, Albert); Sznajer, Y (Sznajer, Yves); Blanchet, P (Blanchet, Patricia); Marlin, S (Marlin, Sandrine); Pingault, V (Pingault, Veronique)	HUMAN MUTATION Volume: 38 Issue: 5 Pages: 581-593 DOI: 10.1002/humu.23206 Published: MAY 2017
Loss of Function of KCNC1 is associated with intellectual disability without seizures	Poirier, K (Poirier, Karine); Viot, G (Viot, Geraldine); Lombardi, L (Lombardi, Laura); Jauny, C (Jauny, Clemence); Billuart, P (Billuart, Pierre); Bienvenu, T (Bienvenu, Thierry)	EUROPEAN JOURNAL OF HUMAN GENETICS Volume: 25 Issue: 5 Pages: 560-564 DOI: 10.1038/ejhg.2017.3 Published: MAY 2017

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Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU	Depienne, C (Depienne, Christel); Nava, C (Nava, Caroline); Keren, B (Keren, Boris); Heide, S (Heide, Solveig); Rastetter, A (Rastetter, AgnSs); Passemard, S (Passemard, Sandrine); Chantot-Bastaraud, S (Chantot-Bastaraud, Sandra); Moutard, ML (Moutard, Marie-Laure); Agrawal, PB (Agrawal, Pankaj B.); VanNoy, G (VanNoy, Grace); Stoler, JM (Stoler, Joan M.); Amor, DJ (Amor, David J.); de Villemeur, TB (de Villemeur, Thierry Billette); Doummar, D (Doummar, Diane); Alby, C (Alby, Caroline); Cormier-Daire, V (Cormier-Daire, Valerie); Garel, C (Garel, Catherine); Marzin, P (Marzin, Pauline); Scheidecker, S (Scheidecker, Sophie); de Saint-Martin, A (de Saint-Martin, Anne); Hirsch, E (Hirsch, Edouard); Korff, C (Korff, Christian); Bottani, A (Bottani, Armand); Faivre, L (Faivre, Laurence); Verloes, A (Verloes, Alain); Orzechowski, C (Orzechowski, Christine); Burglen, L (Burglen, Lydie); Leheup, B (Leheup, Bruno); Roume, J (Roume, Joelle); Andrieux, J (Andrieux, Joris); Sheth, F (Sheth, Frenny); Datar, C (Datar, Chaitanya); Parker, MJ (Parker, Michael J.); Pasquier, L (Pasquier, Laurent); Odent, S (Odent, Sylvie); Naudion, S (Naudion, Sophie); Delrue, MA (Delrue, Marie-Ange); Le Caignec, C (Le Caignec, Cedric); Vincent, M (Vincent, Marie); Isidor, B (Isidor, Bertrand); Renaldo, F (Renaldo, Florence); Stewart, F (Stewart, Fiona); Toutain, A (Toutain, Annick); Koehler, U (Koehler, Udo); Hackl, B (Hackl, Birgit); von Stulpnagel, C (von Stulpnagel, Celina); Kluger, G (Kluger, Gerhard); Moller, RS (Moller, Rikke S.); Pal, D (Pal, Deb); Jonson, T (Jonson, Tord); Soller, M (Soller, Maria); Verbeek, NE (Verbeek, Nienke E.); van Haelst, MM (van Haelst, Mieke M.); de Kovel, C (de Kovel, Carolien); Koeleman, B (Koeleman, Bobby); Monroe, G (Monroe, Glen); van Haaften, G (van Haaften, Gijs); Study, D (Study, D. D. D.); Attie-Bitach, T (Attie-Bitach, Tania); Boutaud, L (Boutaud, Lucile); Heron, D (Heron, Delphine); Mignot, C (Mignot, Cyril)	HUMAN GENETICS Volume: 136 Issue: 4 Pages: 463-479 DOI: 10.1007/s00439-017-1772-0 Published: APR 2017
ORAI1 Mutations with Distinct Channel Gating Defects in Tubular Aggregate Myopathy	Bohm, J (Boehm, Johann); Bulla, M (Bulla, Monica); Urquhart, JE (Urquhart, Jill E.); Malfatti, E (Malfatti, Edoardo); Williams, SG (Williams, Simon G.); O'Sullivan, J (O'Sullivan, James); Szlauer, A (Szlauer, Anastazja); Koch, C (Koch, Catherine); Baranello, G (Baranello, Giovanni); Mora, M (Mora, Marina); Ripolone, M (Ripolone, Michela); Violano, R (Violano, Raffaella); Moggio, M (Moggio, Maurizio); Kingston, H (Kingston, Helen); Dawson, T (Dawson, Timothy); DeGoede, CG (DeGoede, Christian G.); Nixon, J (Nixon, John); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Romero, N (Romero, Norma); Newman, WG (Newman, William G.); Demaurex, N (Demaurex, Nicolas); Laporte, J (Laporte, Jocelyn)	HUMAN MUTATION Volume: 38 Issue: 4 Pages: 426-438 DOI: 10.1002/humu.23172 Published: APR 2017
Dihydropyridine receptor (DHPR, CACNA1S) congenital myopathy	Schartner, V (Schartner, Vanessa); Romero, NB (Romero, Norma B.); Donkervoort, S (Donkervoort, Sandra); Treves, S (Treves, Susan); Munot, P (Munot, Pinki); Pierson, TM (Pierson, Tyler Mark); Dabaj, I (Dabaj, Ivana); Malfatti, E (Malfatti, Edoardo); Zaharieva, IT (Zaharieva, Irina T.); Zorzato, F (Zorzato, Francesco); Neto, OA (Neto, Osorio Abath); Brochier, G (Brochier, Guy); Lornage, X (Lornage, Xaviere); Eymard, B (Eymard, Bruno); Taratuto, AL (Taratuto, Ana Lia); Bohm, J (Boehm, Johann); Gonorazky, H (Gonorazky, Hernan); Ramos-Platt, L (Ramos-Platt, Leigh); Feng, L (Feng, Lucy); Phadke, R (Phadke, Rahul); Bharucha-Goebel, DX (Bharucha-Goebel, Diana X.); Sumner, CJ (Sumner, Charlotte Jane); Bui, MT (Bui, Mai Thao); Lacene, E (Lacene, Emmanuelle); Beuvin, M (Beuvin, Maud); Labasse, C (Labasse, Clemence); Dondaine, N (Dondaine, Nicolas); Schneider, R (Schneider, Raphael); Thompson, J (Thompson, Julie); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Matthews, E (Matthews, Emma); Pakleza, AN (Pakleza, Aleksandra Nadaj); Sewry, CA (Sewry, Caroline A.); Biancalana, V (Biancalana, Valerie); Quijano-Roy, S (Quijano-Roy, Susana); Muntoni, F (Muntoni, Francesco); Fardeau, M (Fardeau, Michel); Bonnemann, CG (Boennemann, Carsten G.); Laporte, J (Laporte, Jocelyn)	ACTA NEUROPATHOLOGICA Volume: 133 Issue: 4 Pages: 517-533 DOI: 10.1007/s00401-016-1656-8 Published: APR 2017
Recurrent KIF2A mutations are responsible for classic lissencephaly	Cavallin, M (Cavallin, Mara); Bijlsma, EK (Bijlsma, Emilia K.); El Morjani, A (El Morjani, Adrienne); Moutton, S (Moutton, Sebastien); Peeters, EAJ (Peeters, Els A. J.); Maillard, C (Maillard, Camille); Pedespan, JM (Pedespan, Jean Michel); Guerrot, AM (Guerrot, Anne-Marie); Drouin-Garaud, V (Drouin-Garaud, Valerie); Coubes, C (Coubes, Christine); Genevieve, D (Genevieve, David); Bole-Feysot, C (Bole-Feysot, Christine); Fourrage, C (Fourrage, Cecile); Steffann, J (Steffann, Julie); Bahi-Buisson, N (Bahi-Buisson, Nadia)	NEUROGENETICS Volume: 18 Issue: 2 Pages: 73-79 DOI: 10.1007/s10048-016-0499-8 Published: APR 2017
Neuropathological Hallmarks of Brain Malformations in Extreme Phenotypes Related to DYNC1H1 Mutations	Laquerriere, A (Laquerriere, Annie); Maillard, C (Maillard, Camille); Cavallin, M (Cavallin, Mara); Chapon, F (Chapon, Francoise); Marguet, F (Marguet, Florent); Molin, A (Molin, Arnaud); Sigaudy, S (Sigaudy, Sabine); Blouet, M (Blouet, Marie); Benoist, G (Benoist, Guillaume); Fernandez, C (Fernandez, Carla); Poirier, K (Poirier, Karine); Chelly, J (Chelly, Jamel); Thomas, S (Thomas, Sophie); Bahi-Buisson, N (Bahi-Buisson, Nadia)	JOURNAL OF NEUROPATHOLOGY AND EXPERIMENTAL NEUROLOGY Volume: 76 Issue: 3 Pages: 195-205 DOI: 10.1093/jnen/nlw124 Published: MAR 2017

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Recessive MYPN Mutations Cause Cap Myopathy with Occasional Nemaline Rods	Lornage, X (Lornage, Xaviere); Malfatti, E (Malfatti, Edoardo); Cheraud, C (Cheraud, Chrystel); Schneider, R (Schneider, Raphael); Biancalana, V (Biancalana, Valerie); Cuisset, JM (Cuisset, Jean-Marie); Garibaldi, M (Garibaldi, Matteo); Eymard, B (Eymard, Bruno); Fardeau, M (Fardeau, Michel); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Thompson, J (Thompson, Julie); Carlier, RY (Carlier, Robert-Yves); Bohm, J (Bohm, Johann); Romero, NB (Romero, Norma B.); Laporte, J (Laporte, Jocelyn)	ANNALS OF NEUROLOGY Volume: 81 Issue: 3 Pages: 467-473 Published: MAR 2017
Prenatal and Postnatal Presentations of Corpus Callosum Agenesis with Polymicrogyria Caused By EGP5 Mutation	Maillard, C (Maillard, Camille); Cavallin, M (Cavallin, Mara); Piquand, K (Piquand, Kevin); Philbert, M (Philbert, Marion); Bault, JP (Bault, Jean Philippe); Millischer, AE (Millischer, Anne Elodie); Moshous, D (Moshous, Despina); Rio, M (Rio, Marlene); Gitiaux, C (Gitiaux, Cyril); Boddaert, N (Boddaert, Nathalie); Masson, C (Masson, Cecile); Thomas, S (Thomas, Sophie); Bahi-Buisson, N (Bahi-Buisson, Nadia)	AMERICAN JOURNAL OF MEDICAL GENETICS PART A Volume: 173 Issue: 3 Pages: 706-711 DOI: 10.1002/ajmg.a.38061 Published: MAR 2017
Clinical phenotypes and outcomes of heritable and sporadic pulmonary veno-occlusive disease: a population-based study	Montani, D (Montani, David); Girerd, B (Girerd, Barbara); Jais, X (Jais, Xavier); Levy, M (Levy, Marilyne); Amar, D (Amar, David); Savale, L (Savale, Laurent); Dorfmller, P (Dorfmller, Peter); Seferian, A (Seferian, Andrei); Lau, EM (Lau, Edmund M.); Eyries, M (Eyries, Melanie); Le Pavec, J (Le Pavec, Jerome); Parent, F (Parent, Florence); Bonnet, D (Bonnet, Damien); Soubrier, F (Soubrier, Florent); Fadel, E (Fadel, Elie); Sitbon, O (Sitbon, Olivier); Simonneau, G (Simonneau, Gerald); Humbert, M (Humbert, Marc)	LANCET RESPIRATORY MEDICINE Volume: 5 Issue: 2 Pages: 125-134 DOI: 10.1016/S2213-2600(16)30438-6 Published: FEB 2017
The genetic make-up of ovarian development and function: the focus on the transcription factor FOXL2	Elzaiat, M (Elzaiat, M.); Todeschini, AL (Todeschini, A. -L.); Caburet, S (Caburet, S.); Veitia, RA (Veitia, R. A.)	CLINICAL GENETICS Volume: 91 Issue: 2 Special Issue: SI Pages: 173-182 DOI: 10.1111/cge.12862 Published: FEB 2017
Clinical spectrum and features of activated phosphoinositide 3-kinase delta syndrome: A large patient cohort study	Coulter, TI (Coulter, Tanya I.); Chandra, A (Chandra, Anita); Bacon, CM (Bacon, Chris M.); Babar, J (Babar, Judith); Curtis, J (Curtis, James); Screaton, N (Screaton, Nick); Goodlad, JR (Goodlad, John R.); Farmer, G (Farmer, George); Steele, CL (Steele, Cathal Laurence); Leahy, TR (Leahy, Timothy Ronan); Doffinger, R (Doffinger, Rainer); Baxendale, H (Baxendale, Helen); Bernatoniene, J (Bernatoniene, Jolanta); Edgar, JDM (Edgar, J. David M.); Longhurst, HJ (Longhurst, Hilary J.); Ehl, S (Ehl, Stephan); Speckmann, C (Speckmann, Carsten); Grimbacher, B (Grimbacher, Bodo); Sediva, A (Sediva, Anna); Milota, T (Milota, Tomas); Faust, SN (Faust, Saul N.); Williams, AP (Williams, Anthony P.); Hayman, G (Hayman, Grant); Kucuk, ZY (Kucuk, Zeynep Yesim); Hague, R (Hague, Rosie); French, P (French, Paul); Brooker, R (Brooker, Richard D); Forsyth, P (Forsyth, Peter); Herriot, R (Herriot, Richard); Cancrini, C (Cancrini, Caterina); Palma, P (Palma, Paolo); Ariganello, P (Ariganello, Paola); Conlon, N (Conlon, Niall); Feighery, C (Feighery, Conleth); Gavin, PJ (Gavin, Patrick J.); Jones, A (Jones, Alison); Imai, K (Imai, Kohsuke); Ibrahim, MAA (Ibrahim, Mohammad A); Markelj, G (Markelj, Gasper); Abinun, M (Abinun, Mario); Rieux-Laucat, F (Rieux-Laucat, Frederic); Latour, S (Latour, Sylvain); Pellier, I (Pellier, Isabelle); Fischer, A (Fischer, Alain); Touzot, F (Touzot, Fabien); Casanova, JL (Casanova, Jean-Laurent); Durandy, A (Durandy, Anne); Burns, SO (Burns, Siobhan O); Savic, S (Savic, Sinisa); Kumararatne, DS (Kumararatne, D. S.); Moshous, D (Moshous, Despina); Kracker, S (Kracker, Sven); Vanhaesebroeck, B (Vanhaesebroeck, Bart); Okkenhaug, K (Okkenhaug, Klaus); Picard, C (Picard, Capucine); Nejentsev, S (Nejentsev, Sergey); Condliffe, AM (Condliffe, Alison M.); Cant, AJ (Cant, Andrew James)	JOURNAL OF ALLERGY AND CLINICAL IMMUNOLOGY Volume: 139 Issue: 2 Pages: 597-+ DOI: 10.1016/j.jaci.2016.06.021 Published: FEB 2017
Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion	Vasli, N (Vasli, Nasim); Harris, E (Harris, Elizabeth); Karamchandani, J (Karamchandani, Jason); Bareke, E (Bareke, Eric); Majewski, J (Majewski, Jacek); Romero, NB (Romero, Norma B.); Stojkovic, T (Stojkovic, Tanya); Barresi, R (Barresi, Rita); Tasfaout, H (Tasfaout, Hichem); Charlton, R (Charlton, Richard); Malfatti, E (Malfatti, Edoardo); Bohm, J (Bohm, Johann); Marini-Bettolo, C (Marini-Bettolo, Chiara); Choquet, K (Choquet, Karine); Dicaire, MJ (Dicaire, Marie-Josee); Shao, YH (Shao, Yi-Hong); Topf, A (Topf, Ana); O'Ferrall, E (O'Ferrall, Erin); Eymard, B (Eymard, Bruno); Straub, V (Straub, Volker); Blanco, G (Blanco, Gonzalo); Lochmuller, H (Lochmuller, Hanns); Brais, B (Brais, Bernard); Laporte, J (Laporte, Jocelyn); Tetreault, M (Tetreault, Martine)	BRAIN Volume: 140 Pages: 37-48 DOI: 10.1093/brain/aww257 Part: 1 Published: JAN 2017
Nonlethal CHRNA1-Related Congenital Myasthenic Syndrome with a Homozygous Null Mutation	Neto, OA (Abath Neto, Osorio); Heise, CO (Heise, Carlos Otto); Moreno, CDM (Martins Moreno, Cristiane de Araujo); Estephan, ED (Estephan, Eduardo de Paula); Mesrob, L (Mesrob, Lilia); Lechner, D (Lechner, Doris); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Oliveira, ASB (Bulle Oliveira, Acary Souza); Reed, UC (Reed, Umbertina Conti); Biancalana, V (Biancalana, Valerie); Laporte, J (Laporte, Jocelyn); Zanoteli, E (Zanoteli, Edmar)	CANADIAN JOURNAL OF NEUROLOGICAL SCIENCES Volume: 44 Issue: 1 Pages: 125-127 Published: JAN 2017

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Familial small-intestine carcinoids: Chromosomal alterations and germline inositol polyphosphate multikinase sequencing	de Mestier, L (de Mestier, Louis); Pasmant, E (Pasmant, Eric); Fleury, C (Fleury, Clemence); Brixi, H (Brixi, Hedia); Sohier, P (Sohier, Pierre); Ferona, T (Ferona, Thomas); Diebold, MD (Diebold, Marie-Daniele); Clauser, E (Clauser, Eric); Cadiot, G (Cadiot, Guillaume)	DIGESTIVE AND LIVER DISEASE Volume: 49 Issue: 1 Pages: 98-102 DOI: 10.1016/j.dld.2016.10.007 Published: JAN 2017
Mosaicism in ATP1A3-related disorders: not just a theoretical risk	Hully, M (Hully, Marie); Ropars, J (Ropars, Juliette); Hubert, L (Hubert, Laurence); Boddaert, N (Boddaert, Nathalie); Rio, M (Rio, Marlene); Bernardelli, M (Bernardelli, Mathieu); Desguerre, I (Desguerre, Isabelle); Cormier-Daire, V (Cormier-Daire, Valerie); Munnich, A (Munnich, Arnold); de Lonlay, P (de Lonlay, Pascale); Reilly, L (Reilly, Louise); Besmond, C (Besmond, Claude); Bahi-Buisson, N (Bahi-Buisson, Nadia)	NEUROGENETICS Volume: 18 Issue: 1 Pages: 23-28 DOI: 10.1007/s10048-016-0498-9 Published: JAN 2017
Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy	Ait-El-Mkadem, S (Ait-El-Mkadem, Samira); Dayem-Quere, M (Dayem-Quere, Manal); Gusic, M (Gusic, Mirjana); Chaussenot, A (Chaussenot, Annabelle); Bannwarth, S (Bannwarth, Sylvie); Francois, B (Francois, Berengere); Genin, EC (Genin, Emmanuelle C.); Fragaki, K (Fragaki, Konstantina); Volker-Touw, CLM (Volker-Touw, Catharina L. M.); Vasnier, C (Vasnier, Christelle); Serre, V (Serre, Valerie); van Gassen, KLI (van Gassen, Koen L. I.); Lespinasse, F (Lespinasse, Francoise); Richter, S (Richter, Susan); Eisenhofer, G (Eisenhofer, Graeme); Rouzier, C (Rouzier, Cecile); Mochel, F (Mochel, Fanny); De Saint-Martin, A (De Saint-Martin, Anne); Warde, MTA (Warde, Marie-Therese Abi); de Sain-van der Velde, MGM (de Sain-van der Velde, Monique G. M.); Jans, JJM (Jans, Judith J. M.); Amiel, J (Amiel, Jeanne); Avsec, Z (Avsec, Ziga); Mertes, C (Mertes, Christian); Haack, TB (Haack, Tobias B.); Strom, T (Strom, Tim); Meitinger, T (Meitinger, Thomas); Bonnen, PE (Bonnen, Penelope E.); Taylor, RW (Taylor, Robert W.); Gagneur, J (Gagneur, Julien); van Hasselt, PM (van Hasselt, Peter M.); Rotig, A (Rotig, Agnes); Delahodde, A (Delahodde, Agnes); Prokisch, H (Prokisch, Holger); Fuchs, SA (Fuchs, Sabine A.); Paquis-Flucklinger, V (Paquis-Flucklinger, Veronique)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 100 Issue: 1 Pages: 151-159 DOI: 10.1016/j.ajhg.2016.11.014 Published: JAN 5 2017
Mutations in Histone Acetylase Modifier BRPF1 Cause an Autosomal-Dominant Form of Intellectual Disability with Associated Ptosis	Mattioli, F (Mattioli, Francesca); Schaefer, E (Schaefer, Elise); Magee, A (Magee, Alex); Mark, P (Mark, Paul); Mancini, GM (Mancini, Grazia M.); Dieterich, K (Dieterich, Klaus); Von Allmen, G (Von Allmen, Gretchen); Alders, M (Alders, Marielle); Coutton, C (Coutton, Charles); van Slegtenhorst, M (van Slegtenhorst, Marjon); Vieville, G (Vieville, Gaelle); Engelen, M (Engelen, Mark); Cobben, JM (Cobben, Jan Maarten); Juusola, J (Juusola, Jane); Pujol, A (Pujol, Aurora); Mandel, JL (Mandel, Jean-Louis); Piton, A (Piton, Amelie)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 100 Issue: 1 Pages: 105-116 DOI: 10.1016/j.ajhg.2016.11.010 Published: JAN 5 2017
International Registry of Patients Carrying TGFBR1 or TGFBR2 Mutations Results of the MAC (Montalcino Aortic Consortium)	Jondeau, G (Jondeau, Guillaume); Ropers, J (Ropers, Jacques); Regalado, E (Regalado, Ellen); Braverman, A (Braverman, Alan); Evangelista, A (Evangelista, Arturo); Teixedo, G (Teixedo, Guisela); De Backer, J (De Backer, Julie); Muino-Mosquera, L (Muino-Mosquera, Laura); Naudion, S (Naudion, Sophie); Zordan, C (Zordan, Cecile); Morisaki, T (Morisaki, Takayuki); Morisaki, H (Morisaki, Hiroto); Von Kodolitsch, Y (Von Kodolitsch, Yskert); Dupuis-Girod, S (Dupuis-Girod, Sophie); Morris, SA (Morris, Shaine A.); Jeremy, R (Jeremy, Richmond); Odent, S (Odent, Sylvie); Ades, LC (Ades, Leslie C.); Bakshi, M (Bakshi, Madhura); Holman, K (Holman, Katherine); LeMaire, S (LeMaire, Scott); Milleron, O (Milleron, Olivier); Langeois, M (Langeois, Maud); Spentchian, M (Spentchian, Myrtille); Aubart, M (Aubart, Melodie); Boileau, C (Boileau, Catherine); Pyeritz, R (Pyeritz, Reed); Milewicz, DM (Milewicz, Dianna M.); Group Author(s): Montalcino Aortic Consortium	CIRCULATION-CARDIOVASCULAR GENETICS Volume: 9 Issue: 6 Pages: 548-+ DOI: 10.1161/CIRCGENETICS.116.001485 Published: DEC 2016
Whole-exome sequencing of familial cases of multiple morphological abnormalities of the sperm flagella (MMAF) reveals new DNAH1 mutations	Amiri-Yekta, A (Amiri-Yekta, Amir); Coutton, C (Coutton, Charles); Kherraf, ZE (Kherraf, Zine-Eddine); Karaouzene, T (Karaouzene, Thomas); Le Tanno, P (Le Tanno, Pauline); Sanati, MH (Sanati, Mohammad Hossein); Sabbaghian, M (Sabbaghian, Marjan); Almadani, N (Almadani, Navid); Gilani, MAS (Gilani, Mohammad Ali Sadighi); Hosseini, SH (Hosseini, Seyedeh Hanieh); Bahrami, S (Bahrami, Salahadin); Daneshipour, A (Daneshipour, Abbas); Bini, M (Bini, Maurizio); Arnoult, C (Arnoult, Christophe); Colombo, R (Colombo, Roberto); Gourabi, H (Gourabi, Hamid); Ray, PF (Ray, Pierre F.)	HUMAN REPRODUCTION Volume: 31 Issue: 12 Pages: 2872-2880 DOI: 10.1093/humrep/dew262 Published: DEC 2016
Mutations in the HECT domain of NEDD4L lead to AKT-mTOR pathway deregulation and cause periventricular nodular heterotopia	Broix, L (Broix, Loic); Jagline, H (Jagline, Helene); Ivanova, EL (Ivanova, Ekaterina L.); Schmucker, S (Schmucker, Stephane); Drouot, N (Drouot, Nathalie); Clayton-Smith, J (Clayton-Smith, Jill); Pagnamenta, AT (Pagnamenta, Alistair T.); Metcalfe, KA (Metcalfe, Kay A.); Isidor, B (Isidor, Bertrand); Louvier, UW (Louvier, Ulrike Walther); Poduri, A (Poduri, Annapurna); Taylor, JC (Taylor, Jenny C.); Tilly, P (Tilly, Peggy); Poirier, K (Poirier, Karine); Saillour, Y (Saillour, Yoann); Lebrun, N (Lebrun, Nicolas); Stemmelen, T (Stemmelen, Tristan); Rudolf, G (Rudolf, Gabrielle); Muraca, G (Muraca, Giuseppe); Saintpierre, B (Saintpierre, Benjamin); Elmorjani, A (Elmorjani, Adrienne); Moise, M (Moise, Martin); Weirauch, NB (Weirauch, Nathalie Bednarek); Guerrini, R (Guerrini, Renzo); Boland, A (Boland, Anne); Olaso, R (Olaso, Robert); Masson, C (Masson, Cecile); Tripathy, R (Tripathy, Ratna); Keays, D (Keays, David); Beldjord, C (Beldjord, Cherif); Nguyen, L (Nguyen, Laurent); Godin, J (Godin, Juliette); Kini, U (Kini, Usha); Nischke, P (Nischke, Patrick); Deleuze, JF (Deleuze, Jean-Francois); Bahi-Buisson, N (Bahi-Buisson, Nadia); Sumara, I (Sumara, Izabela); Hinckelmann, MV (Hinckelmann, Maria-Victoria); Chelly, J (Chelly, Jamel); Group Author(s): Deciphering Dev Disorders Study	NATURE GENETICS Volume: 48 Issue: 11 Pages: 1349-1358 DOI: 10.1038/ng.3676 Published: NOV 2016

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Mutations in MYT1, encoding the myelin transcription factor 1, are a rare cause of OAVS	Lopez, E (Lopez, Estelle); Berenguer, M (Berenguer, Marie); Tingaud-Sequeira, A (Tingaud-Sequeira, Angele); Marlin, S (Marlin, Sandrine); Toutain, A (Toutain, Annick); Denoyelle, F (Denoyelle, Francoise); Picard, A (Picard, Arnaud); Charron, S (Charron, Sabine); Mathieu, G (Mathieu, Guilaine); de Belvalet, H (de Belvalet, Harmony); Arveiler, B (Arveiler, Benoit); Babin, PJ (Babin, Patrick J.); Lacombe, D (Lacombe, Didier); Rooryck, C (Rooryck, Caroline)	JOURNAL OF MEDICAL GENETICS Volume: 53 Issue: 11 Pages: 752-760 DOI: 10.1136/jmedgenet-2016-103774 Published: NOV 2016
Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization	O'Grady, GL (O'Grady, Gina L.); Best, HA (Best, Heather A.); Sztal, TE (Sztal, Tamar E.); Schartner, V (Schartner, Vanessa); Sanjuan-Vazquez, T (Sanjuan-Vazquez, Tvlyriam); Donkervoort, S (Donkervoort, Sandra); Neto, OA (Neto, Osorio Abath); Sutton, RB (Sutton, Roger Bryan); Ilkovski, B (Ilkovski, Biljana); Romero, NB (Romero, Norma Beatriz); Stojkovic, T (Stojkovic, Tanya); Dastgir, J (Dastgir, Jahannaz); Waddell, LB (Waddell, Leigh B.); Boland, A (Boland, Anne); Hu, Y (Hu, Ying); Williams, C (Williams, Caitlin); Ruparelia, AA (Ruparelia, Avnika A.); Maisonobe, T (Maisonobe, Thierry); Peduto, AJ (Peduto, Anthony J.); Reddel, SW (Reddel, Stephen W.); Lek, M (Lek, Monkol); Tukiainen, T (Tukiainen, Tam); Cummings, BB (Cummings, Beryl B.); Joshi, H (Joshi, Himanshu); Nectoux, J (Nectoux, Juliette); Brammah, S (Brammah, Susan); Deleuze, JF (Deleuze, Jean-Francois); Ing, VO (Ing, Viola Oorschot); Ramm, G (Ramm, Georg); Ardicli, D (Ardicli, Didem); Nowak, KJ (Nowak, Kristen J.); Talim, B (Talim, Beril); Topaloglu, H (Topaloglu, Haluk); Laing, NG (Laing, Nigel G.); North, KN (North, Kathryn N.); MacArthur, DG (MacArthur, Daniel G.); Friant, S (Friant, Sylvie); Clarke, NF (Clarke, Nigel F.); Bryson-Richardson, RJ (Bryson-Richardson, Robert J.); Bonnemann, CG (Bonnemann, Carsten G.); Laporte, J (Laporte, Jocelyn); Cooper, ST (Cooper, Sandra T.)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 99 Issue: 5 Pages: 1086-1105 DOI: 10.1016/j.ajhg.2016.09.005 Published: NOV 3 2016
Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2-2q11.2	Henden, L (Henden, Lyndal); Freytag, S (Freytag, Saskia); Afawi, Z (Afawi, Zaid); Baldassari, S (Baldassari, Sara); Berkovic, SF (Berkovic, Samuel F.); Bisulli, F (Bisulli, Francesca); Canafoglia, L (Canafoglia, Laura); Casari, G (Casari, Giorgio); Crompton, DE (Crompton, Douglas Ewan); Depienne, C (Depienne, Christel); Gecz, J (Gecz, Jozef); Guerrini, R (Guerrini, Renzo); Helbig, I (Helbig, Ingo); Hirsch, E (Hirsch, Edouard); Keren, B (Keren, Boris); Klein, KM (Klein, Karl Martin); Labauge, P (Labauge, Pierre); LeGuern, E (LeGuern, Eric); Licchetta, L (Licchetta, Laura); Mei, D (Mei, Davide); Nava, C (Nava, Caroline); Pippucci, T (Pippucci, Tommaso); Rudolf, G (Rudolf, Gabrielle); Scheffer, IE (Scheffer, Ingrid Eileen); Striano, P (Striano, Pasquale); Tinuper, P (Tinuper, Paolo); Zara, F (Zara, Federico); Corbett, M (Corbett, Mark); Bahlo, M (Bahlo, Melanie)	HUMAN GENETICS Volume: 135 Issue: 10 Pages: 1117-1125 DOI: 10.1007/s00439-016-1700-8 Published: OCT 2016
Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea	Bauche, S (Bauche, Stephanie); O'Regan, S (O'Regan, Seana); Azuma, Y (Azuma, Yoshiteru); Laffargue, F (Laffargue, Fanny); McMacken, G (McMacken, Grace); Sternberg, D (Sternberg, Damien); Brochier, G (Brochier, Guy); Buon, C (Buon, Celine); Bouzidi, N (Bouzidi, Nassima); Topf, A (Topf, Ana); Lacene, E (Lacene, Emmanuelle); Remerand, G (Remerand, Ganaelle); Beaufrere, AM (Beaufrere, Anne-Marie); Pebrel-Richard, C (Pebrel-Richard, Celine); Thevenon, J (Thevenon, Julien); El Chehadeh-Djebbar, S (El Chehadeh-Djebbar, Salima); Faivre, L (Faivre, Laurence); Duffourd, Y (Duffourd, Yannis); Ricci, F (Ricci, Federica); Mongini, T (Mongini, Tiziana); Fiorillo, C (Fiorillo, Chiara); Astrea, G (Astrea, Guja); Burloiu, CM (Burloiu, Carmen Magdalena); Butoianu, N (Butoianu, Niculina); Sandu, C (Sandu, Carmen); Servais, L (Servais, Laurent); Bonne, G (Bonne, Gisele); Nelson, I (Nelson, Isabelle); Desguerre, I (Desguerre, Isabelle); Nougues, MC (Nougues, Marie-Christine); Boeuf, B (Boeuf, Benoit); Romero, N (Romero, Norma); Laporte, J (Laporte, Jocelyn); Boland, A (Boland, Anne); Lechner, D (Lechner, Doris); Deleuze, JF (Deleuze, Jean-Francois); Fontaine, B (Fontaine, Bertrand); Strochlic, L (Strochlic, Laure); Lochmuller, H (Lochmuller, Hanns); Eymard, B (Eymard, Bruno); Mayer, M (Mayer, Michele); Nicole, S (Nicole, Sophie)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 99 Issue: 3 Pages: 753-761 DOI: 10.1016/j.ajhg.2016.06.033 Published: SEP 1 2016
QIL1 mutation causes MICOS disassembly and early onset fatal mitochondrial encephalopathy with liver disease	Guarani, V (Guarani, Virginia); Jardel, C (Jardel, Claude); Chretien, D (Chretien, Dominique); Lombes, A (Lombes, Anne); Benit, P (Benit, Paule); Labasse, C (Labasse, Clemence); Lacene, E (Lacene, Emmanuelle); Bourillon, A (Bourillon, Agnes); Imbard, A (Imbard, Appolline); Benoist, JF (Benoist, Jean Francois); Dorboz, I (Dorboz, Imen); Gilleron, M (Gilleron, Mylene); Goetzman, ES (Goetzman, Eric S.); Gaignard, P (Gaignard, Pauline); Slama, A (Slama, Abdelhamid); Elmaleh-Berges, M (Elmaleh-Berges, Monique); Romero, NB (Romero, Norma B.); Rustin, P (Rustin, Pierre); de Baulny, HO (de Baulny, Helene Ogier); Paulo, JA (Paulo, Joao A.); Harper, JW (Harper, J. Wade); Schiff, M (Schiff, Manuel)	ELIFE Volume: 5 Article Number: e17163 DOI: 10.7554/eLife.17163 Published: SEP 13 2016
Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline	Hardies, K (Hardies, Katia); Cai, YY (Cai, Yiying); Jardel, C (Jardel, Claude); Jansen, AC (Jansen, Anna C.); Cao, M (Cao, Mian); May, P (May, Patrick); Djemie, T (Djemie, Tania); Le Camus, CH (Le Camus, Caroline Hachon); Keymolen, K (Keymolen, Kathelijn); Deconinck, T (Deconinck, Tine); Bhambhani, V (Bhambhani, Vikas); Long, C (Long, Catherine); Sajan, SA (Sajan, Samin A.); Helbig, KL (Helbig, Katherine L.); Suls, A (Suls, Arvid); Balling, R (Balling, Rudi); Helbig, I (Helbig, Ingo); De Jonghe, P (De Jonghe, Peter); Depienne, C (Depienne, Christel); De Camilli, P (De Camilli, Pietro); Weckhuysen, S (Weckhuysen, Sarah); Group Author(s): AR Working Grp; EuroEPINOMICS RES Consortium	BRAIN Volume: 139 Pages: 2420-2430 DOI: 10.1093/brain/aww180 Part: 9 Published: SEP 2016

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Mutation Update for Kabuki Syndrome Genes KMT2D and KDM6A and Further Delineation of X-Linked Kabuki Syndrome Subtype 2	Bogershausen, N (Boegershausen, Nina); Gatinois, V (Gatinois, Vincent); Riehmer, V (Riehmer, Vera); Kayserili, H (Kayserili, Huelya); Becker, J (Becker, Jutta); Thoenes, M (Thoenes, Michaela); Simsek-Kiper, PO (Simsek-Kiper, Pelin OEzlem); Barat-Houari, M (Barat-Houari, Mouna); Elcioglu, NH (Elcioglu, Nursel H.); Wieczorek, D (Wieczorek, Dagmar); Tinschert, S (Tinschert, Sigrid); Sarrabay, G (Sarrabay, Guillaume); Strom, TM (Strom, Tim M.); Fabre, A (Fabre, Aurelie); Baynam, G (Baynam, Gareth); Sanchez, E (Sanchez, Elodie); Nurnberg, G (Nuernberg, Gudrun); Altunoglu, U (Altunoglu, Umut); Capri, Y (Capri, Yline); Isidor, B (Isidor, Bertrand); Lacombe, D (Lacombe, Didier); Corsini, C (Corsini, Carole); Cormier-Daire, V (Cormier-Daire, Valerie); Sanlaville, D (Sanlaville, Damien); Giuliano, F (Giuliano, Fabienne); Le Quan Sang, KH (Le Quan Sang, Kim-Hanh); Kayirangwa, H (Kayirangwa, Honorine); Nurnberg, P (Nuernberg, Peter); Meitinger, T (Meitinger, Thomas); Boduroglu, K (Boduroglu, Koray); Zoll, B (Zoll, Barbara); Lyonnet, S (Lyonnet, Stanislas); Tzschach, A (Tzschach, Andreas); Verloes, A (Verloes, Alain); Di Donato, N (Di Donato, Nataliya); Touitou, I (Touitou, Isabelle); Netzer, C (Netzer, Christian); Li, Y (Li, Yun); Genevieve, D (Genevieve, David); Yigit, G (Yigit, Goekhan); Wollnik, B (Wollnik, Bernd)	HUMAN MUTATION Volume: 37 Issue: 9 Pages: 847-864 DOI: 10.1002/humu.23026 Published: SEP 2016
Truncating Mutations in the Adhesion G ProteinCoupled Receptor G2 Gene ADGRG2 Cause an X-Linked Congenital Bilateral Absence of Vas Deferens	Patat, O (Patat, Olivier); Pagin, A (Pagin, Adrien); Siegfried, A (Siegfried, Aurore); Mitchell, V (Mitchell, Valerie); Chassaing, N (Chassaing, Nicolas); Faguer, S (Faguer, Stanislas); Monteil, L (Monteil, Laetitia); Gaston, V (Gaston, Veronique); Bujan, L (Bujan, Louis); Courtade-Saidi, M (Courtade-Saidi, Monique); Marcelli, F (Marcelli, Francois); Lalau, G (Lalau, Guy); Rigot, JM (Rigot, Jean-Marc); Mieusset, R (Mieusset, Roger); Bieth, E (Bieth, Eric)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 99 Issue: 2 Pages: 437-442 DOI: 10.1016/j.ajhg.2016.06.012 Published: AUG 4 2016
NANS-mediated synthesis of sialic acid is required for brain and skeletal development	van Karnebeek, CDM (van Karnebeek, Clara D. M.); Bonafe, L (Bonafe, Luisa); Wen, XY (Wen, Xiao-Yan); Tarailo-Graovac, M (Tarailo-Graovac, Maja); Balzano, S (Balzano, Sara); Royer-Bertrand, B (Royer-Bertrand, Beryl); Ashikov, A (Ashikov, Angel); Garavelli, L (Garavelli, Livia); Mammi, I (Mammi, Isabella); Turolla, L (Turolla, Licia); Breen, C (Breen, Catherine); Donnai, D (Donnai, Dian); Cormier, V (Cormier, Valerie); Heron, D (Heron, Delphine); Nishimura, G (Nishimura, Gen); Uchikawa, S (Uchikawa, Shinichi); Campos-Xavier, B (Campos-Xavier, Belinda); Rossi, A (Rossi, Antonio); Hennet, T (Hennet, Thierry); Brand-Arzamendi, K (Brand-Arzamendi, Koroboshka); Rozmus, J (Rozmus, Jacob); Harshman, K (Harshman, Keith); Stevenson, BJ (Stevenson, Brian J.); Girardi, E (Girardi, Enrico); Superti-Furga, G (Superti-Furga, Giulio); Dewan, T (Dewan, Tammie); Collingridge, A (Collingridge, Alissa); Halparin, J (Halparin, Jessie); Ross, CJ (Ross, Colin J.); Van Allen, MI (Van Allen, Margot I.); Rossi, A (Rossi, Andrea); Engelke, UF (Engelke, Udo F.); Kluijtmans, LAJ (Kluijtmans, Leo A. J.); van der Heeft, E (van der Heeft, Ed); Renkema, H (Renkema, Herma); de Brouwer, A (de Brouwer, Arjan); Huijben, K (Huijben, Karin); Zijlstra, F (Zijlstra, Fokje); Heisse, T (Heisse, Thorben); Boltje, T (Boltje, Thomas); Wasserman, WW (Wasserman, Wyeth W.); Rivolta, C (Rivolta, Carlo); Unger, S (Unger, Sheila); Lefeber, DJ (Lefeber, Dirk J.); Wevers, RA (Wevers, Ron A.); Superti-Furga, A (Superti-Furga, Andrea)	NATURE GENETICS Volume: 48 Issue: 7 Pages: 777-+ DOI: 10.1038/ng.3578 Published: JUL 2016
Confirmation of TENM3 Involvement in Autosomal Recessive Colobomatous Microphthalmia	Chassaing, N (Chassaing, Nicolas); Ragge, N (Ragge, Nicola); Plaisancie, J (Plaisancie, Julie); Patat, O (Patat, Oliver); Genevieve, D (Genevieve, David); Rivier, F (Rivier, Francois); Malrieu-Eliaou, C (Malrieu-Eliaou, Claudie); Hamel, C (Hamel, Christian); Kaplan, J (Kaplan, Josseline); Calvas, P (Calvas, Patrick)	AMERICAN JOURNAL OF MEDICAL GENETICS PART A Volume: 170 Issue: 7 Pages: 1895-1898 DOI: 10.1002/ajmg.a.37667 Published: JUL 2016
LRBA deficiency with autoimmunity and early onset chronic erosive polyarthritis	Levy, E (Levy, Eva); Stolzenberg, MC (Stolzenberg, Marie-Claude); Bruneau, J (Bruneau, Julie); Breton, S (Breton, Sylvain); Neven, B (Neven, Benedicte); Sauvion, S (Sauvion, Sylvie); Zarhrate, M (Zarhrate, Mohammed); Nitschke, P (Nitschke, Patrick); Fischer, A (Fischer, Alain); Magerus-Chatinet, A (Magerus-Chatinet, Aude); Quartier, P (Quartier, Pierre); Rieux-Laucat, F (Rieux-Laucat, Frederic)	CLINICAL IMMUNOLOGY Volume: 168 Pages: 88-93 DOI: 10.1016/j.clim.2016.03.006 Published: JUL 2016
Complex mode of inheritance in holoprosencephaly revealed by whole exome sequencing	Mouden, C (Mouden, C.); Dubourg, C (Dubourg, C.); Carre, W (Carre, W.); Rose, S (Rose, S.); Quelin, C (Quelin, C.); Akloul, L (Akloul, L.); Hamdi-Roze, H (Hamdi-Roze, H.); Viot, G (Viot, G.); Salhi, H (Salhi, H.); Darnault, P (Darnault, P.); Odent, S (Odent, S.); Dupe, V (Dupe, V.); David, V (David, V.)	CLINICAL GENETICS Volume: 89 Issue: 6 Pages: 659-668 DOI: 10.1111/cge.12722 Published: JUN 2016

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The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery	Toriyama, M (Toriyama, Michinori); Lee, CJ (Lee, Chanjae); Taylor, SP (Taylor, S. Paige); Duran, I (Duran, Ivan); Cohn, DH (Cohn, Daniel H.); Bruel, AL (Bruel, Ange-Line); Tabler, JM (Tabler, Jacqueline M.); Drew, K (Drew, Kevin); Kelly, MR (Kelly, Marcus R.); Kim, S (Kim, Sukyoung); Park, TJ (Park, Tae Joo); Braun, DA (Braun, Daniela A.); Pierquin, G (Pierquin, Ghislaine); Biver, A (Biver, Armand); Wagner, K (Wagner, Kerstin); Malfroot, A (Malfroot, Anne); Panigrahi, I (Panigrahi, Inusha); Franco, B (Franco, Brunella); Al-Lami, HA (Al-Lami, Hadeel Adel); Yeung, Y (Yeung, Yvonne); Choi, YJ (Choi, Yeon Ja); Duffourd, Y (Duffourd, Yannis); Faivre, L (Faivre, Laurence); Riviere, JB (Riviere, Jean-Baptiste); Chen, J (Chen, Jiang); Liu, KJ (Liu, Karen J.); Marcotte, EM (Marcotte, Edward M.); Hildebrandt, F (Hildebrandt, Friedhelm); Thauvin-Robinet, C (Thauvin-Robinet, Christel); Krakow, D (Krakow, Deborah); Jackson, PK (Jackson, Peter K.); Wallingford, JB (Wallingford, John B.)	NATURE GENETICS Volume: 48 Issue: 6 Pages: 648-+ DOI: 10.1038/ng.3558 Published: JUN 2016
A non-sense MCM9 mutation in a familial case of primary ovarian insufficiency	Fauchereau, F (Fauchereau, F.); Shalev, S (Shalev, S.); Chervinsky, E (Chervinsky, E.); Beck-Fruchter, R (Beck-Fruchter, R.); Legois, B (Legois, B.); Fellous, M (Fellous, M.); Caburet, S (Caburet, S.); Veitia, RA (Veitia, R. A.)	CLINICAL GENETICS Volume: 89 Issue: 5 Pages: 603-607 DOI: 10.1111/cge.12736 Published: MAY 2016
De novo TUBB2B mutation causes fetal akinesia deformation sequence with microlissencephaly: An unusual presentation of tubulinopathy	Laquerriere, A (Laquerriere, Annie); Gonzales, M (Gonzales, Marie); Saillour, Y (Saillour, Yoann); Cavallin, M (Cavallin, Mara); Joye, N (Joye, Nicole); Quelin, C (Quelin, Chloe); Bidat, L (Bidat, Laurent); Dommergues, M (Dommergues, Marc); Plessis, G (Plessis, Ghislaine); Encha-Razavi, F (Encha-Razavi, Ferechte); Chelly, J (Chelly, Jamel); Bahi-Buisson, N (Bahi-Buisson, Nadia); Poirier, K (Poirier, Karine)	EUROPEAN JOURNAL OF MEDICAL GENETICS Volume: 59 Issue: 4 Pages: 249-256 DOI: 10.1016/j.ejmg.2015.12.007 Published: APR 2016
Mosaic parental germline mutations causing recurrent forms of malformations of cortical development	Zillhardt, JL (Zillhardt, Julia Lauer); Poirier, K (Poirier, Karine); Broix, L (Broix, Loic); Lebrun, N (Lebrun, Nicolas); Elmorjani, A (Elmorjani, Adrienne); Martinovic, J (Martinovic, Jelena); Saillour, Y (Saillour, Yoann); Muraca, G (Muraca, Giuseppe); Nectoux, J (Nectoux, Juliette); Bessieres, B (Bessieres, Bettina); Fallet-Bianco, C (Fallet-Bianco, Catherine); Lyonnet, S (Lyonnet, Stanislas); Dulac, O (Dulac, Olivier); Odent, S (Odent, Sylvie); Rejeb, I (Rejeb, Imen); Ben Jemaa, L (Ben Jemaa, Lamia); Rivier, F (Rivier, Francois); Pinson, L (Pinson, Lucile); Genevieve, D (Genevieve, David); Musizzano, Y (Musizzano, Yuri); Bigi, N (Bigi, Nicole); Leboucq, N (Leboucq, Nicolas); Giuliano, F (Giuliano, Fabienne); Philip, N (Philip, Nicole); Vilain, C (Vilain, Catheline); Van Bogaert, P (Van Bogaert, Patrick); Maurey, H (Maurey, Helsne); Beldjord, C (Beldjord, Cherif); Artiguenave, F (Artiguenave, Franois); Boland, A (Boland, Anne); Olaso, R (Olaso, Robert); Masson, C (Masson, Cecile); Nitschke, P (Nitschke, Patrick); Deleuze, JF (Deleuze, Jean-Francois); Bahi-Buisson, N (Bahi-Buisson, Nadia); Chelly, J (Chelly, Jamel)	EUROPEAN JOURNAL OF HUMAN GENETICS Volume: 24 Issue: 4 Pages: 611-614 DOI: 10.1038/ejhg.2015.192 Published: APR 2016
Potassium Channel Subfamily K Member 3 (KCNK3) Contributes to the Development of Pulmonary Arterial Hypertension	Antigny, F (Antigny, Fabrice); Hautefort, A (Hautefort, Aurelie); Meloche, J (Meloche, Jolyane); Belacel-Ouari, M (Belacel-Ouari, Milia); Manoury, B (Manoury, Boris); Rucker-Martin, C (Rucker-Martin, Catherine); Pechoux, C (Pechoux, Christine); Potus, F (Potus, Francois); Nadeau, V (Nadeau, Valerie); Tremblay, E (Tremblay, Eve); Ruffenach, G (Ruffenach, Gregoire); Bourgeois, A (Bourgeois, Alice); Dorfmuller, P (Dorfmueller, Peter); Breuils-Bonnet, S (Breuils-Bonnet, Sandra); Fadel, E (Fadel, Elie); Ranchoux, B (Ranchoux, Benoit); Jourdon, P (Jourdon, Philippe); Girerd, B (Girerd, Barbara); Montani, D (Montani, David); Provencher, S (Provencher, Steeve); Bonnet, S (Bonnet, Sebastien); Simonneau, G (Simonneau, Gerald); Humbert, M (Humbert, Marc); Perros, F (Perros, Frederic)	CIRCULATION Volume: 133 Issue: 14 Pages: 1371-1385 DOI: 10.1161/CIRCULATIONAHA.115.020951 Published: APR 5 2016
Whole-exome sequencing improves the diagnosis yield in sporadic infantile spasm syndrome	Dimassi, S (Dimassi, S.); Labalme, A (Labalme, A.); Ville, D (Ville, D.); Calender, A (Calender, A.); Mignot, C (Mignot, C.); Boutry-Kryza, N (Boutry-Kryza, N.); De Bellescize, J (De Bellescize, J.); Rivier-Ringenbach, C (Rivier-Ringenbach, C.); Bourel-Ponchel, E (Bourel-Ponchel, E.); Cheillan, D (Cheillan, D.); Simonet, T (Simonet, T.); Maincent, K (Maincent, K.); Rossi, M (Rossi, M.); Till, M (Till, M.); Mougou-Zerelli, S (Mougou-Zerelli, S.); Edery, P (Edery, P.); Saad, A (Saad, A.); Heron, D (Heron, D.); des Portes, V (des Portes, V.); Sanlaville, D (Sanlaville, D.); Lesca, G (Lesca, G.)	CLINICAL GENETICS Volume: 89 Issue: 2 Pages: 198-204 DOI: 10.1111/cge.12636 Published: FEB 2016
The localisation of the apical Par/Cdc42 polarity module is specifically affected in microvillus inclusion disease	Michaux, G (Michaux, Gregoire); Massey-Harroche, D (Massey-Harroche, Dominique); Nicolle, O (Nicolle, Ophelie); Rabant, M (Rabant, Marion); Brousse, N (Brousse, Nicole); Goulet, O (Goulet, Olivier); Le Bivic, A (Le Bivic, Andre); Ruemmele, FM (Ruemmele, Frank M.)	BIOLOGY OF THE CELL Volume: 108 Issue: 1 Pages: 19-28 DOI: 10.1111/boc.201500034 Published: JAN 2016
Early-Onset Encephalopathy with Epilepsy Associated with a Novel Splice Site Mutation in SMC1A	Lebrun, N (Lebrun, Nicolas); Lebon, S (Lebon, Sebastien); Jeannet, PY (Jeannet, Pierre-Yves); Jacquemont, S (Jacquemont, Sebastien); Billuart, P (Billuart, Pierre); Bienvenu, T (Bienvenu, Thierry)	AMERICAN JOURNAL OF MEDICAL GENETICS PART A Volume: 167 Issue: 12 Pages: 3076-3081 DOI: 10.1002/ajmg.a.37364 Published: DEC 2015

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New Practical Definitions for the Diagnosis of Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	Pilliod, J (Pilliod, Julie); Moutton, S (Moutton, Sebastien); Lavie, J (Lavie, Julie); Maurat, E (Maurat, Elise); Hubert, C (Hubert, Christophe); Bellance, N (Bellance, Nadege); Anheim, M (Anheim, Mathieu); Forlani, S (Forlani, Sylvie); Mochel, F (Mochel, Fanny); N'Guyen, K (N'Guyen, Karine); Thauvin-Robinet, C (Thauvin-Robinet, Christel); Verny, C (Verny, Christophe); Milea, D (Milea, Dan); Lesca, G (Lesca, Gaetan); Koenig, M (Koenig, Michel); Rodriguez, D (Rodriguez, Diana); Houcinat, N (Houcinat, Nada); Van-Gils, J (Van-Gils, Julien); Durand, CM (Durand, Christelle M.); Guichet, A (Guichet, Agnes); Barth, M (Barth, Magalie); Bonneau, D (Bonneau, Dominique); Convers, P (Convers, Philippe); Maillart, E (Maillart, Elisabeth); Guyant-Marechal, L (Guyant-Marechal, Lucie); Hannequin, D (Hannequin, Didier); Fromager, G (Fromager, Guillaume); Afenjar, A (Afenjar, Alexandra); Chantot-Bastaraud, S (Chantot-Bastaraud, Sandra); Valence, S (Valence, Stephanie); Charles, P (Charles, Perrine); Berquin, P (Berquin, Patrick); Rooryck, C (Rooryck, Caroline); Bouron, J (Bouron, Julie); Brice, A (Brice, Alexis); Lacombe, D (Lacombe, Didier); Rossignol, R (Rossignol, Rodrigue); Stevanin, G (Stevanin, Giovanni); Benard, G (Benard, Giovanni); Burglen, L (Burglen, Lydie); Durr, A (Durr, Alexandra); Goizet, C (Goizet, Cyril); Coupry, I (Coupry, Isabelle)	ANNALS OF NEUROLOGY Volume: 78 Issue: 6 Pages: 871-886 DOI: 10.1002/ana.24509 Published: DEC 2015
Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies	Angebault, C (Angebault, Claire); Guichet, PO (Guichet, Pierre-Olivier); Talmat-Amar, Y (Talmat-Amar, Yasmina); Charif, M (Charif, Majida); Gerber, S (Gerber, Sylvie); Fares-Taie, L (Fares-Taie, Lucas); Gueguen, N (Gueguen, Naig); Halloyn, F (Halloyn, Francois); Moore, D (Moore, David); Amati-Bonneau, P (Amati-Bonneau, Patrizia); Manes, G (Manes, Gael); Hebrard, M (Hebrard, Maxime); Bocquet, B (Bocquet, Beatrice); Quiles, M (Quiles, Melanie); Piro-Megy, C (Piro-Megy, Camille); Teigell, M (Teigell, Marisa); Delettre, C (Delettre, Cecile); Rossel, M (Rossel, Mireille); Meunier, I (Meunier, Isabelle); Preising, M (Preising, Markus); Lorenz, B (Lorenz, Birgit); Carelli, V (Carelli, Valerio); Chinnery, PF (Chinnery, Patrick F.); Yu-Wai-Man, P (Yu-Wai-Man, Patrick); Kaplan, J (Kaplan, Josseline); Roubertie, A (Roubertie, Agathe); Barakat, A (Barakat, Abdelhamid); Bonneau, D (Bonneau, Dominique); Reynier, P (Reynier, Pascal); Rozet, JM (Rozet, Jean-Michel); Bomont, P (Bomont, Pascale); Hamel, CP (Hamel, Christian P.); Lenaers, G (Lenaers, Guy)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 97 Issue: 5 Pages: 754-760 DOI: 10.1016/j.ajhg.2015.09.012 Published: NOV 5 2015
Skeletal muscle sodium channelopathies	Nicole, S (Nicole, Sophie); Fontaine, B (Fontaine, Bertrand)	CURRENT OPINION IN NEUROLOGY Volume: 28 Issue: 5 Pages: 508-514 DOI: 10.1097/WCO.0000000000000238 Published: OCT 2015
A strategy to discover decoy chemokine ligands with an anti-inflammatory activity	Abboud, D (Abboud, Dayana); Daubeuf, F (Daubeuf, Francois); Do, QT (Quoc Tuan Do); Utard, V (Utard, Valerie); Villa, P (Villa, Pascal); Haiech, J (Haiech, Jacques); Bonnet, D (Bonnet, Dominique); Hibert, M (Hibert, Marcel); Bernard, P (Bernard, Philippe); Galzi, JL (Galzi, Jean-Luc); Frossard, N (Frossard, Nelly)	SCIENTIFIC REPORTS Volume: 5 Article Number: 14746 DOI: 10.1038/srep14746 Published: OCT 7 2015
The Intracellular Domain of the Coxsackievirus and Adenovirus Receptor Differentially Influences Adenovirus Entry	Loustalot, F (Loustalot, Fabien); Kremer, EJ (Kremer, Eric J.); Salinas, S (Salinas, Sara)	JOURNAL OF VIROLOGY Volume: 89 Issue: 18 Pages: 9417-9426 DOI: 10.1128/JVI.01488-15 Published: SEP 2015
Rare ACTG1 variants in fetal microlissencephaly	Poirier, K (Poirier, Karine); Martinovic, J (Martinovic, Jelena); Laquerriere, A (Laquerriere, Annie); Cavallin, M (Cavallin, Mara); Fallet-Bianco, C (Fallet-Bianco, Catherine); Desguerre, I (Desguerre, Isabelle); Valence, S (Valence, Stephanie); Grande-Goburghun, J (Grande-Goburghun, Jocelyne); Francannet, C (Francannet, Christine); Deleuze, JF (Deleuze, Jean-Francois); Boland, A (Boland, Anne); Chelly, J (Chelly, Jamel); Bahi-Buisson, N (Bahi-Buisson, Nadia)	EUROPEAN JOURNAL OF MEDICAL GENETICS Volume: 58 Issue: 8 Pages: 416-418 DOI: 10.1016/j.ejmg.2015.06.006 Published: AUG 2015
Adenovirus Tales: From the Cell Surface to the Nuclear Pore Complex	Kremer, EJ (Kremer, Eric J.); Nemerow, GR (Nemerow, Glen R.)	PLOS PATHOGENS Volume: 11 Issue: 6 Article Number: e1004821 DOI: 10.1371/journal.ppat.1004821 Published: JUN 2015
Limiting hepatic Bmp-Smad signaling by matriptase-2 is required for erythropoietin-mediated hepcidin suppression in mice	Nai, A (Nai, Antonella); Rubio, A (Rubio, Aude); Campanella, A (Campanella, Alessandro); Gourbeyre, O (Gourbeyre, Ophelie); Artuso, I (Artuso, Irene); Bordini, J (Bordini, Jessica); Gineste, A (Gineste, Aurelie); Latour, C (Latour, Chloe); Besson-Fournier, C (Besson-Fournier, Celine); Lin, HY (Lin, Herbert Y.); Coppin, H (Coppin, Helene); Roth, MP (Roth, Marie-Paule); Camaschella, C (Camaschella, Clara); Silvestri, L (Silvestri, Laura); Meynard, D (Meynard, Delphine)	BLOOD Volume: 127 Issue: 19 Pages: 2327-2336 DOI: 10.1182/blood-2015-11-681494 Published: MAY 12 2016

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X-linked Inhibitor of Apoptosis Protein Deficiency: More than an X-linked Lymphoproliferative Syndrome	Aguilar, C (Aguilar, Claire); Latour, S (Latour, Sylvain)	JOURNAL OF CLINICAL IMMUNOLOGY Volume: 35 Issue: 4 Pages: 331-338 DOI: 10.1007/s10875-015-0141-9 Published: MAY 2015
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