SUPPLEMENTAL INFORMATION

Table S1 – Genetic discoveries in cardiovascular disorders made with next-generation sequencing.

Disease	Inheritance	Gene(s)	Approach	References
Cardiomyopathies				
Dilated cardiomyopathy	AR	*GATAD1*	homozyosity mapping, exome	1
Dilated cardiomyopathy	AD	*BAG3*	CNV analysis, exome	2
Dilated cardiomyopathy	AD	*TTN*	linkage analysis, exome	3
Dilated cardiomyopathy	AD	*DES*	exome	4
Dilated cardiomyopathy	AD	*RBM20*	exome	5
Dilated cardiomyopathy	AD	*LMNA*	exome	6
Dilated cardiomyopathy	AD	*TNNT2*	exome	7
Hypertrophic cardiomyopathy	AD	*Multiple*	41 gene panel	8
Hypertrophic cardiomyopathy	X-linked	*FHL1*	exome	9
X-linked cardiomegaly	X-linked	*CLIC2*	X chromosome exons	10
Left ventricular noncompaction	digenic	*MYH7B, ITGA7*	exome	11
Infantile cardiomyopathy	mitochondrial	*MRPL44*	exome	12
Congenital heart disease				
Pleotropic congenital heart disease	AD	*MYH6*	exome	13
Multiple types	*de novo*	histone-modifying genes	exome	14
Isolated truncus arteriosus	AR	*PLXND1*	homozyosity mapping, exome	15
Atrial septal defect	AD	*ACTC1*	exome	16
Arrhythmia				
Long-QT syndrome	*de novo*	*CALM1, CALM2*	exome	17
Long-QT syndrome	AD	*CACNA1C*	exome	18
Idiopathic ventricular fibrillation	AD	*CALM1*	exome	19
Cardiac conduction disease	AD	*LMNA*	exome	20
Sudden unexplained death	unknown	*MYH7*	exome	21
Sudden unexplained death	unknown	*KCNQ1, KCNH2*	exome	22

Table 3 - *continued*

Disease	Inheritance	Gene(s)	Approach	Reference
Dyslipidemia				
Hypercholesterolemia	AR or *de novo*	*ABCG5*	whole genome	23
Hypercholesterolemia	not defined	*LDLR, APOB*	exome	24
Hypercholesterolemia	AD	*APOE*	exome	25
Hypercholesterolemia	AD	*APOE*	exome	26
Hypercholesterolemia	AR	*LIPA*	exome	27
Hypercholesterolemia and CAD	AD	*ST6GALNAC5*	linkage analysis, exome	28
Hypertriglyceridemia	AD	*SLC25A40*	linkage analysis, exome	29
Vascular disorders				
Familial thoracic aortic aneurysm	AD	*SMAD3*	exome	30
Familial thoracic aortic aneurysm	AD	*TGFB2*	linkage analysis, exome	31
Familial thoracic aortic aneurysm	AD	*PRKG1*	exome	32
Primary lymphedema	AD	*VEGFC*	exome	33
Vascular anomaly syndrome	not defined	*BMP9*	exome	34
Hypertension				
Pseudohypoaldosteronism II	AD	*KLHL3, CUL3*	exome	35
Familial hyperkalemic hypertension	AD	*KLHL3*	exome	36
Pulmonary arterial hypertension	AD	*CAV1*	exome	37
Aldosterone-producing adenoma	somatic, AD	*KCNJ5*	exome	38
Aldosterone-producing adenoma	somatic, *de novo*	*CACNA1D*	exome	39
Aldosterone-producing adenoma	somatic	*ATP1A1, ATP2B3*	exome	40,41
Syndromes with cardiac findings				
Cantú syndrome	AD	*ABCC9*	exome	42,43
CHIME syndrome	AR	*PIGL*	exome	44
Noonan syndrome	AD	*PTPN11*	exome	45
Marfan syndrome	AD	*LRP1*	exome	46
Phosphoglucomutase 1 deficiency	AR	*PGM1*	exome	47

Abbreviations: AR, autosomal recessive; AD, autosomal dominant; CAD, coronary artery disease

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SUPPLEMENTAL REFERENCES

1. Theis JL, Sharpe KM, Matsumoto ME, Chai HS, Nair AA, Theis JD, de AM, Wieben ED, Michels VV, Olson TM. Homozygosity mapping and exome sequencing reveal GATAD1 mutation in autosomal recessive dilated cardiomyopathy. *Circ Cardiovasc Genet.* 2011;4:585-594.

2. Norton N, Li D, Rieder MJ, Siegfried JD, Rampersaud E, Zuchner S, Mangos S, Gonzalez-Quintana J, Wang L, McGee S, Reiser J, Martin E, Nickerson DA, Hershberger RE. Genome-wide studies of copy number variation and exome sequencing identify rare variants in BAG3 as a cause of dilated cardiomyopathy. *Am J Hum Genet.* 2011;88:273-282.

3. Norton N, Li D, Rampersaud E, Morales A, Martin ER, Zuchner S, Guo S, Gonzalez M, Hedges DJ, Robertson PD, Krumm N, Nickerson DA, Hershberger RE. Exome sequencing and genome-wide linkage analysis in 17 families illustrate the complex contribution of TTN truncating variants to dilated cardiomyopathy. *Circ Cardiovasc Genet.* 2013;6:144-153.

4. Tse HF, Ho JC, Choi SW, Lee YK, Butler AW, Ng KM, Siu CW, Simpson MA, Lai WH, Chan YC, Au KW, Zhang J, Lay KW, Esteban MA, Nicholls JM, Colman A, Sham PC. Patient-specific induced-pluripotent stem cells-derived cardiomyocytes recapitulate the pathogenic phenotypes of dilated cardiomyopathy due to a novel DES mutation identified by whole exome sequencing. *Hum Mol Genet.* 2013;22:1395-1403.

5. Wells QS, Becker JR, Su YR, Mosley JD, Weeke P, D'Aoust L, Ausborn NL, Ramirez AH, Pfotenhauer JP, Naftilan AJ, Markham L, Exil V, Roden DM, Hong CC. Whole exome sequencing identifies a causal RBM20 mutation in a large pedigree with familial dilated cardiomyopathy. *Circ Cardiovasc Genet.* 2013;6:317-326.

6. Roncarati R, Viviani AC, Krawitz P, Lattanzi G, von KY, Perrot A, di PE, Papa L, Portararo P, Columbaro M, Forni A, Faggian G, Condorelli G, Robinson PN. Doubly heterozygous LMNA and TTN mutations revealed by exome sequencing in a severe form of dilated cardiomyopathy. *Eur J Hum Genet.* 2013;21:1105-1111.

7. Campbell N, Sinagra G, Jones KL, Slavov D, Gowan K, Merlo M, Carniel E, Fain PR, Aragona P, Di LA, Mestroni L, Taylor MR. Whole exome sequencing identifies a troponin T mutation hot spot in familial dilated cardiomyopathy. *PLoS ONE.* 2013;8:e78104.

SUPPLEMENTAL INFORMATION

8. Lopes LR, Zekavati A, Syrris P, Hubank M, Giambartolomei C, Dalageorgou C, Jenkins S, McKenna W, Plagnol V, Elliott PM. Genetic complexity in hypertrophic cardiomyopathy revealed by high-throughput sequencing. *J Med Genet.* 2013;50:228-239.

9. Hartmannova H, Kubanek M, Sramko M, Piherova L, Noskova L, Hodanova K, Stranecky V, Pristoupilova A, Sovova J, Marek T, Maluskova J, Ridzon P, Kautzner J, Hulkova H, Kmoch S. Isolated X-linked hypertrophic cardiomyopathy caused by a novel mutation of the four-and-a-half LIM domain 1 gene. *Circ Cardiovasc Genet.* 2013;6:543-551.

10. Takano K, Liu D, Tarpey P, Gallant E, Lam A, Witham S, Alexov E, Chaubey A, Stevenson RE, Schwartz CE, Board PG, Dulhunty AF. An X-linked channelopathy with cardiomegaly due to a CLIC2 mutation enhancing ryanodine receptor channel activity. *Hum Mol Genet.* 2012;21:4497-4507.

11. Esposito T, Sampaolo S, Limongelli G, Varone A, Formicola D, Diodato D, Farina O, Napolitano F, Pacileo G, Gianfrancesco F, Di IG. Digenic mutational inheritance of the integrin alpha 7 and the myosin heavy chain 7B genes causes congenital myopathy with left ventricular non-compact cardiomyopathy. *Orphanet J Rare Dis.* 2013;8:91.

12. Carroll CJ, Isohanni P, Poyhonen R, Euro L, Richter U, Brilhante V, Gotz A, Lahtinen T, Paetau A, Pihko H, Battersby BJ, Tyynismaa H, Suomalainen A. Whole-exome sequencing identifies a mutation in the mitochondrial ribosome protein MRPL44 to underlie mitochondrial infantile cardiomyopathy. *J Med Genet.* 2013;50:151-159.

13. Arrington CB, Bleyl SB, Matsunami N, Bonnell GD, Otterud BE, Nielsen DC, Stevens J, Levy S, Leppert MF, Bowles NE. Exome analysis of a family with pleiotropic congenital heart disease. *Circ Cardiovasc Genet.* 2012;5:175-182.

14. Zaidi S, Choi M, Wakimoto H, Ma L, Jiang J, Overton JD, Romano-Adesman A, Bjornson RD, Breitbart RE, Brown KK, Carriero NJ, Cheung YH, Deanfield J, DePalma S, Fakhro KA, Glessner J, Hakonarson H, Italia MJ, Kaltman JR, Kaski J, Kim R, Kline JK, Lee T, Leipzig J, Lopez A, Mane SM, Mitchell LE, Newburger JW, Parfenov M, Pe'er I, Porter G, Roberts AE, Sachidanandam R, Sanders SJ, Seiden HS, State MW, Subramanian S, Tikhonova IR, Wang W, Warburton D, White PS, Williams IA, Zhao H, Seidman JG, Brueckner M, Chung WK, Gelb BD, Goldmuntz E, Seidman CE, Lifton RP. De novo mutations in histone-modifying genes in congenital heart disease. *Nature.* 2013;498:220-223.

15. Ta-Shma A, Pierri CL, Stepensky P, Shaag A, Zenvirt S, Elpeleg O, Rein AJ. Isolated truncus arteriosus associated with a mutation in the plexin-D1 gene. *Am J Med Genet A.* 2013;161A:3115-3120.

16. Greenway SC, McLeod R, Hume S, Roslin NM, Alvarez N, Giuffre M, Zhan SH, Shen Y, Preuss C, Andelfinger G, Jones SJ, Gerull B. Exome sequencing identifies a novel variant in ACTC1 associated with familial atrial septal defect. *Can J Cardiol.* 2014;30:181-187.

17. Crotti L, Johnson CN, Graf E, De Ferrari GM, Cuneo BF, Ovadia M, Papagiannis J, Feldkamp MD, Rathi SG, Kunic JD, Pedrazzini M, Wieland T, Lichtner P, Beckmann BM, Clark T, Shaffer C, Benson DW, Kaab S, Meitinger T, Strom TM, Chazin WJ, Schwartz PJ, George AL, Jr. Calmodulin mutations associated with recurrent cardiac arrest in infants. *Circulation.* 2013;127:1009-1017.

18. Boczek NJ, Best JM, Tester DJ, Giudicessi JR, Middha S, Evans JM, Kamp TJ, Ackerman MJ. Exome sequencing and systems biology converge to identify novel mutations in the L-type calcium channel, CACNA1C, linked to autosomal dominant long QT syndrome. *Circ Cardiovasc Genet.* 2013;6:279-289.

19. Marsman RF, Barc J, Beekman L, Alders M, Dooijes D, van den WA, Ratbi I, Sefiani A, Bhuiyan ZA, Wilde AA, Bezzina CR. A mutation in CALM1 encoding calmodulin in familial idiopathic ventricular fibrillation in childhood and adolescence. *J Am Coll Cardiol.* 2014;63:259-266.

20. Lai CC, Yeh YH, Hsieh WP, Kuo CT, Wang WC, Chu CH, Hung CL, Cheng CY, Tsai HY, Lee JL, Tang CY, Hsu LA. Whole-exome sequencing to identify a novel LMNA gene mutation associated with inherited cardiac conduction disease. *PLoS ONE.* 2013;8:e83322.

21. Loporcaro CG, Tester DJ, Maleszewski JJ, Kruisselbrink T, Ackerman MJ. Confirmation of Cause and Manner of Death Via a Comprehensive Cardiac Autopsy Including Whole Exome Next-Generation Sequencing. *Arch Pathol Lab Med.* 2013.

22. Bagnall RD, Das KJ, Duflou J, Semsarian C. Exome analysis-based molecular autopsy in cases of sudden unexplained death in the young. *Heart Rhythm.* 2014;11:655-662.

23. Rios J, Stein E, Shendure J, Hobbs HH, Cohen JC. Identification by whole-genome resequencing of gene defect responsible for severe hypercholesterolemia. *Hum Mol Genet.* 2010;19:4313-4318.

24. Futema M, Plagnol V, Whittall RA, Neil HA, Humphries SE. Use of targeted exome sequencing as a diagnostic tool for Familial Hypercholesterolaemia. *J Med Genet.* 2012;49:644-649.

25. Marduel M, Ouguerram K, Serre V, Bonnefont-Rousselot D, Marques-Pinheiro A, Erik BK, Devillers M, Luc G, Lecerf JM, Tosolini L, Erlich D, Peloso GM, Stitziel N, Nitchke P, Jais JP, Abifadel M, Kathiresan S, Leren TP, Rabes JP, Boileau C, Varret M. Description of a large family with autosomal dominant hypercholesterolemia associated with the APOE p.Leu167del mutation. *Hum Mutat.* 2013;34:83-87.

26. Awan Z, Choi HY, Stitziel N, Ruel I, Bamimore MA, Husa R, Gagnon MH, Wang RH, Peloso GM, Hegele RA, Seidah NG, Kathiresan S, Genest J. APOE p.Leu167del mutation in familial hypercholesterolemia. *Atherosclerosis.* 2013;231:218-222.

27. Stitziel NO, Fouchier SW, Sjouke B, Peloso GM, Moscoso AM, Auer PL, Goel A, Gigante B, Barnes TA, Melander O, Orho-Melander M, Duga S, Sivapalaratnam S, Nikpay M, Martinelli N, Girelli D, Jackson RD, Kooperberg C, Lange LA, Ardissino D, McPherson R, Farrall M, Watkins H, Reilly MP, Rader DJ, de FU, Schunkert H, Erdmann J, Samani NJ, Charnas L, Altshuler D, Gabriel S, Kastelein JJ, Defesche JC, Nederveen AJ, Kathiresan S, Hovingh GK. Exome sequencing and directed clinical phenotyping diagnose cholesterol ester storage disease presenting as autosomal recessive hypercholesterolemia. *Arterioscler Thromb Vasc Biol.* 2013;33:2909-2914.

28. InanlooRahatloo K, Parsa AF, Huse K, Rasooli P, Davaran S, Platzer M, Kramer M, Fan JB, Turk C, Amini S, Steemers F, Gunderson K, Ronaghi M, Elahi E. Mutation in ST6GALNAC5 identified in family with coronary artery disease. *Sci Rep.* 2014;4:3595.

29. Rosenthal EA, Ranchalis J, Crosslin DR, Burt A, Brunzell JD, Motulsky AG, Nickerson DA, Wijsman EM, Jarvik GP. Joint linkage and association analysis with exome sequence data implicates SLC25A40 in hypertriglyceridemia. *Am J Hum Genet.* 2013;93:1035-1045.

30. Regalado ES, Guo DC, Villamizar C, Avidan N, Gilchrist D, McGillivray B, Clarke L, Bernier F, Santos-Cortez RL, Leal SM, Bertoli-Avella AM, Shendure J, Rieder MJ, Nickerson DA, Milewicz DM. Exome sequencing identifies SMAD3 mutations as a cause of familial thoracic aortic aneurysm and dissection with intracranial and other arterial aneurysms. *Circ Res.* 2011;109:680-686.

31. Boileau C, Guo DC, Hanna N, Regalado ES, Detaint D, Gong L, Varret M, Prakash SK, Li AH, d'Indy H, Braverman AC, Grandchamp B, Kwartler CS, Gouya L, Santos-Cortez RL, Abifadel M, Leal SM, Muti C, Shendure J, Gross MS, Rieder MJ, Vahanian A, Nickerson DA, Michel JB, Jondeau G, Milewicz DM. TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. *Nat Genet.* 2012;44:916-921.

32. Guo DC, Regalado E, Casteel DE, Santos-Cortez RL, Gong L, Kim JJ, Dyack S, Horne SG, Chang G, Jondeau G, Boileau C, Coselli JS, Li Z, Leal SM, Shendure J, Rieder MJ, Bamshad MJ, Nickerson DA, Kim C, Milewicz DM. Recurrent gain-of-function mutation in PRKG1 causes thoracic aortic aneurysms and acute aortic dissections. *Am J Hum Genet.* 2013;93:398-404.

33. Gordon K, Schulte D, Brice G, Simpson MA, Roukens MG, van IA, Connell F, Kalidas K, Jeffery S, Mortimer PS, Mansour S, Schulte-Merker S, Ostergaard P. Mutation in vascular endothelial growth factor-C, a ligand for vascular endothelial growth factor receptor-3, is associated with autosomal dominant milroy-like primary lymphedema. *Circ Res.* 2013;112:956-960.

34. Wooderchak-Donahue WL, McDonald J, O'Fallon B, Upton PD, Li W, Roman BL, Young S, Plant P, Fulop GT, Langa C, Morrell NW, Botella LM, Bernabeu C, Stevenson DA, Runo JR, Bayrak-Toydemir P. BMP9 mutations cause a vascular-anomaly syndrome with phenotypic overlap with hereditary hemorrhagic telangiectasia. *Am J Hum Genet.* 2013;93:530-537.

35. Boyden LM, Choi M, Choate KA, Nelson-Williams CJ, Farhi A, Toka HR, Tikhonova IR, Bjornson R, Mane SM, Colussi G, Lebel M, Gordon RD, Semmekrot BA, Poujol A, Valimaki MJ, De Ferrari ME, Sanjad SA, Gutkin M, Karet FE, Tucci JR, Stockigt JR, Keppler-Noreuil KM, Porter CC, Anand SK, Whiteford ML, Davis ID, Dewar SB, Bettinelli A, Fadrowski JJ, Belsha CW, Hunley TE, Nelson RD, Trachtman H, Cole TR, Pinsk M, Bockenhauer D, Shenoy M, Vaidyanathan P, Foreman JW, Rasoulpour M, Thameem F, Al-Shahrouri HZ, Radhakrishnan J, Gharavi AG, Goilav B, Lifton RP. Mutations in kelch-like 3 and cullin 3 cause hypertension and electrolyte abnormalities. *Nature.* 2012;482:98-102.

36. Louis-Dit-Picard H, Barc J, Trujillano D, Miserey-Lenkei S, Bouatia-Naji N, Pylypenko O, Beaurain G, Bonnefond A, Sand O, Simian C, Vidal-Petiot E, Soukaseum C, Mandet C, Broux F, Chabre O, Delahousse M, Esnault V, Fiquet B, Houillier P, Bagnis CI, Koenig J, Konrad M, Landais P, Mourani C, Niaudet P, Probst V, Thauvin C, Unwin RJ, Soroka SD, Ehret G, Ossowski S, Caulfield M, Bruneval P, Estivill X, Froguel P, Hadchouel J, Schott JJ, Jeunemaitre X. KLHL3 mutations cause familial hyperkalemic hypertension by impairing ion transport in the distal nephron. *Nat Genet.* 2012;44:456-3.

37. Austin ED, Ma L, LeDuc C, Berman RE, Borczuk A, Phillips JA, III, Palomero T, Sumazin P, Kim HR, Talati MH, West J, Loyd JE, Chung WK. Whole exome sequencing to identify a novel gene (caveolin-1) associated with human pulmonary arterial hypertension. *Circ Cardiovasc Genet.* 2012;5:336-343.

38. Choi M, Scholl UI, Yue P, Bjorklund P, Zhao B, Nelson-Williams C, Ji W, Cho Y, Patel A, Men CJ, Lolis E, Wisgerhof MV, Geller DS, Mane S, Hellman P, Westin G, Akerstrom G, Wang W, Carling T, Lifton RP. K+ channel mutations in adrenal aldosterone-producing adenomas and hereditary hypertension. *Science.* 2011;331:768-772.

39. Scholl UI, Goh G, Stolting G, de Oliveira RC, Choi M, Overton JD, Fonseca AL, Korah R, Starker LF, Kunstman JW, Prasad ML, Hartung EA, Mauras N, Benson MR, Brady T, Shapiro JR, Loring E, Nelson-Williams C, Libutti SK, Mane S, Hellman P, Westin G, Akerstrom G, Bjorklund P, Carling T, Fahlke C, Hidalgo P, Lifton RP. Somatic and germline CACNA1D calcium channel mutations in aldosterone-producing adenomas and primary aldosteronism. *Nat Genet.* 2013;45:1050-1054.

40. Beuschlein F, Boulkroun S, Osswald A, Wieland T, Nielsen HN, Lichtenauer UD, Penton D, Schack VR, Amar L, Fischer E, Walther A, Tauber P, Schwarzmayr T, Diener S, Graf E, Allolio B, Samson-Couterie B, Benecke A, Quinkler M, Fallo F, Plouin PF, Mantero F, Meitinger T, Mulatero P, Jeunemaitre X, Warth R, Vilsen B, Zennaro MC, Strom TM, Reincke M. Somatic mutations in ATP1A1 and ATP2B3 lead to aldosterone-producing adenomas and secondary hypertension. *Nat Genet.* 2013;45:440-442.

41. Azizan EA, Poulsen H, Tuluc P, Zhou J, Clausen MV, Lieb A, Maniero C, Garg S, Bochukova EG, Zhao W, Shaikh LH, Brighton CA, Teo AE, Davenport AP, Dekkers T, Tops B, Kusters B, Ceral J, Yeo GS, Neogi SG, McFarlane I, Rosenfeld N, Marass F, Hadfield J, Margas W, Chaggar K, Solar M, Deinum J, Dolphin AC, Farooqi IS, Striessnig J, Nissen P, Brown MJ. Somatic mutations in ATP1A1 and CACNA1D underlie a common subtype of adrenal hypertension. *Nat Genet.* 2013;45:1055-1060.

42. Harakalova M, van Harssel JJ, Terhal PA, van LS, Duran K, Renkens I, Amor DJ, Wilson LC, Kirk EP, Turner CL, Shears D, Garcia-Minaur S, Lees MM, Ross A, Venselaar H, Vriend G, Takanari H, Rook MB, van der Heyden MA, Asselbergs FW, Breur HM, Swinkels ME, Scurr IJ, Smithson SF, Knoers NV, van der Smagt JJ, Nijman IJ, Kloosterman WP, van Haelst MM, van HG, Cuppen E. Dominant missense mutations in ABCC9 cause Cantú syndrome. *Nat Genet.* 2012;44:793-796.

43. van Bon BW, Gilissen C, Grange DK, Hennekam RC, Kayserili H, Engels H, Reutter H, Ostergaard JR, Morava E, Tsiakas K, Isidor B, Le MM, Eser M, Wieskamp N, de VP, Steehouwer M, Veltman JA, Robertson SP, Brunner HG, de Vries BB, Hoischen A. Cantú syndrome is caused by mutations in ABCC9. *Am J Hum Genet.* 2012;90:1094-1101.

44. Ng BG, Hackmann K, Jones MA, Eroshkin AM, He P, Wiliams R, Bhide S, Cantagrel V, Gleeson JG, Paller AS, Schnur RE, Tinschert S, Zunich J, Hegde MR, Freeze HH. Mutations in the glycosylphosphatidylinositol gene PIGL cause CHIME syndrome. *Am J Hum Genet.* 2012;90:685-688.

45. Carapito R, Paul N, Untrau M, Ott L, Corradini N, Poignant S, Geffroy L, Caldagues E, Heymann MF, Cassagnau E, Isidor B, Bahram S. A new mutation in the C-SH2 domain of PTPN11 causes Noonan syndrome with multiple giant cell lesions. *J Hum Genet.* 2014;59:57-59.

46. Li G, Yu J, Wang K, Wang B, Wang M, Zhang S, Qin S, Yu Z. Exome sequencing identified new mutations in a Marfan syndrome family. *Diagn Pathol.* 2014;9:25.

47. Tegtmeyer LC, Rust S, van SM, Ng BG, Losfeld ME, Timal S, Raymond K, He P, Ichikawa M, Veltman J, Huijben K, Shin YS, Sharma V, Adamowicz M, Lammens M, Reunert J, Witten A, Schrapers E, Matthijs G, Jaeken J, Rymen D, Stojkovic T, Laforet P, Petit F, Aumaitre O, Czarnowska E, Piraud M, Podskarbi T, Stanley CA, Matalon R, Burda P, Seyyedi S, Debus V, Socha P, Sykut-Cegielska J, van SF, De ML, Vajro P, DeClue T, Ficicioglu C, Wada Y, Wevers RA, Vanderschaeghe D, Callewaert N, Fingerhut R, van SE, Freeze HH, Morava E, Lefeber DJ, Marquardt T. Multiple phenotypes in phosphoglucomutase 1 deficiency. *N Engl J Med.* 2014;370:533-542.