FONDATION maladies rares

PUBLICATIONS DES PROJETS SOUTENUS PAR LA FONDATION

2019

Title	Author(s)	Source
A new case of SMA phenotype without epilepsy due to biallelic variants in ASAH1	van der Beek, NAME (van der Beek, Nadine A. M. E.); Nelson, I (Nelson, Isabelle); Froissart, R (Froissart, Roseline); Levade, T (Levade, Thierry); Garcia, V (Garcia, Virginie); Lacene, E (Lacene, Emmanuelle); Boland, A (Boland, Anne); Masson, C (Masson, Cecile); Romero, NB (Romero, Norma B.); Stojkovic, T (Stojkovic, Tanya); Bonne, G (Bonne, Gisele); Behin, A (Behin, Anthony)	EUROPEAN JOURNAL OF HUMAN GENETICS Volume: 27 Issue: 3 Pages: 337-339 DOI: 10.1038/s41431-018-0250-z Published: MAR 2019
Bi-allelic Mutations in ARMC2 Lead to Severe Astheno-T eratozoospermia Due to Sperm Flagellum Malformations in Humans and Mice	Coutton, C (Coutton, Charles); Martinez, G (Martinez, Guillaume); Kherraf, ZE (Kherraf, Zine-Eddine); Amiri-Yekta, A (Amiri-Yekta, Amir); Boguenet, M (Boguenet, Magalie); Saut, A (Saut, Antoine); He, XJ (He, Xiaojin); Zhang, F (Zhang, Feng); Cristou-Kent, M (Cristou-Kent, Marie); Escoffier, J (Escoffier, Jessica); Bidart, M (Bidart, Marie); Satre, V (Satre, Veronique); Conne, B (Conne, Beatrice); Ben Mustapha, SF (Ben Mustapha, Selima Fourati); Halouani, L (Halouani, Lazhar); Marrakchi, O (Marrakchi, Ouafi); Makni, M (Makni, Mounir); Latrous, H (Latrous, Habib); Kharouf, M (Kharouf, Mahmoud); Pernet-Gallay, K (Pernet-Gallay, Karin); Bonhivers, M (Bonhivers, Melanie); Hennebicq, S (Hennebicq, Sylviane); Rives, N (Rives, Nathalie); Dulioust, E (Dulioust, Emmanuel); Toure, A (Toure, Aminata); Gourabi, H (Gourabi, Hamid); Cao, YX (Cao, Yunxia); Zouari, R (Zouari, Raoudha); Hosseini, SH (Hosseini, Seyedeh Hanieh); Nef, S (Nef, Serge); Thierry-Mieg, N (Thierry-Mieg, Nicolas); Arnoult, C (Arnoult, Christophe); Ray, PF (Ray, Pierre F.)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 104 Issue: 2 Pages: 331-340 DOI: 10.1016/j.ajhg.2018.12.013 Published: FEB 7 2019
Prospective memory in narcolepsy type 1 patients	Tonetti, L (Tonetti, Lorenzo); Natale, V (Natale, Vincenzo); Gauriau, C (Gauriau, Caroline); Faraut, B (Faraut, Brice); Philip, P (Philip, Pierre); Leger, D (Leger, Damien)	JOURNAL OF PSYCHOSOMATIC RESEARCH Volume: 117 Pages: 30-31 DOI: 10.1016/j.jpsychores.2018.12.008 Published: FEB 2019
Identification and Characterization o Known Biallelic Mutations in the IFT27 (BBS19) Gene in a Novel Family With Bardet-Biedl Syndrome	Schaefer, E (Schaefer, Elise); Delvallee, C (Delvallee, Clarisse); Mary, L (Mary, Laura); Stoetzel, C (Stoetzel, Corinne); Geoffroy, V (Geoffroy, Veronique); Marks-Delesalle, C (Marks-Delesalle, Caroline); Holder-Espinasse, M (Holder-Espinasse, Muriel); Ghoumid, J (Ghoumid, Jamal); Dollfus, H (Dollfus, Helene); Muller, J (Muller, Jean)	FRONTIERS IN GENETICS Volume: 10 Article Number: 21 DOI: 10.3389/fgene.2019.00021 Published: JAN 30 2019

Title	Author(s)	Source
Genetics meets DNA methylation in rare diseases	Velasco, G (Velasco, Guillaume); Francastel, C (Francastel, Claire)	CLINICAL GENETICS Volume: 95 Issue: 2 Pages: 210-220 DOI: 10.1111/cge.13480 Published: FEB 2019
Sarcomeric disorganization and nemaline bodies in muscle biopsies of patients with EXOSC3-related type 1 pontocerebellar hypoplasia	Pinto, MM (Pinto, Miguel M.); Monges, S (Monges, Soledad); Malfatti, E (Malfatti, Edoardo); Lubieniecki, F (Lubieniecki, Fabiana); Lornage, X (Lornage, Xaviere); Alias, L (Alias, Laura); Labasse, C (Labasse, Clemence); Madelaine, A (Madelaine, Angeline); Fardeau, M (Fardeau, Michel); Laporte, J (Laporte, Jocelyn); Tizzano, EF (Tizzano, Eduardo F.); Romero, NB (Romero, Norma B.)	MUSCLE & NERVE Volume: 59 Issue: 1 Pages: 137-141 DOI: 10.1002/mus.26305 Published: JAN 2019
Mental stress test: a rapid, simple, and efficient test to unmask long QT syndrome	Etienne, P (Etienne, Pauline); Huchet, F (Huchet, Francois); Gaborit, N (Gaborit, Nathalie); Barc, J (Barc, Julien); Thollet, A (Thollet, Aurelie); Kyndt, F (Kyndt, Florence); Guyomarch, B (Guyomarch, Beatrice); Le Marec, H (Le Marec, Herve); Charpentier, F (Charpentier, Flavien); Schott, JJ (Schott, Jean-Jacques); Redon, R (Redon, Richard); Probst, V (Probst, Vincent); Gourraud, JB (Gourraud, Jean-Baptiste)	EUROPACE Volume: 20 Issue: 12 Pages: 2014-2020 DOI: 10.1093/europace/euy078 Published: DEC 2018
Loss of Sarcomeric Scaffolding as a Common Baseline Histopathologic Lesion in Titin-Related Myopathies	Avila-Polo, R (Avila-Polo, Rainiero); Malfatti, E (Malfatti, Edoardo); Lornage, X (Lornage, Xaviere); Cheraud, C (Cheraud, Chrystel); Nelson, I (Nelson, Isabelle); Nectoux, J (Nectoux, Juliette); Bohm, J (Bohm, Johann); Schneider, R (Schneider, Raphael); Hedberg-Oldfors, C (Hedberg-Oldfors, Carola); Eymard, B (Eymard, Bruno); Monges, S (Monges, Soledad); Lubieniecki, F (Lubieniecki, Fabiana); Brochier, G (Brochier, Guy); Bui, MT (Bui, Mai Thao); Madelaine, A (Madelaine, Angeline); Labasse, C (Labasse, Clemence); Beuvin, M (Beuvin, Maud); Lacene, E (Lacene, Emmanuelle); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Thompson, J (Thompson, Julie); Richard, I (Richard, Isabelle); Taratuto, AL (Taratuto, Ana Lia); Udd, B (Udd, Bjarne); Leturcq, F (Leturcq, France); Bonne, G (Bonne, Gisele); Oldfors, A (Oldfors, Anders); Laporte, J (Laporte, Jocelyn); Romero, NB (Romero, Norma Beatriz)	JOURNAL OF NEUROPATHOLOGY AND EXPERIMENTAL NEUROLOGY Volume: 77 Issue: 12 Pages: 1101-1114 DOI: 10.1093/jnen/nly095 Published: DEC 2018
LRP10 in alpha-synucleinopathies	Tesson, C (Tesson, Christelle); Brefel-Courbon, C (Brefel-Courbon, Christine); Corvol, JC (Corvol, Jean-Christophe); Lesage, S (Lesage, Suzanne); Brice, A (Brice, Alexis)	LANCET NEUROLOGY Volume: 17 Issue: 12 Pages: 1034-1034 DOI: 10.1016/S1474-4422(18)30400-9 Published: DEC 2018
Delineating FOXG1 syndrome From congenital microcephaly to hyperkinetic encephalopathy	Vegas, N (Vegas, Nancy); Cavallin, M (Cavallin, Mara); Maillard, C (Maillard, Camille); Boddaert, N (Boddaert, Nathalie); Toulouse, J (Toulouse, Joseph); Schaefer, E (Schaefer, Elise); Lerman-Sagie, T (Lerman-Sagie, Tally); Lev, D (Lev, Dorit); Magalie, B (Magalie, Barth); Moutton, S (Moutton, Sebastien); Haan, E (Haan, Eric); Isidor, B (Isidor, Bertrand); Heron, D (Heron, Delphine); Milh, M (Milh, Mathieu); Rondeau, S (Rondeau, Stephane); Michot, C (Michot, Caroline); Valence, S (Valence, Stephanie); Wagner, S (Wagner, Sabrina); Hully, M (Hully, Marie); Mignot, C (Mignot, Cyril); Masurel, A (Masurel, Alice); Datta, A (Datta, Alexandre); Odent, S (Odent, Sylvie); Nizon, M (Nizon, Mathilde); Lazaro, L (Lazaro, Leila); Vincent, M (Vincent, Marie); Cogne, B (Cogne, Benjamin); Guerrot, AM (Guerrot, Anne Marie); Arpin, S (Arpin, Stephanie); Pedespan, JM (Pedespan, Jean Michel); Caubel, I (Caubel, Isabelle); Pontier, B (Pontier, Benedicte); Troude, B (Troude, Baptiste); Rivier, F (Rivier, Francois); Philippe, C (Philippe, Christophe); Bienvenu, T (Bienvenu, Thierry); Spitz, MA (Spitz, Marie-Aude); Bery, A (Bery, Amandine); Bahi-Buisson, N (Bahi-Buisson, Nadia)	NEUROLOGY-GENETICS Volume: 4 Issue: 6 Article Number: UNSP e281 DOI: 10.1212/NXG.0000000000000281 Published: DEC 2018
Using actigraphy to assess sleep and wake rhythms of narcolepsy type 1 patients: a comparison with primary insomniacs and healthy controls	Leger, D (Leger, Damien); Gauriau, C (Gauriau, Caroline); Tonetti, L (Tonetti, Lorenzo); Lantin, M (Lantin, Michaele); Filardi, M (Filardi, Marco); Philip, P (Philip, Pierre); Faraut, B (Faraut, Brice); Natale, V (Natale, Vincenzo)	SLEEP MEDICINE Volume: 52 Pages: 88-91 DOI: 10.1016/j.sleep.2018.07.024 Published: DEC 2018
Further refinement of COL4A1 and COL4A2 related cortical malformations	Cavallin, M (Cavallin, Mara); Mine, M (Mine, Manuele); Philbert, M (Philbert, Marion); Boddaert, N (Boddaert, Nathalie); Lepage, JM (Lepage, Jean Marie); Coste, T (Coste, Thibault); Lopez-Gonzalez, V (Lopez-Gonzalez, Vanessa); Sanchez-Soler, MJ (Sanchez-Soler, Maria Jose); Ballesta-Martinez, MJ (Ballesta-Martinez, Maria Juliana); Remerand, G (Remerand, Ganaelle); Pasquier, L (Pasquier, Laurent); Guet, A (Guet, Agnes); Chelly, J (Chelly, Jamel); Lascelles, K (Lascelles, Karine); Prieto-Morin, C (Prieto-Morin, Carol); Kossorotoff, M (Kossorotoff, Manoelle); Lasserve, ET (Lasserve, Elisabeth Tournier); Bahi-Buisson, N (Bahi-Buisson, Nadia)	EUROPEAN JOURNAL OF MEDICAL GENETICS Volume: 61 Issue: 12 Pages: 765-772 DOI: 10.1016/j.ejmg.2018.10.004 Published: DEC 2018
Mutations in TBR1 gene leads to cortical malformations and intellectual disability	Vegas, N (Vegas, Nancy); Cavallin, M (Cavallin, Mara); Kleefstra, T (Kleefstra, Tjitske); de Boer, L (de Boer, Lonneke); Philbert, M (Philbert, Marion); Maillard, C (Maillard, Camille); Boddaert, N (Boddaert, Nathalie); Munnich, A (Munnich, Arnold); Hubert, L (Hubert, Laurence); Bery, A (Bery, Amandine); Besmond, C (Besmond, Claude); Bahi-Buisson, N (Bahi-Buisson, Nadia)	EUROPEAN JOURNAL OF MEDICAL GENETICS Volume: 61 Issue: 12 Pages: 759-764 DOI: 10.1016/j.ejmg.2018.09.012 Published: DEC 2018

Title	Author(s)	Source
Recurrent RTTN mutation leading to severe microcephaly, polymicrogyria and growth restriction	Cavallin, M (Cavallin, Mara); Bery, A (Bery, Amandine); Maillard, C (Maillard, Camille); Salomon, LJ (Salomon, Laurent J.); Bole, C (Bole, Christine); Reilly, ML (Reilly, Madeline Louise); Nitschke, P (Nitschke, Patrick); Boddaert, N (Boddaert, Nathalie); Bahi-Buisson, N (Bahi-Buisson, Nadia)	EUROPEAN JOURNAL OF MEDICAL GENETICS Volume: 61 Issue: 12 Pages: 755-758 DOI: 10.1016/j.ejmg.2018.08.001 Published: DEC 2018
TLE1, a key player in neurogenesis, a new andidate gene for a utosomal recessive postnatal microcephaly	Cavallin, M (Cavallin, Mara); Maillard, C (Maillard, Camille); Hully, M (Hully, Marie); Philbert, M (Philbert, Marion); Boddaert, N (Boddaert, Nathalie); Reilly, ML (Reilly, Madeline Louise); Nitschke, P (Nitschke, Patrick); Bery, A (Bery, Amandine); Bahi-Buisson, N (Bahi-Buisson, Nadia)	EUROPEAN JOURNAL OF MEDICAL GENETICS Volume: 61 Issue: 12 Pages: 729-732 DOI: 10.1016/j.ejmg.2018.05.002 Published: DEC 2018
SynAggreg: A Multifunctional High-Throughput Technology for Precision Study of Amyloid Aggregation and Systematic Discovery of Synergistic Inhibitor Compounds	Aviolat, H (Aviolat, Hubert); Nomine, Y (Nomine, Yves); Gloria, S (Gloria, Sophie); Bonhoure, A (Bonhoure, Anna); Hoffmann, D (Hoffmann, David); Ruhlmann, C (Ruhlmann, Christine); Nierengarten, H (Nierengarten, Helene); Ruffenach, F (Ruffenach, Frank); Villa, P (Villa, Pascal); Trottier, Y (Trottier, Yvon); Klein, FAC (Klein, Fabrice A. C.)	JOURNAL OF MOLECULAR BIOLOGY Volume: 430 Issue: 24 Pages: 5257-5279 DOI: 10.1016/j.jmb.2018.09.009 Published: DEC 7 2018
A Lacanian Approach to Medical Demand, With a Focus on Pediatric Genetics: A Plea for Subjectivization	Potier, R (Potier, Remy); Putois, O (Putois, Olivier)	FRONTIERS IN PSYCHOLOGY Volume: 9 Article Number: 2021 DOI: 10.3389/fpsyg.2018.02021 Published: NOV 1 2018
Genetic evaluation of patients with non-syndromic male infertility	Okutman, O (Okutman, Ozlem); Ben Rhouma, M (Ben Rhouma, Maroua); Benkhalifa, M (Benkhalifa, Moncef); Muller, J (Muller, Jean); Viville, S (Viville, Stephane)	JOURNAL OF ASSISTED REPRODUCTION AND GENETICS Volume: 35 Issue: 11 Pages: 1939-1951 DOI: 10.1007/s10815-018-1301-7 Published: NOV 2018
Prenatal retinoic acid exposure reveals candidate genes for craniofacial disorders	Berenguer, M (Berenguer, Marie); Darnaudery, M (Darnaudery, Muriel); Claverol, S (Claverol, Stephane); Bonneu, M (Bonneu, Marc); Lacombe, D (Lacombe, Didier); Rooryck, C (Rooryck, Caroline)	SCIENTIFIC REPORTS Volume: 8 Article Number: 17492 DOI: 10.1038/s41598-018-35681-0 Published: NOV 30 2018
Whole genome sequencing identifies a de novo 2.1 Mb balanced paracentric i nversion disrupting FOXP1 and leading to severe intellectual disability	Vuillaume, ML (Vuillaume, M-L); Cogne, B (Cogne, B.); Jeanne, M (Jeanne, M.); Boland, A (Boland, A.); Ung, DC (Ung, D-C); Quinquis, D (Quinquis, D.); Besnard, T (Besnard, T.); Deleuze, JF (Deleuze, J-F); Redon, R (Redon, R.); Bezieau, S (Bezieau, S.); Laumonnier, F (Laumonnier, F.); Toutain, A (Toutain, A.)	CLINICA CHIMICA ACTA Volume: 485 Pages: 218-223 DOI: 10.1016/j.cca.2018.06.048 Published: OCT 2018
Whole-exome sequencing identifies mutations in FSIP2 as a recurrent cause of multiple morphological abnormalities of the sperm flagella	Martinez, G (Martinez, Guillaume); Kherraf, ZE (Kherraf, Zine-Eddine); Zouari, R (Zouari, Raoudha); Ben Mustapha, SF (Ben Mustapha, Selima Fourati); Saut, A (Saut, Antoine); Pernet-Gallay, K (Pernet-Gallay, Karin); Bertrand, A (Bertrand, Anne); Bidart, M (Bidart, Marie); Hograindleur, JP (Hograindleur, Jean Pascal); Amiri-Yekta, A (Amiri-Yekta, Amir); Kharouf, M (Kharouf, Mahmoud); Karaouzene, T (Karaouzene, Thomas); Thierry-Mieg, N (Thierry-Mieg, Nicolas); Dacheux-Deschamps, D (Dacheux-Deschamps, Denis); Satre, V (Satre, Veronique); Bonhivers, M (Bonhivers, Melanie); Toure, A (Toure, Aminata); Arnoult, C (Arnoult, Christophe); Ray, PF (Ray, Pierre F.); Coutton, C (Coutton, Charles)	HUMAN REPRODUCTION Volume: 33 Issue: 10 Pages: 1973-1984 DOI: 10.1093/humrep/dey264 Published: OCT 2018
Preference heterogeneity with respect to whole genome sequencing. A discrete choice experiment among parents of children with rare genetic diseases	Peyron, C (Peyron, Christine); Pelissier, A (Pelissier, Aurore); Bejean, S (Bejean, Sophie)	SOCIAL SCIENCE & MEDICINE Volume: 214 Pages: 125-132 DOI: 10.1016/j.socscimed.2018.08.015 Published: OCT 2018
An African loss-of-function CACNA1C variant p.T1787M associated with a risk of ventricular fibrillation	Blancard, M (Blancard, Malorie); Debbiche, A (Debbiche, Amal); Kato, K (Kato, Koichi); Cardin, C (Cardin, Christelle); Sabrina, G (Sabrina, Guichard); Gandjbakhch, E (Gandjbakhch, Estelle); Probst, V (Probst, Vincent); Haissaguerre, M (Haissaguerre, Michel); Extramiana, F (Extramiana, Fabrice); Hocini, M (Hocini, Meleze); Olivier, G (Olivier, Geoffroy); Leenhardt, A (Leenhardt, Antoine); Guicheney, P (Guicheney, Pascale); Rougier, JS (Rougier, Jean-Sebastien)	SCIENTIFIC REPORTS Volume: 8 Article Number: 14619 DOI: 10.1038/s41598-018-32867-4 Published: OCT 2 2018
Mutations in the SRP54 gene cause severe congenital neutropenia as well as Shwachman-Diamond-like syndrome	Bellanne-Chantelot, C (Bellanne-Chantelot, Christine); Schmaltz-Panneau, B (Schmaltz-Panneau, Barbara); Marty, C (Marty, Caroline); Fenneteau, O (Fenneteau, Odile); Callebaut, I (Callebaut, Isabelle); Clauin, S (Clauin, Severine); Docet, A (Docet, Aurelie); Damaj, GL (Damaj, Gandhi-Laurent); Leblanc, T (Leblanc, Thierry); Pellier, I (Pellier, Isabelle); Stoven, C (Stoven, Cecile); Souquere, S (Souquere, Sylvie); Antony-Debre, I (Antony-Debre, Ileana); Beaupain, B (Beaupain, Blandine); Aladjidi, N (Aladjidi, Nathalie); Barlogis, V (Barlogis, Vincent); Bauduer, F (Bauduer, Frederic); Bensaid, P (Bensaid, Philippe); Boespflug-Tanguy, O (Boespflug-Tanguy, Odile); Berger, C (Berger, Claire); Bertrand, Y (Bertrand, Yves); Carausu, L (Carausu, Liana); Fieschi, C (Fieschi, Claire); Galambrun, C (Galambrun, Claire); Schmidt, A (Schmidt, Aline); Journel, H (Journel, Hubert); Mazingue, F (Mazingue, Francoise); Nelken, B (Nelken, Brigitte); Quah, TC (Thuan Chong Quah); Oksenhendler, E (Oksenhendler, Eric); Ouachee, M (Ouachee, Marie); Pasquet, M (Pasquet, Marlene); Saada, V (Saada, Veronique); Suarez, F (Suarez, Felipe); Pierron, G (Pierron, Gerard); Vainchenker, W (Vainchenker, William); Plo, I (Plo, Isabelle); Donadieu, J (Donadieu, Jean)	BLOOD Volume: 132 Issue: 12 Pages: 1318-1331 DOI: 10.1182/blood-2017-12-820308 Published: SEP 20 2018

Title	Author(s)	Source
Cortical progenitor biology: key features mediating proliferation versus differentiation	Uzquiano, A (Uzquiano, Ana)[1,2,3] ; Gladwyn-Ng, I (Gladwyn-Ng, Ivan) [4] ; Nguyen, L (Nguyen, Laurent)[4] ; Reiner, O (Reiner, Orly)[5] ; Gotz, M (Goetz, Magdalena)[6,7,8] ; Matsuzaki, F (Matsuzaki, Fumio)[9] ; Francis, F (Francis, Fiona)	JOURNAL OF NEUROCHEMISTRY Volume: 146 Issue: 5 Pages: 500-525 DOI: 10.1111/jnc.14338 Published:SEP 2018
A Homozygous Ancestral SVA-Insertion-Mediated Deletion in WDR66 Induces Multiple Morphological Abnormalities of the Sperm Flagellum and Male Infertility	Kherraf, ZE (Kherraf, Zine-Eddine)[1,2] ; Amiri-Yekta, A (Amiri-Yekta, Amir)[1,2,3] ; Dacheux, D (Dacheux, Denis)[4,5] ; Karaouzene, T (Karaouzene, Thomas)[1] ; Coutton, C (Coutton, Charles)[1,6] ; Christou-Kent, M (Christou-Kent, Marie)[1] ; Martinez, G (Martinez, Guillaume)[1,6] ; Landrein, N (Landrein, Nicolas)[4] ; Le Tanno, P (Le Tanno, Pauline)[1] ; Ben Mustapha, SF (Ben Mustapha, Selima Fourati)[7] ; Halouani, L (Halouani, Lazhar)[7] ; Marrakchi, O (Marrakchi, Ouafi) [7] ; Makni, M (Makni, Mounir)[7] ; Latrous, H (Latrous, Habib)[7] ; Kharouf, M (Kharouf, Mahmoud)[7] ; Pernet-Gallay, K (Pernet-Gallay, Karin)[8] ; Gourabi, H (Gourabi, Hamid)[3] ; Robinson, DR (Robinson, Derrick R.)[4] ; Crouzy, S (Crouzy, Serge)[10] ; Blum, M (Blum, Michael)[9] ; Thierry-Mieg, N (Thierry-Mieg, Nicolas)[9] ; Toure, A (Toure, Aminata)[11,12,13] ; Zouari, R (Zouari, Raoudha)[7] ; Arnoult, C (Arnoult, Christophe)[1] ; Bonhivers, M (Bonhivers, Melanie)[4] ; Ray, PF (Ray, Pierre F.)[1,2)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 103 Issue: 3 Pages: 400-412 DOI: 10.1016/j.ajhg.2018.07.014 Published:SEP 6 2018
Genetic Evidence Supporting the Role of the Calcium Channel, CACNA1S, in Tooth Cusp and Root Patterning	Laugel-Haushalter, V (Laugel-Haushalter, Virginie); Morkmued, S (Morkmued, Supawich); Stoetzel, C (Stoetzel, Corinne); Geoffroy, V (Geoffroy, Veronique); Muller, J (Muller, Jean); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Chennen, K (Chennen, Kirsley); Pitiphat, W (Pitiphat, Waranuch); Dollfus, H (Dollfus, Helene); Niederreither, K (Niederreither, Karen); Bloch-Zupan, A (Bloch-Zupan, Agnes); Pungchanchaikul, P (Pungchanchaikul, Patimaporn)	FRONTIERS IN PHYSIOLOGY Volume: 9 Article Number: 1329 DOI: 10.3389/fphys.2018.01329 Published: SEP 26 2018
Diagnostic Yield of Next-generation Sequencing in Very Early-onset Inflammatory Bowel Diseases: A Multicentre Study	Charbit-Henrion, F (Charbit-Henrion, Fabienne); Parlato, M (Parlato, Marianna); Hanein, S (Hanein, Sylvain); Duclaux-Loras, R (Duclaux-Loras, Remi); Nowak, J (Nowak, Jan); Begue, B (Begue, Bernadette); Rakotobe, S (Rakotobe, Sabine); Bruneau, J (Bruneau, Julie); Fourrage, C (Fourrage, Cecile); Alibeu, O (Alibeu, Olivier); Rieux-Laucat, F (Rieux-Laucat, Frederic); Levy, E (Levy, Eva); Stolzenberg, MC (Stolzenberg, Marie-Claude); Mazerolles, F (Mazerolles, Fabienne); Latour, S (Latour, Sylvain); Lenoir, C (Lenoir, Christelle); Fischer, A (Fischer, Alain); Picard, C (Picard, Capucine); Aloi, M (Aloi, Marina); Dias, JA (Dias, Jorge Amil); Ben Hariz, M (Ben Hariz, Mongi); Bourrier, A (Bourrier, Anne); Breuer, C (Breuer, Christian); Breton, A (Breton, Anne); Bronski, J (Bronski, Jiri); Buderus, S (Buderus, Stephan); Cananzi, M (Cananzi, Mara); Coopman, S (Coopman, Stephanie); Cremilleux, C (Cremilleux, Clara); Dabadie, A (Dabadie, Alain); Dumant-Forest, C (Dumant-Forest, Clementine); Gurkan, OE (Gurkan, Odul Egritas); Fabre, A (Fabre, Alexandre); Fischer, A (Fischer, Aude); Diaz, MG (German Diaz, Marta); Gonzalez-Lama, Y (Gonzalez-Lama, Yago); Goulet, O (Goulet, Olivier); Guariso, G (Guariso, Graziella); Gurcan, N (Gurcan, Neslihan); Homan, M (Homan, Matjaz); Hugot, JP (Hugot, Jean-Pierre); Jeziorski, E (Jeziorski, Eric); Karanika, E (Karanika, Evi); Lachaux, A (Lachaux, Alain); Lewindon, P (Lewindon, Peter); Lima, R (Lima, Rosa); Magro, F (Magro, Fernando); Major, J (Major, Janos); Malamut, G (Malamut, Georgia); Mas, E (Mas, Emmanuel); Mattyus, I (Mattyus, Istvan); Mearin, LM (Mearin, Luisa M.); Melek, J (Melek, Jan); Navas-Lopez, VM (Manuel Navas-Lopez, Victor); Paerregaard, A (Paerregaard, Anders); Pelatan, C (Pelatan, Cecile); Pigneur, B (Pigneur, Benedicte); Pais, IP (Pais, Isabel Pinto); Rebeuh, J (Rebeuh, Julie); Romano, C (Romano, Claudio); Siala, N (Siala, Nadia); Strisciuglio, C (Strisciuglio, Caterina); Tempia-Caliera, M (Tempia-Caliera, Michela); Tounian, P (Tounian, Patrick); Turner, D (Turner, Dan); Urbonas, V (Urbonas, Vaidotas); Willot, S (Willot, Stephanie); Ruemmele, FM (Ruemmele, Frank M.); Cerf-Bensussan, N (Cerf-Bensussan, Nadine)	JOURNAL OF CROHNS & COLITIS Volume: 12 Issue: 9 Pages: 1104-1112 DOI: 10.1093/ecco-jcc/jjy068 Published: SEP 2018
Dominant-negative IKZF1 mutations cause a T, B, and myeloid cell combined immunodeficiency	Boutboul, D (Boutboul, David); Kuehn, HS (Kuehn, Hye Sun); de Wyngaert, ZV (de Wyngaert, Zoe Van); Niemela, JE (Niemela, Julie E.); Callebaut, I (Callebaut, Isabelle); Stoddard, J (Stoddard, Jennifer); Lenoir, C (Lenoir, Christelle); Barlogis, V (Barlogis, Vincent); Farnarier, C (Farnarier, Catherine); Vely, F (Vely, Frederic); Yoshida, N (Yoshida, Nao); Kojima, S (Kojima, Seiji); Kanegane, H (Kanegane, Hirokazu); Hoshino, A (Hoshino, Akihiro); Hauck, F (Hauck, Fabian); Lhermitte, L (Lhermitte, Ludovic); Asnafi, V (Asnafi, Vahid); Roehrs, P (Roehrs, Philip); Chen, S (Chen, Shaoying); Verbsky, JW (Verbsky, James W.); Calvo, KR (Calvo, Katherine R.); Husami, A (Husami, Ammar); Zhang, KJ (Zhang, Kejian); Roberts, J (Roberts, Joseph); Amrol, D (Amrol, David); Sleaseman, J (Sleaseman, John); Hsu, AP (Hsu, Amy P.); Holland, SM (Holland, Steven M.); Marsh, R (Marsh, Rebecca); Fischer, A (Fischer, Alain); Fleisher, TA (Fleisher, Thomas A.); Picard, C (Picard, Capucine); Latour, S (Latour, Sylvain); Rosenzweig, SD (Rosenzweig, Sergio D.)	Source: JOURNAL OF CLINICAL INVESTIGATION Volume: 128 Issue: 7 Pages: 3071-3087 DOI: 10.1172/JCI98164 Published: JUL 2 2018
Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140	Geoffroy, V (Geoffroy, Veronique); Stoetzel, C (Stoetzel, Corinne); Scheidecker, S (Scheidecker, Sophie); Schaefer, E (Schaefer, Elise); Perrault, I (Perrault, Isabelle); Bar, S (Baer, Severine); Kroll, A (Kroll, Ariane); Delbarre, M (Delbarre, Marion); Antin, M (Antin, Manuela); Leuvrey, AS (Leuvrey, Anne-Sophie); Henry, C (Henry, Charline); Blanche, H (Blanche, Helene); Decker, E (Decker, Eva); Kloth, K (Kloth, Katja); Klaus, G (Klaus, Guenter); Mache, C (Mache, Christoph); Martin-Coignard, D (Martin-Coignard, Dominique); McGinn, S (McGinn, Steven); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Friant, S (Friant, Sylvie); Saunier, S (Saunier, Sophie); Rozet, JM (Rozet, Jean-Michel); Bergmann, C (Bergmann, Carsten); Dollfus, H (Dollfus, Helene); Muller, J (Muller, Jean)	HUMAN MUTATION Volume: 39 Issue: 7 Pages: 983-992 DOI: 10.1002/humu.23539 Published: JUL 2018

Title	Author(s)	Source
A heterodimer formed by bone morphogenetic protein 9 (BMP9) and BMP10 provides most BMP biological activity in plasma	Tillet, E (Tillet, Emmanuelle)[1] ; Ouarne, M (Ouarne, Marie)[1] ; Desroches-Castan, A (Desroches-Castan, Agnes)[1] ; Mallet, C (Mallet, Christine)[1] ; Subileau, M (Subileau, Mariela)[1] ; Didier, R (Didier, Robin)[1,2] ; Lioutsko, A (Lioutsko, Anna)[1,3] ; Belthier, G (Belthier, Guillaume)[1,4] ; Feige, JJ (Feige, Jean-Jacques)[1] ; Bailly, S (Bailly, Sabine)[1]	JOURNAL OF BIOLOGICAL CHEMISTRY Volume: 293 Issue: 28 Pages: 10963-10974 DOI: 10.1074/jbc.RA118.002968 Published:JUL 13 2018
Thyroid Hypoplasia in Congenital Hypothyroidism Associated with Thyroid Peroxidase Mutations	Stoupa, A (Stoupa, Athanasia)[1,2,3] ; Chaabane, R (Chaabane, Rim)[4] ; Gueriouz, M (Gueriouz, Manelle)[2] ; Raynaud-Ravni, C (Raynaud-Ravni, Catherine)[5] ; Nitschke, P (Nitschke, Patrick)[6] ; Bole-Feysot, C (Bole-Feysot, Christine)[7] ; Mnif, M (Mnif, Mouna)[8] ; Ammar Keskes, L (Ammar Keskes, Leila)[4] ; Hachicha, M (Hachicha, Mongia)[9] ; Belguith, N (Belguith, Neila)[4,10] ; Polak, M (Polak, Michel)[1,2,3,11,12] ; Carre, A (Carre, Aurore)[2,3]	THYROID Volume: 28 Issue: 7 Pages: 941-944 DOI: 10.1089/thy.2017.0502 Published:JUL 2018
INTU-related oral-facial-digital syndrome type VI: A confirmatory report	Bruel, AL (Bruel, A. -L.); Levy, J (Levy, J.); Elenga, N (Elenga, N.); Defo, A (Defo, A.); Favre, A (Favre, A.); Lucron, H (Lucron, H.); Capri, Y (Capri, Y.); Perrin, L (Perrin, L.); Passemard, S (Passemard, S.); Vial, Y (Vial, Y.); Tabet, AC (Tabet, A. -C.); Faivre, L (Faivre, L.); Thauvin-Robinet, C (Thauvin-Robinet, C.); Verloes, A (Verloes, A.)	CLINICAL GENETICS Volume: 93 Issue: 6 Pages: 1205-1209 DOI: 10.1111/cge.13238 Published: JUN 2018
Whole-exome sequence analysis highlights the role of unmasked recessive mutations in copy number variants with incomplete penetrance	Egloff, M (Egloff, Matthieu)[1,2,3] ; Nguyen, LS (Nguyen, Lam-Son)[2,3] ; Siquier-Pernet, K (Siquier-Pernet, Karine)[2,3] ; Cormier-Daire, V (Cormier-Daire, Valerie)[3,4] ; Baujat, G (Baujat, Genevieve)[4] ; Attie-Bitach, T (Attie-Bitach, Tania)[1,3] ; Bole-Feysot, C (Bole-Feysot, Christine)[5] ; Nitschke, P (Nitschke, Patrick)[6] ; Vekemans, M (Vekemans, Michel)[1,3] ; Colleaux, L (Colleaux, Laurence)[2,3] ; Malan, V (Malan, Valerie)[1,2,3]	EUROPEAN JOURNAL OF HUMAN GENETICS Volume: 26 Issue: 6 Pages: 912-918 DOI: 10.1038/s41431-018-0124-4 Published:JUN 2018
A MEI1 homozygous missense mutation associated with meiotic arrest in a consanguineous family	Ben Khelifa, M (Ben Khelifa, M.); Ghieh, F (Ghieh, F.); Boudjenah, R (Boudjenah, R.); Hue, C (Hue, C.); Fauvert, D (Fauvert, D.); Dard, R (Dard, R.); Garchon, HJ (Garchon, H. J.); Vialard, F (Vialard, F.	HUMAN REPRODUCTION Volume: 33 Issue: 6 Pages: 1034-1037 DOI: 10.1093/humrep/dey073 Published: JUN 2018
Novel mutations in DNAJB6 cause LGMD1D and distal myopathy in French families	Jonson, PH (Jonson, P. H.)[1] ; Palmio, J (Palmio, J.)[2] ; Johari, M (Johari, M.)[1] ; Penttila, S (Penttila, S.)[2] ; Evila, A (Evila, A.)[1] ; Nelson, I (Nelson, I.)[3] ; Bonne, G (Bonne, G.)[3] ; Wiart, N (Wiart, N.)[4] ; Meyer, V (Meyer, V.)[4] ; Boland, A (Boland, A.)[4] ; Deleuze, JF (Deleuze, J. -F.)[4] ; Masson, C (Masson, C.)[5] ; Stojkovic, T (Stojkovic, T.)[3] ; Chapon, F (Chapon, F.)[6] ; Romero, NB (Romero, N. B.)[7] ; Sole, G (Sole, G.)[8] ; Ferrer, X (Ferrer, X.)[8] ; Ferreiro, A (Ferreiro, A.)[9,10] ; Hackman, P (Hackman, P.)[1] ; Richard, I (Richard, I.)[11] ; Udd, B (Udd, B.)[1,2,12]	EUROPEAN JOURNAL OF NEUROLOGY Volume: 25 Issue: 5 Pages: 790-794 DOI: 10.1111/ene.13598 Published:MAY 2018
PATL2 is a key actor of oocyte maturation whose invalidation causes infertility in women and mice	Christou-Kent, M (Christou-Kent, Marie)[1] ; Kherraf, ZE (Kherraf, Zine-Eddine)[1] ; Amiri-Yekta, A (Amiri-Yekta, Amir)[1,2,3] ; Le Blevec, E (Le Blevec, Emilie)[1] ; Karaouzene, T (Karaouzene, Thomas)[1] ; Conne, B (Conne, Beatrice)[4] ; Escoffier, J (Escoffier, Jessica)[1] ; Assou, S (Assou, Said)[5] ; Guttin, A (Guttin, Audrey)[6] ; Lambert, E (Lambert, Emeline)[1] ; Martinez, G (Martinez, Guillaume)[1,2,7] ; Boguenet, M (Boguenet, Magalie)[1] ; Ben Mustapha, SF (Ben Mustapha, Selima Fourati)[8] ; Cedrin Durnerin, I (Cedrin Durnerin, Isabelle)[9] ; Halouani, L (Halouani, Lazhar)[8] ; Marrakchi, O (Marrakchi, Ouafi)[8] ; Makni, M (Makni, Mounir)[8] ; Latrous, H (Latrous, Habib)[8] ; Kharouf, M (Kharouf, Mahmoud)[8] ; Coutton, C (Coutton, Charles)[1,2,7] ; Thierry-Mieg, N (Thierry-Mieg, Nicolas)[10] ; Nef, S (Nef, Serge)[4] ; Bottari, SP (Bottari, Serge P.)[1] ; Zouari, R (Zouari, Raoudha)[8] ; Issartel, JP (Issartel, Jean Paul)[6] ; Ray, PF (Ray, Pierre F.)[1,2] ; Arnoult, C (Arnoult, Christophe)[1]	EMBO MOLECULAR MEDICINE Volume: 10 Issue: 5Article Number: UNSP e8515 DOI: 10.15252/emmm.201708515 Published:MAY 2018
Missense mutation of TTC7A mimicking tricho-hepato-enteric (SD/THE) syndrome in a patient with very-early onset inflammatory bowel disease	Neves, JF (Neves, Joao Farela)[1,2] ; Afonso, I (Afonso, Isabel)[3] ; Borrego, L (Borrego, Luis)[2,4] ; Martins, C (Martins, Catarina)[2] ; Cordeiro, AI (Cordeiro, Ana Isabel)[1] ; Neves, C (Neves, Conceicao)[1] ; Lacoste, C (Lacoste, Caroline)[5] ; Badens, C (Badens, Catherine)[5,6] ; Fabre, A (Fabre, Alexandre)[6,7]	EUROPEAN JOURNAL OF MEDICAL GENETICS Volume: 61 Issue: 4 Pages: 185-188 DOI: 10.1016/j.ejmg.2017.11.014 Published:APR 2018
Human ALPI deficiency causes inflammatory bowel disease and highlights a key mechanism of gut homeostasis	Parlato, M (Parlato, Marianna)[1,2,3] ; Charbit-Henrion, F (Charbit-Henrion, Fabienne)[1,2,3,4,5] ; Pan, J (Pan, Jie)[6,7] ; Romano, C (Romano, Claudio)[3,8] ; Duclaux-Loras, R (Duclaux-Loras, Remi)[1,2,3,4] ; Le Du, MH (Le Du, Marie-Helene)[9] ; Warner, N (Warner, Neil)[6,7] ; Francalanci, P (Francalanci, Paola)[10,11] ; Bruneau, J (Bruneau, Julie)[4,12] ; Bras, M (Bras, Marc)[2,13] ; Zarhrate, M (Zarhrate, Mohammed)[14] ; Begue, B (Begue, Bernadette)[1,2,3] ; Guegan, N (Guegan, Nicolas)[1,2,4] ; Rakotobe, S (Rakotobe, Sabine)[1,2,3] ; Kapel, N (Kapel, Nathalie)[15] ; De Angelis, P (De Angelis, Paola)[10,11] ; Griffiths, AM (Griffiths, Anne M.)[6,7] ; Fiedler, K (Fiedler, Karoline)[6,7] ; Crowley, E (Crowley, Eileen)[6,7] ; Ruemmele, F (Ruemmele, Frank)[1,2,3,4,5] ; Muise, AM (Muise, Aleixo M.)[6,7,16,17] ; Cerf-Bensussan, N (Cerf-Bensussan, Nadine)[1,2,3,4]	EMBO MOLECULAR MEDICINE Volume: 10 Issue: 4 Article Number: UNSP e8483 DOI: 10.15252/emmm.201708483 Published:APR 2018

Title	Author(s)	Source
Absence of CFAP69 Causes Male Infertility due to Multiple Morphological Abnormalities of the Flagella in Human and Mouse	Dong, FN (Dong, Frederick N.)[1] ; Amiri-Yekta, A (Amiri-Yekta, Amir) [2,3] ; Martinez, G (Martinez, Guillaume)[3,4] ; Saut, A (Saut, Antoine) [3,4] ; Tek, J (Tek, Julie)[6,7] ; Stouvenel, L (Stouvenel, Laurence)[5,6,7] ; Lores, P (Lores, Patrick)[5,6,7] ; Karaouzene, T (Karaouzene, Thomas)[3,8,9] ; Thierry-Mieg, N (Thierry-Mieg, Nicolas)[9] ; Satre, V (Satre, Veronique)[3,4] ; Brouillet, S (Brouillet, Sophie)[10] ; Daneshipour, A (Daneshipour, Abbas)[2] ; Hosseini, SH (Hosseini, Seyedeh Hanieh)[11] ; Bonhivers, M (Bonhivers, Melanie)[12,13] ; Gourabi, H (Gourabi, Hamid)[2] ; Dulioust, E (Dulioust, Emmanuel) [7,14] ; Arnoult, C (Arnoult, Christophe)[3] ; Toure, A (Toure, Aminata) [5,6,7] ; Ray, PF (Ray, Pierre F.)[3,8] ; Zhao, HQ (Zhao, Haiqing)[1] ; Coutton, C (Coutton, Charles)[3,4]	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 102 Issue: 4 Pages: 636-648 DOI: 10.1016/j.ajhg.2018.03.007 Published:APR 5 2018
Autoimmune Lymphoproliferative Syndrome-FAS Patients Have an Abnormal Regulatory T Cell (Treg) Phenotype but Display Normal Natural Treg-Suppressive Function on T Cell Proliferation	Mazerolles, F (Mazerolles, Fabienne)[1,2] ; Stolzenberg, MC (Stolzenberg, Marie-Claude)[1,2] ; Pelle, O (Pelle, Olivier)[1,3] ; Picard, C (Picard, Capucine)[2,4,5,6] ; Neven, B (Neven, Benedicte)[1,2,4] ; Fischer, A (Fischer, Alain)[2,4,7] ; Magerus-Chatinet, A (Magerus-Chatinet, Aude)[1,2] ; Rieux-Laucat, F (Rieux-Laucat, Frederic)[1,2]	FRONTIERS IN IMMUNOLOGY Volume: 9 Article Number: 718 DOI: 10.3389/fimmu.2018.00718 Published:APR 9 2018
Cardiac function and exercise adaptation in 8 children with LPIN1 mutations	Legendre, A (Legendre, Antoine)[1] ; Khraiche, D (Khraiche, Diala)[1] ; Ou, P (Ou, Phalla)[2] ; Mauvais, FX (Mauvais, Francois-Xavier)[3,4] ; Madrange, M (Madrange, Marine)[3] ; Guemann, AS (Guemann, Anne-Sophie)[3] ; Jais, JP (Jais, Jean-Philippe)[5] ; Bonnet, D (Bonnet, Damien)[1,6] ; Hamel, Y (Hamel, Yamina)[3] ; de Lonlay, P (de Lonlay, Pascale)[3]	MOLECULAR GENETICS AND METABOLISM Volume: 123 Issue: 3 Pages: 375-381 DOI: 10.1016/j.ymgme.2017.12.429 Published:MAR 2018+C107
Pediatric-onset Evans syndrome: Heterogeneous presentation and high frequency of monogenic disorders including LRBA and CTLA4 mutations	Besnard, C (Besnard, Caroline)[1,3,4] ; Levy, E (Levy, Eva)[1,2] ; Aladjidi, N (Aladjidi, Nathalie)[5] ; Stolzenberg, MC (Stolzenberg, Marie-Claude)[1,2] ; Magerus-Chatinet, A (Magerus-Chatinet, Aude)[1,2] ; Alibeu, O (Alibeu, Olivier)[6] ; Nitschke, P (Nitschke, Patrick)[2,7] ; Blanche, S (Blanche, Stephane)[2,3] ; Hermine, O (Hermine, Olivier) [2,8] ; Jeziorski, E (Jeziorski, Eric)[9] ; Landman-Parker, J (Landman-Parker, Judith)[4,10] ; Leverger, G (Leverger, Guy)[10] ; Mahlaoui, N (Mahlaoui, Nizar)[3] ; Michel, G (Michel, Gerard)[11] ; Pellier, I (Pellier, Isabelle)[12] ; Suarez, F (Suarez, Felipe)[2,13,14] ; Thuret, I (Thuret, Isabelle)[15] ; de Saint-Basile, G (de Saint-Basile, Genevieve)[2,16,17] ; Picard, C (Picard, Capucine)[2,3,17] ; Fischer, A (Fischer, Alain)[1,2,3,18] ; Neven, B (Neven, Benedicte)[1,2,3] ; Rieux-Laucat, F (Rieux-Laucat, Frederic)[1,2] ; Quartier, P (Quartier, Pierre)[1,2,3]	CLINICAL IMMUNOLOGY Volume: 188 Pages: 52-57 DOI: 10.1016/j.clim.2017.12.009 Published:MAR 2018
Loss-of-Function Mutations in UNC45A Cause a Syndrome Associating Cholestasis, Diarrhea, Impaired Hearing, and Bone Fragility	Emmanuelle Ecochard-Dugelay, Ge´raldine Hery, Fre´de´ric Huet, Philippe Gauchez, Emmanuel Gonzales, Catherine Guettier-Bouttier, Mina Komuta, Caroline Lacoste, Raphaelle Maudinas, Karin Mazodier, Yves Rimet, Jean-Baptiste Rivie`re, Bertrand Roquelaure, Sabine Sigaudy, Xavier Stephenne, Christel Thauvin-Robinet, Julien Thevenon, Jacques Sarles, Nicolas Levy, Catherine Badens, Olivier Goulet, Jean-Pierre Hugot, Nicholas Katsanis, Laurence Faivre, and Alexandre Fabre,	The American Journal of Human Genetics (2018), https://doi.org/10.1016/j.ajhg.2018.01.009
Whole exome sequencing in three families segregating a pediatric case of sarcoidosis	Calender, A (Calender, Alain)[1,3] ; Farnier, PAR (Farnier, Pierre Antoine Rollat)[2] ; Buisson, A (Buisson, Adrien)[1] ; Pinson, S (Pinson, Stephane)[1] ; Bentaher, A (Bentaher, Abderrazzaq)[3] ; Lebecque, S (Lebecque, Serge)[4] ; Corvol, H (Corvol, Harriet)[5] ; Abou Taam, R (Abou Taam, Rola)[6] ; Houdouin, V (Houdouin, Veronique)[7] ; Bardel, C (Bardel, Claire)[2] ; Roy, P (Roy, Pascal)[2] ; Devouassoux, G (Devouassoux, Gilles)[8] ; Cottin, V (Cottin, Vincent)[9] ; Seve, P (Seve, Pascal)[10] ; Bernaudin, JF (Bernaudin, Jean-Francois)[11] ; Lim, CX (Lim, Clarice X.)[12] ; Weichhart, T (Weichhart, Thomas)[12] ; Valeyre, D (Valeyre, Dominique)[13,14] ; Pacheco, Y (Pacheco, Yves)[3] ; Clement, A (Clement, Annick)[15] ; Nathan, N (Nathan, Nadia)[15]	BMC MEDICAL GENOMICS Volume: 11; Article Number: 23 DOI: 10.1186/s12920-018-0338-x Published:MAR 6 2018
G908R NOD2 variant in a family with sarcoidosis	Besnard, V (Besnard, Valerie)[1] ; Calender, A (Calender, Alain)[2] ; Bouvry, D (Bouvry, Diane)[1,3] ; Pacheco, Y (Pacheco, Yves)[4,5] ; Chapelon-Abric, C (Chapelon-Abric, Catherine)[6] ; Jeny, F (Jeny, Florence)[1,3] ; Nunes, H (Nunes, Hilario)[1,3] ; Planes, C (Planes, Carole)[1,3] ; Valeyre, D (Valeyre, Dominique)[1,3]	RESPIRATORY RESEARCH Volume: 19; Article Number: 44 DOI: 10.1186/s12931-018-0748-5 Published:MAR 20 2018
Discordant manifestations in Italian brothers with GNE myopathy	Dotti, MT (Dotti, Maria-Teresa); Malandrini, A (Malandrini, Alessandro); Lornage, X (Lornage, Xaviere); Mignarri, A (Mignarri, Andrea); Cantisani, TA (Cantisani, Teresa Anna); Bohm, J (Bohm, Johann); Laporte, J (Laporte, Jocelyn); Malfatti, E (Malfatti, Edoardo)	JOURNAL OF THE NEUROLOGICAL SCIENCES Volume: 386 Pages: 1-3 DOI: 10.1016/j.jns.2018.01.002 Published: MAR 15 2018

Title	Author(s)	Source
Clinical and genetic heterogeneity in familial steroid-sensitive nephrotic syndrome	Dorval, G (Dorval, Guillaume); Gribouval, O (Gribouval, Olivier); Martinez-Barquero, V (Martinez-Barquero, Vanesa); Machuca, E (Machuca, Eduardo); Tete, MJ (Tete, Marie-Josephe); Baudouin, V (Baudouin, Veronique); Benoit, S (Benoit, Stephane); Chabchoub, I (Chabchoub, Imen); Champion, G (Champion, Gerard); Chauveau, D (Chauveau, Dominique); Chehade, H (Chehade, Hassib); Chouchane, C (Chouchane, Chokri); Cloarec, S (Cloarec, Sylvie); Cochat, P (Cochat, Pierre); Dahan, K (Dahan, Karin); Dantal, J (Dantal, Jacques); Delmas, Y (Delmas, Yahsou); Deschenes, G (Deschenes, Georges); Dolhem, P (Dolhem, Phillippe); Durand, D (Durand, Dominique); Ekinci, Z (Ekinci, Zelal); El Karoui, K (El Karoui, Khalil); Fischbach, M (Fischbach, Michel); Grunfeld, JP (Grunfeld, Jean-Pierre); Guigonis, V (Guigonis, Vincent); Hachicha, M (Hachicha, Mongia); Hogan, J (Hogan, Julien); Hourmant, M (Hourmant, Maryvonne); Hummel, A (Hummel, Aurelie); Kamar, N (Kamar, Nassim); Krummel, T (Krummel, Thierry); Lacombe, D (Lacombe, Didier); Llanas, B (Llanas, Brigitte); Mesnard, L (Mesnard, Laurent); Mohsin, N (Mohsin, Nabil); Niaudet, P (Niaudet, Patrick); Nivet, H (Nivet, Hubert); Parvex, P (Parvex, Paloma); Pietrement, C (Pietrement, Christine); de Pontual, L (de Pontual, Loic); Noble, CP (Noble, Claire Pouteil); Ribes, D (Ribes, David); Ronco, P (Ronco, Pierre); Rondeau, E (Rondeau, Eric); Sallee, M (Sallee, Marion); Tsimaratos, M (Tsimaratos, Michel); Ulinski, T (Ulinski, Tim); Salomon, R (Salomon, Remi); Antignac, C (Antignac, Corinne); Boyer, O (Boyer, Olivia)	PEDIATRIC NEPHROLOGY Volume: 33 Issue: 3 Pages: 473-483 DOI: 10.1007/s00467-017-3819-9 Published: MAR 2018
Loss of RASGRP1 in humans impairs T-cell expansion leading to Epstein-Barr virus susceptibility	Winter, S (Winter, Sarah); Martin, E (Martin, Emmanuel); Boutboul, D (Boutboul, David); Lenoir, C (Lenoir, Christelle); Boudjemaa, S (Boudjemaa, Sabah); Petit, A (Petit, Arnaud); Picard, C (Picard, Capucine); Fischer, A (Fischer, Alain); Leverger, G (Leverger, Guy); Latour, S (Latour, Sylvain)	EMBO MOLECULAR MEDICINE Volume: 10 Issue: 2 Pages: 188-199 DOI: 10.15252/emmm.201708292 Published: FEB 2018
Mutations in CFAP43 and CFAP44 cause male infertility and flagellum defects in Trypanosoma and human	Thierry-Mieg, N (Thierry-Mieg, Nicolas); Conne, B (Conne, Beatrice); Dacheux, D (Dacheux, Denis); Landrein, N (Landrein, Nicolas); Schmitt, A (Schmitt, Alain); Stouvenel, L (Stouvenel, Laurence); Lores, P (Lores, Patrick); El Khouri, E (El Khouri, Elma); Bottari, SP (Bottari, Serge P.); Faure, J (Faure, Julien); Wolf, JP (Wolf, Jean-Philippe); Pernet-Gallay, K (Pernet-Gallay, Karin); Escoffier, J (Escoffier, Jessica); Gourabi, H (Gourabi, Hamid); Robinson, DR (Robinson, Derrick R.); Nef, S (Nef, Serge); Dulioust, E (Dulioust, Emmanuel); Zouari, R (Zouari, Raoudha); Bonhivers, M (Bonhivers, Melanie); Toure, A (Toure, Aminata); Arnoult, C (Arnoult, Christophe); Ray, PF (Ray, Pierre F.)	NATURE COMMUNICATIONS Volume: 9 Article Number: 686 DOI: 10.1038/s41467-017-02792-7 Published: FEB 15 2018
Genetic disruption of the oncogenic HMGA2-PLAG1-IGF2 pathway causes fetal growth restriction	Habib, WA (Habib, Walid Abi); Brioude, F (Brioude, Frederic); Edouard, T (Edouard, Thomas); Bennett, JT (Bennett, James T.); Lienhardt-Roussie, A (Lienhardt-Roussie, Anne); Tixier, F (Tixier, Frederique); Salem, J (Salem, Jennifer); Yuen, T (Yuen, Tony); Azzi, S (Azzi, Salah); Le Bouc, Y (Le Bouc, Yves); Harbison, MD (Harbison, Madeleine D.); Netchine, I (Netchine, Irene)	GENETICS IN MEDICINE Volume: 20 Issue: 2 Pages: 250-258 DOI: 10.1038/gim.2017.105 Published: FEB 2018
Identification of PITX3 mutations in individuals with various ocular developmental defects	Seco, CZ (Seco, Celia Zazo); Plaisancie, J (Plaisancie, Julie); Lupasco, T (Lupasco, Tatiana); Michot, C (Michot, Caroline); Pechmeja, J (Pechmeja, Jacmine); Delanne, J (Delanne, Julian); Cottereau, E (Cottereau, Edouard); Ayuso, C (Ayuso, Carmen); Corton, M (Corton, Marta); Calvas, P (Calvas, Patrick); Ragge, N (Ragge, Nicola); Chassaing, N (Chassaing, Nicolas)	OPHTHALMIC GENETICS Volume: 39 Issue: 3 Pages: 314-320 DOI: 10.1080/13816810.2018.1430243 Published: 2018
Using multi-criteria decision analysis to appraise orphan drugs: a systematic review	Friedmann, C (Friedmann, Carlotta); Levy, P (Levy, Pierre); Hensel, P (Hensel, Paul); Hiligsmann, M (Hiligsmann, Mickael)	EXPERT REVIEW OF PHARMACOECONOMICS & OUTCOMES RESEARCH Volume: 18 Issue: 2 Pages: 135-146 DOI: 10.1080/14737167.2018.1414603 Published: 2018
Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults (vol 75, pg 105, 2017)	Charif, M (Charif, M.); Nasca, A (Nasca, A.); Thompson, K (Thompson, K.	JAMA NEUROLOGY Volume: 75 Issue: 1 Pages: 133-133 DOI: 10.1001/jamaneurol.2017.4634
Ciliogenesis and cell cycle alterations contribute to KIF2A-related malformations of cortical development	(Drouot, Nathalie); Reilly, ML (Reilly, Madeline Louise); Francis, F (Francis, Fiona); Benmerah, A (Benmerah, Alexandre); Bahi-Buisson, N (Bahi-Buisson, Nadia); Belvindrah, R (Belvindrah, Richard); Nguyen, L (Nguyen, Laurent); Godin, JD (Godin, Juliette D.); Chelly, J (Chelly, Jamel); Hinckelmann, MV (Hinckelmann, Maria-Victoria)	HUMAN MOLECULAR GENETICS Volume: 27 Issue: 2 Pages: 224-238 DOI: 10.1093/hmg/ddx384 Published: JAN 15 2018

Title	Author(s)	Source
Affected female carriers of MTM1 mutations display a wide spectrum of clinical and pathological involvement: delineating diagnostic clues	Biancalana, V (Biancalana, Valerie); Scheidecker, S (Scheidecker, Sophie); Miguet, M (Miguet, Marguerite); Laquerriere, A (Laquerriere, Annie); Romero, NB (Romero, Norma B.); Stojkovic, T (Stojkovic, Tanya); Neto, OA (Neto, Osorio Abath); Mercier, S (Mercier, Sandra); Voermans, N (Voermans, Nicol); Tanner, L (Tanner, Laura); Rogers, C (Rogers, Curtis); Ollagnon-Roman, E (Ollagnon-Roman, Elisabeth); Roper, H (Roper, Helen); Boutte, C (Boutte, Celia); Ben-Shachar, S (Ben-Shachar, Shay); Lornage, X (Lornage, Xaviere); Vasli, N (Vasli, Nasim); Schaefer, E (Schaefer, Elise); Laforet, P (Laforet, Pascal); Pouget, J (Pouget, Jean); Moerman, A (Moerman, Alexandre); Pasquier, L (Pasquier, Laurent); Marcorelle, P (Marcorelle, Pascale); Magot, A (Magot, Armelle); Kusters, B (Kusters, Benno); Streichenberger, N (Streichenberger, Nathalie); Tranchant, C (Tranchant, Christine); Dondaine, N (Dondaine, Nicolas); Schneider, R (Schneider, Raphael); Gasnier, C (Gasnier, Claire); Calmels, N (Calmels, Nadege); Kremer, V (Kremer, Valerie); Nguyen, K (Nguyen, Karine); Perrier, J (Perrier, Julie); Kamsteeg, EJ (Kamsteeg, Erik Jan); Carlier, P (Carlier, Pierre); Carlier, RY (Carlier, Robert-Yves); Thompson, J (Thompson, Julie); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Fardeau, M (Fardeau, Michel); Zanoteli, E (Zanoteli, Edmar); Eymard, B (Eymard, Bruno); Laporte, J (Laporte, Jocelyn)	ACTA NEUROPATHOLOGICA Volume: 134 Issue: 6 Pages: 889-904 DOI: 10.1007/s00401-017-1748-0 Published: DEC 2017
Autism spectrum disorder recurrence, resulting of germline mosaicism for a CHD2 gene missense variant	Lebrun, N (Lebrun, N.); Parent, P (Parent, P.); Gendras, J (Gendras, J.); Billuart, P (Billuart, P.); Poirier, K (Poirier, K.); Bienvenu, T (Bienvenu, T.)	CLINICAL GENETICS Volume: 92 Issue: 6 Pages: 669-670 DOI: 10.1111/cge.13073 Published: DEC 2017
Normal human adipose tissue functions and differentiation in patients with biallelic LPIN1 inactivating mutations	Pelosi, M (Pelosi, Michele); Testet, E (Testet, Eric); Le Lay, S (Le Lay, Soazig); Dugail, I (Dugail, Isabelle); Tang, XY (Tang, Xiaoyun); Mabilleau, G (Mabilleau, Guillaume); Hamel, Y (Hamel, Yamina); Madrange, M (Madrange, Marine); Blanc, T (Blanc, Thomas); Odent, T (Odent, Thierry); McMullen, TPW (McMullen, Todd P. W.); Alfo, M (Alfo, Marco); Brindley, DN (Brindley, David N.); de Lonlay, P (de Lonlay, Pascale)	JOURNAL OF LIPID RESEARCH Volume: 58 Issue: 12 Pages: 2348-2364 DOI: 10.1194/jlr.P075440 Published: DEC 2017
A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations	(Kivuva, Emma); Scott, RH (Scott, Richard H.); Rendon, A (Rendon, Augusto); Munnich, A (Munnich, Arnold); Newman, W (Newman, William); Kerr, B (Kerr, Bronwyn); Besmond, C (Besmond, Claude); Rosenfeld, JA (Rosenfeld, Jill A.); Amiel, J (Amiel, Jeanne); Field, M (Field, Michael); Gecz, J (Gecz, Jozef)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 101 Issue: 6 Pages: 995-1005 DOI: 10.1016/j.ajhg.2017.10.009 Published: DEC 7 2017
Genetic Characterization of a French Cohort of GNE-mutation negative inclusion body myopathy patients with exome sequencing	Cerino, M (Cerino, Mathieu); Gorokhova, S (Gorokhova, Svetlana); Laforet, P (Laforet, Pascal); Ben Yaou, R (Ben Yaou, Rabah); Salort-Campana, E (Salort-Campana, Emmanuelle); Pouget, J (Pouget, Jean); Attarian, S (Attarian, Shahram); Eymard, B (Eymard, Bruno); Deleuze, JF (Deleuze, Jean-Francois); Boland, A (Boland, Anne); Behin, A (Behin, Anthony); Stojkovic, T (Stojkovic, Tanya); Bonne, G (Bonne, Gisele); Levy, N (Levy, Nicolas); Bartoli, M (Bartoli, Marc); Krahn, M (Krahn, Martin)	MUSCLE & NERVE Volume: 56 Issue: 5 Pages: 993-997 DOI: 10.1002/mus.25638 Published: NOV 2017
Sclérodermie systémique et difficultés professionnelles : résultats d'une enquête prospective **Systemic sclerosis and occupational difficulties: Results of a prospective study**	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 Nov;38(11):718-724. doi: 10.1016/j.revmed.2017.06.006. Epub 2017 Jul 10.
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
Common and variable clinical, histological, and imaging findings of recessive RYR1-related centronuclear myopathy patients	Neto, OA (Neto, Osorio Abath); Moreno, CDM (Martins Moreno, Cristiane de Araujo); Malfatti, E (Malfatti, Edoardo); Donkervoort, S (Donkervoort, Sandra); Bohm, J (Bohm, Johann); Guimaraes, JB (Guimaraes, Julio Brandao); Foley, AR (Foley, A. Reghan); Mohassel, P (Mohassel, Payam); Dastgir, J (Dastgir, Jahannaz); Bharucha-Goebel, DX (Bharucha-Goebel, Diana Xerxes); Monges, S (Monges, Soledad); Lubieniecki, F (Lubieniecki, Fabiana); Collins, J (Collins, James); Medne, L (Medne, Livija); Santi, M (Santi, Mariarita); Yum, S (Yum, Sabrina); Banwell, B (Banwell, Brenda); Salort-Campana, E (Salort-Campana, Emmanuelle); Rendu, J (Rendu, John); Faure, J (Faure, Julien); Yis, U (Yis, Uluc); Eymard, B (Eymard, Bruno); Cheraud, C (Cheraud, Chrystel); Schneider, R (Schneider, Raphael); Thompson, J (Thompson, Julie); Lornage, X (Lornage, Xaviere); Mesrob, L (Mesrob, Lilia); Lechner, D (Lechner, Doris); Boland, A (Boland, Anne); Deleuze, JF (Deleuze, Jean-Francois); Reed, UC (Reed, Umbertina Conti); Oliveira, ASB (Bulle Oliveira, Acary Souza); Biancalana, V (Biancalana, Valerie); Romero, NB (Romero, Norma B.); Bonnemann, CG (Bonnemann, Carsten G.); Laporte, J (Laporte, Jocelyn); Zanoteli, E (Zanoteli, Edmar)	NEUROMUSCULAR DISORDERS Volume: 27 Issue: 11 Pages: 975-985 DOI: 10.1016/j.nmd.2017.05.016 Published: NOV 2017

Title	Author(s)	Source
Optimized approach for the identification of highly efficient correctors of nonsense mutations in human diseases	Benhabiles, H (Benhabiles, Hana); Gonzalez-Hilarion, S (Gonzalez-Hilarion, Sara); Amand, S (Amand, Severine); Bailly, C (Bailly, Christine); Prevotat, A (Prevotat, Anne); Reix, P (Reix, Philippe); Hubert, D (Hubert, Dominique); Adriaenssens, E (Adriaenssens, Eric); Rebuffat, S (Rebuffat, Sylvie); Tulasne, D (Tulasne, David); Lejeune, F (Lejeune, Fabrice)	PLOS ONE Volume: 12 Issue: 11 Article Number: e0187930 DOI: 10.1371/journal.pone.0187930 Published: NOV 13 2017
Juvenile-Onset Diabetes and Congenital Cataract: «Double-Gene» Mutations Mimicking a Syndromic Diabetes Presentation	Lenfant, C (Lenfant, Caroline); Baz, P (Baz, Patrick); Degavre, A (Degavre, Anne); Philippi, A (Philippi, Anne); Senee, V (Senee, Valerie); Vandiedonck, C (Vandiedonck, Claire); Derbois, C (Derbois, Celine); Nicolino, M (Nicolino, Marc); Zalloua, P (Zalloua, Pierre); Julier, C (Julier, Cecile)	GENES Volume: 8 Issue: 11 Article Number: 309 DOI: 10.3390/genes8110309 Published: NOV 2017
Mutations in DNM1L, as in OPA1, result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission	Procaccio, Patrizia Amati-Bonneau, Pascal Reynier, Stephanie Leruez, Raphael Calmon, Nathalie Boddaert, Benoit Funalot, Marlene Rio, Didier Bouccara, Isabelle Meunier, Hiromi Sesaki, Josseline Kaplan, Christian P. Hamel, Jean-Michel Rozet and Guy Lenaers	BRAIN 2017 Oct 1;140(10):2586-2596. doi: 10.1093/brain/awx219.
CASQ1 mutations impair calsequestrin polymerization and cause tubular aggregate myopathy	Chevessier Catherine Birck Simona Zanotti Paola Cudia Monica Bulla· Florence Granger Mai Thao Bui Maxime Sartori Christiane Schneider-Gold Edoardo Malfatti Norma B. Romero Marina Mora Jocelyn Laporte	ACTA NEUROPATHOLOGICA 2017 Oct 16. doi: 10.1007/s00401-017-1775-x
WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells	Alain); Martinovic, J (Martinovic, Jelena); Bidat, L (Bidat, Laurent); Rio, M (Rio, Marlene); Lyonnet, S (Lyonnet, Stanislas); Reilly, ML (Reilly, M. Louise); Boddaert, N (Boddaert, Nathalie); Jenneson-Liver, M (Jenneson-Liver, Melanie); Motte, J (Motte, Jacques); Doco-Fenzy, M (Doco-Fenzy, Martine); Chelly, J (Chelly, Jamel); Attie-Bitach, T (Attie-Bitach, Tania); Simons, M (Simons, Matias); Cantagrel, V (Cantagrel, Vincent); Passemard, S (Passemard, Sandrine); Baffet, A (Baffet, Alexandre); Thomas, S (Thomas, Sophie); Bahi-Buisson, N (Bahi-Buisson, Nadia)	BRAIN Volume: 140 Pages: 2597-2609 DOI: 10.1093/brain/awx203 Part: 10 Published: OCT 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	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
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
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
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

Title	Author(s)	Source
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

Title	Author(s)	Source
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

Title	Author(s)	Source
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

Title	Author(s)	Source
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

Title	Author(s)	Source
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

Title	Author(s)	Source
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

Title	Author(s)	Source
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

Title	Author(s)	Source
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

Title	Author(s)	Source
Autosomal dominant polycystic kidney disease: the changing face of clinical management	Ong, ACM (Ong, Albert C. M.); Devuyst, O (Devuyst, Olivier); Knebelmann, B (Knebelmann, Bertrand); Walz, G (Walz, Gerd); Group Author(s): ERA-EDTA Working Grp Inherited Kid	LANCET Volume: 385 Issue: 9981 Pages: 1993-2002 Published: MAY 16 2015
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
Identification of Variants in the 4q35 Gene FAT1 in Patients with a Facioscapulohumeral Dystrophy-Like Phenotype	Puppo, F (Puppo, Francesca); Dionnet, E (Dionnet, Eugenie); Gaillard, MC (Gaillard, Marie-Cecile); Gaildrat, P (Gaildrat, Pascaline); Castro, C (Castro, Christel); Vovan, C (Vovan, Catherine); Bertaux, K (Bertaux, Karine); Bernard, R (Bernard, Rafaelle); Attarian, S (Attarian, Shahram); Goto, K (Goto, Kanako); Nishino, I (Nishino, Ichizo); Hayashi, Y (Hayashi, Yukiko); Magdinier, F (Magdinier, Frederique); Krahn, M (Krahn, Martin); Helmbacher, F (Helmbacher, Francoise); Bartoli, M (Bartoli, Marc); Levy, N (Levy, Nicolas)	HUMAN MUTATION Volume: 36 Issue: 4 Pages: 443-453 DOI: 10.1002/humu.22760 Published: APR 2015
Premature Skin Aging Features Rescued by Inhibition of NADPH Oxidase Activity in XPC-Deficient Mice	Hosseini, M (Hosseini, Mohsen); Mahfouf, W (Mahfouf, Walid); Serrano-Sanchez, M (Serrano-Sanchez, Martin); Raad, H (Raad, Houssam); Harfouche, G (Harfouche, Ghida); Bonneu, M (Bonneu, Marc); Claverol, S (Claverol, Stephane); Mazurier, F (Mazurier, Frederic); Rossignol, R (Rossignol, Rodrigue); Taieb, A (Taieb, Alain); Rezvani, HR (Rezvani, Hamid Reza)	JOURNAL OF INVESTIGATIVE DERMATOLOGY Volume: 135 Issue: 4 Pages: 1108-1118 DOI: 10.1038/jid.2014.511 Published: APR 2015
XIAP deficiency syndrome in humans	Latour, S (Latour, Sylvain); Aguilar, C (Aguilar, Claire)	SEMINARS IN CELL & DEVELOPMENTAL BIOLOGY Volume: 39 Pages: 115-123 DOI: 10.1016/j.semcdb.2015.01.015 Published: MAR 2015
Functional variants of POC5 identified in patients with idiopathic scoliosis	Patten, SA (Patten, Shunmoogum A.); Margaritte-Jeannin, P (Margaritte-Jeannin, Patricia); Bernard, JC (Bernard, Jean-Claude); Alix, E (Alix, Eudeline); Labalme, A (Labalme, Audrey); Besson, A (Besson, Alicia); Girard, SL (Girard, Simon L.); Fendri, K (Fendri, Khaled); Fraisse, N (Fraisse, Nicolas); Biot, B (Biot, Bernard); Poizat, C (Poizat, Coline); Campan-Fournier, A (Campan-Fournier, Amandine); Abelin-Genevois, K (Abelin-Genevois, Kariman); Cunin, V (Cunin, Vincent); Zaouter, C (Zaouter, Charlotte); Liao, MJ (Liao, Meijiang); Lamy, R (Lamy, Raphaelle); Lesca, G (Lesca, Gaetan); Menassa, R (Menassa, Rita); Marcaillou, C (Marcaillou, Charles); Letexier, M (Letexier, Melanie); Sanlaville, D (Sanlaville, Damien); Berard, J (Berard, Jerome); Rouleau, GA (Rouleau, Guy A.); Clerget-Darpoux, F (Clerget-Darpoux, Francoise); Drapeau, P (Drapeau, Pierre); Moldovan, F (Moldovan, Florina); Edery, P (Edery, Patrick)	JOURNAL OF CLINICAL INVESTIGATION Volume: 125 Issue: 3 Pages: 1124-1128 DOI: 10.1172/JCI77262 Published: MAR 2015
Oxidative and Energy Metabolism as Potential Clues for Clinical Heterogeneity in Nucleotide Excision Repair Disorders	Hosseini, M (Hosseini, Mohsen); Ezzedine, K (Ezzedine, Khaled); Taieb, A (Taieb, Alain); Rezvani, HR (Rezvani, Hamid R.)	JOURNAL OF INVESTIGATIVE DERMATOLOGY Volume: 135 Issue: 2 Pages: 341-351 DOI: 10.1038/jid.2014.365 Published: FEB 2015
Inherited STING-activating mutation underlies a familial inflammatory syndrome with lupus-like manifestations	Jeremiah, N (Jeremiah, Nadia); Neven, B (Neven, Benedicte); Gentili, M (Gentili, Matteo); Callebaut, I (Callebaut, Isabelle); Maschalidi, S (Maschalidi, Sophia); Stolzenberg, MC (Stolzenberg, Marie-Claude); Goudin, N (Goudin, Nicolas); Fremond, ML (Fremond, Marie-Louis); Nitschke, P (Nitschke, Patrick); Molina, TJ (Molina, Thierry J.); Blanche, S (Blanche, Stephane); Picard, C (Picard, Capucine); Rice, GI (Rice, Gillian I.); Crow, YJ (Crow, Yanick J.); Manel, N (Manel, Nicolas); Fischer, A (Fischer, Alain); Bader-Meunier, B (Bader-Meunier, Brigitte); Rieux-Laucat, F (Rieux-Laucat, Frederic)	JOURNAL OF CLINICAL INVESTIGATION Volume: 124 Issue: 12 Pages: 5516-5520 DOI: 10.1172/JCI79100 Published: DEC 2014
Haploinsufficiency of Dmxl2, Encoding a Synaptic Protein, Causes Infertility Associated with a Loss of GnRH Neurons in Mouse	Tata, B (Tata, Brooke); Huijbregts, L (Huijbregts, Lukas); Jacquier, S (Jacquier, Sandrine); Csaba, Z (Csaba, Zsolt); Genin, E (Genin, Emmanuelle); Meyer, V (Meyer, Vincent); Leka, S (Leka, Sofia); Dupont, J (Dupont, Joelle); Charles, P (Charles, Perrine); Chevenne, D (Chevenne, Didier); Carel, JC (Carel, Jean-Claude); Leger, J (Leger, Juliane); de Roux, N (de Roux, Nicolas)	PLOS BIOLOGY Volume: 12 Issue: 9 Article Number: e1001952 DOI: 10.1371/journal.pbio.1001952 Published: SEP 2014

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JAGN1 deficiency causes aberrant myeloid cell homeostasis and congenital neutropenia	Boztug, K (Boztug, Kaan); Jarvinen, PM (Jaervinen, Paeivi M.); Salzer, E (Salzer, Elisabeth); Racek, T (Racek, Tomas); Monch, S (Moench, Sebastian); Garncarz, W (Garncarz, Wojciech); Gertz, EM (Gertz, E. Michael); Schaffer, AA (Schaeffer, Alejandro A.); Antonopoulos, A (Antonopoulos, Aristotelis); Haslam, SM (Haslam, Stuart M.); Schieck, L (Schieck, Lena); Puchalka, J (Puchalka, Jacek); Diestelhorst, J (Diestelhorst, Jana); Appaswamy, G (Appaswamy, Giridharan); Lescoeur, B (Lescoeur, Brigitte); Giambruno, R (Giambruno, Roberto); Bigenzahn, JW (Bigenzahn, Johannes W.); Elling, U (Elling, Ulrich); Pfeifer, D (Pfeifer, Dietmar); Conde, CD (Conde, Cecilia Dominguez); Albert, MH (Albert, Michael H.); Welte, K (Welte, Karl); Brandes, G (Brandes, Gudrun); Sherkat, R (Sherkat, Roya); ten Bosch, JV (ten Bosch, Jutte van der Werff); Rezaei, N (Rezaei, Nima); Etzioni, A (Etzioni, Amos); Bellanne-Chantelot, C (Bellanne-Chantelot, Christine); Superti-Furga, G (Superti-Furga, Giulio); Penninger, JM (Penninger, Josef M.); Bennett, KL (Bennett, Keiryn L.); von Blume, J (von Blume, Julia); Dell, A (Dell, Anne); Donadieu, J (Donadieu, Jean); Klein, C (Klein, Christoph)	NATURE GENETICS Volume: 46 Issue: 9 Pages: 1021-+ DOI: 10.1038/ng.3069 Published: SEP 2014
Nuclear matrix, nuclear envelope and premature aging syndromes in a translational research perspective	Cau, P (Cau, Pierre); Navarro, C (Navarro, Claire); Harhouri, K (Harhouri, Karim); Roll, P (Roll, Patrice); Sigaudy, S (Sigaudy, Sabine); Kaspi, E (Kaspi, Elise); Perrin, S (Perrin, Sophie); De Sandre-Giovannoli, A (De Sandre-Giovannoli, Annachiara); Levy, N (Levy, Nicolas)	SEMINARS IN CELL & DEVELOPMENTAL BIOLOGY Volume: 29 Pages: 125-147 DOI: 10.1016/j.semcdb.2014.03.021 Published: MAY 2014
Mutations in QARS, encoding glutaminyl-tRNA synthetase, cause progressive microcephaly, cerebral-cerebellar atrophy, and intractable seizures	Zhang, XC (Zhang, Xiaochang); Ling, JQ (Ling, Jiqiang); Barcia, G (Barcia, Giulia); Jing, LL (Jing, Lili); Wu, J (Wu, Jiang); Barry, BJ (Barry, Brenda J.); Mochida, GH (Mochida, Ganeshwaran H.); Hill, RS (Hill, R. Sean); Weimer, JM (Weimer, Jill M.); Stein, Q (Stein, Quinn); Poduri, A (Poduri, Annapurna); Partlow, JN (Partlow, Jennifer N.); Ville, D (Ville, Dorothee); Dulac, O (Dulac, Olivier); Yu, TW (Yu, Tim W.); Lam, ATN (Lam, Anh-Thu N.); Servattalab, S (Servattalab, Sarah); Rodriguez, J (Rodriguez, Jacqueline); Boddaert, N (Boddaert, Nathalie); Munnich, A (Munnich, Arnold); Colleaux, L (Colleaux, Laurence); Zon, LI (Zon, Leonard I.); Soll, D (Soell, Dieter); Walsh, CA (Walsh, Christopher A.); Nabbout, R (Nabbout, Rima)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 94 Issue: 4 Pages: 547-558 DOI: 10.1016/j.ajhg.2014.03.003 Published: APR 3 2014
Finger creases lend a hand in Kabuki syndrome	Michot, C (Michot, Caroline); Corsini, C (Corsini, Carole); Sanlaville, D (Sanlaville, Damien); Baumann, C (Baumann, Clarisse); Toutain, A (Toutain, Annick); Philip, N (Philip, Nicole); Busa, T (Busa, Tiffany); Holder, M (Holder, Muriel); Faivre, L (Faivre, Laurence); Odent, S (Odent, Sylvie); Delrue, MA (Delrue, Marie-Ange); Till, M (Till, Marianne); Jacquemont, ML (Jacquemont, Marie-Line); Cordier, MP (Cordier, Marie-Pierre); Goldenberg, A (Goldenberg, Alice); Sanchez, E (Sanchez, Elodie); Alix, E (Alix, Eudeline); Poisson, S (Poisson, Sylvain); Kayirangwa, H (Kayirangwa, Honorine); Lacombe, D (Lacombe, Didier); Gilbert-Dussardier, B (Gilbert-Dussardier, Brigitte); Pelet, A (Pelet, Anna); Roume, J (Roume, Joelle); Jacquette, A (Jacquette, Aurelia); Isidor, B (Isidor, Bertrand); Giuliano, F (Giuliano, Fabienne); Burglen, L (Burglen, Lydie); Fradin, M (Fradin, Melanie); Schaefer, E (Schaefer, Elise); Alembick, Y (Alembick, Yves); Doray, B (Doray, Berenice); Moncla, A (Moncla, Anne); Heron, D (Heron, Delphine); Willems, M (Willems, Marjolaine); Pinson, L (Pinson, Lucile); Sang, KHLQ (Sang, Kim Hanh Le Quan); Le Merrer, M (Le Merrer, Martine); Cormier-Daire, V (Cormier-Daire, Valerie); Sarda, P (Sarda, Pierre); Amiel, J (Amiel, Jeanne); Lyonnet, S (Lyonnet, Stanislas); Genevieve, D (Genevieve, David)	EUROPEAN JOURNAL OF MEDICAL GENETICS Volume: 56 Issue: 10 Pages: 556-560 DOI: 10.1016/j.ejmg.2013.07.005 Published: OCT 2013
Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly	Poirier, K (Poirier, Karine); Lebrun, N (Lebrun, Nicolas); Broix, L (Broix, Loic); Tian, G (Tian, Guoling); Saillour, Y (Saillour, Yoann); Boscheron, C (Boscheron, Cecile); Parrini, E (Parrini, Elena); Valence, S (Valence, Stephanie); Saint Pierre, B (Saint Pierre, Benjamin); Oger, M (Oger, Madison); Lacombe, D (Lacombe, Didier); Genevieve, D (Genevieve, David); Fontana, E (Fontana, Elena); Darra, F (Darra, Transcesca); Cances, C (Cances, Claude); Barth, M (Barth, Magalie); Bonneau, D (Bonneau, Dominique); Dalla Bernadina, B (Dalla Bernadina, Bernardo); N'Guyen, S (N'Guyen, Sylvie); Gitiaux, C (Gitiaux, Cyril); Parent, P (Parent, Philippe); Portes, VD (Portes, Vincent des); Pedespan, JM (Pedespan, Jean Michel); Legrez, V (Legrez, Victoire); Castelnau-Ptakine, L (Castelnau-Ptakine, Laetitia); Nitschke, P (Nitschke, Patrick); Hieu, T (Hieu, Thierry); Masson, C (Masson, Cecile); Zelenika, D (Zelenika, Diana); Andrieux, A (Andrieux, Annie); Francis, F (Francis, Fiona); Guerrini, R (Guerrini, Renzo); Cowan, NJ (Cowan, Nicholas J.); Bahi-Buisson, N (Bahi-Buisson, Nadia); Chelly, J (Chelly, Jamel)	NATURE GENETICS Volume: 45 Issue: 6 Pages: 639-+ DOI: 10.1038/ng.2613 Published: JUN 2013
Constitutive Activation of the Calcium Sensor STIM1 Causes Tubular-Aggregate Myopathy	Bohm, J (Boehm, Johann); Chevessier, F (Chevessier, Frederic); De Paula, AM (De Paula, Andre Maues); Koch, C (Koch, Catherine); Attarian, S (Attarian, Shahram); Feger, C (Feger, Claire); Hantai, D (Hantai, Daniel); Laforet, P (Laforet, Pascal); Ghorab, K (Ghorab, Karima); Vallat, JM (Vallat, Jean-Michel); Fardeau, M (Fardeau, Michel); Figarella-Branger, D (Figarella-Branger, Dominique); Pouget, J (Pouget, Jean); Romero, NB (Romero, Norma B.); Koch, M (Koch, Marc); Ebel, C (Ebel, Claudine); Levy, N (Levy, Nicolas); Krahn, M (Krahn, Martin); Eymard, B (Eymard, Bruno); Bartoli, M (Bartoli, Marc); Laporte, J (Laporte, Jocelyn)	AMERICAN JOURNAL OF HUMAN GENETICS Volume: 92 Issue: 2 Pages: 271-278 DOI: 10.1016/j.ajhg.2012.12.007 Published: FEB 7 2013