

**Total:****266****GLA Mutations associated with Classic/early-onset Fabry disease ( total number: 266)**

Abbreviations	Literature Phenotype	Reference 1
M1T	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
M1R	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
M1I	Classic	<a href="#">Blanch (1996) Hum Mutat 8, 38</a>
M1K	Classic	<a href="#">Dobrovolny (2008) Am Soc Hum Genet Meet Abs . 150</a>
C12_L14del	Classic	<a href="#">Chien (2011) Mol Genet Metab 102, 57</a>
L14P	Classic	<a href="#">Tse (2003) Nephrol Dial Transplant 18, 182</a>
L16P	Classic	<a href="#">Garzuly (2005) Brain 128, 2078</a>
A20_W24del	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
A31V	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
N34K	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
N34S	Classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
P40H	Classic	<a href="#">Meng (2010) Zhonghua yi Xue Za Zhi 90,551</a>
P40L	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
P40S	classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
T41I	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
M42T	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
M42V	Classic	<a href="#">Davies (1996) Eur J Hum Genet 4, 219</a>
G43D	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
G43V	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
G43R	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
W44C	Classic	<a href="#">Wang (2005) Zhonghua Yi Xue Yi Chuan Xue Za Zhi 22, 489</a>
L45R/H46S	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
H46R	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
H46L	Classic	<a href="#">Tsuboi (2012) Genet Med 14, 779</a>
H46Y	Classic	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
E48K	Classic	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
R49G	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
R49L	Classic	<a href="#">Davies (1994) Hum Mol Genet 3, 667</a>
R49P	Classic	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
R49S	Classic	<a href="#">Davies (1996) Eur J Hum Genet 4, 219</a>
F50C	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>

M51K	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
C52G	Classic	<a href="#">Cruz (2011) Acta Reumatol Port 36, 399</a>
C52R	Classic	<a href="#">Blanch (1996) Hum Mutat 8, 38</a>
C52S	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
C52Y	Classic	<a href="#">Filoni (2010) Biochim Biophys Acta 1802, 247</a>
C56F	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
C56G	Classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
C56Y	Classic	<a href="#">Davies (1996) Eur J Hum Genet 4, 219</a>
E59K	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
C63S	Classic	<a href="#">Brito (2014) Rev Port Cardiol 33, 247.e1</a>
L68F	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
M72I	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
W81S	Classic	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
Y86C	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
Y86D	Classic	<a href="#">Dobrovolny ASHG Symp., Abstract 633/W, p 150 Philadelphia, 10/2008</a>
E87_D92del	Classic	<a href="#">Davies (1996) Eur J Hum Genet 4, 219</a>
L89P	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
L89R	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
D92N	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
D92H	Classic	<a href="#">Davies (1996) Eur J Hum Genet 4, 219</a>
D92Y	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
D92V	classic	<a href="#">Meng (2010) Zhonghua yi Xue Za Zhi 90,551</a>
D93G	Classic	<a href="#">Davies (1996) Eur J Hum Genet 4, 219</a>
D93N	Classic	<a href="#">Shabbeer (2005) Hum Mutat 25, 299</a>
D93V	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
D93E	Classic	<a href="#">Burlina (2008) J Neurol 255, 738</a>
C94Y	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
C94S	Classic	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
W95L	Classic	<a href="#">Dobrovolny ASHG Symp., Abstract 633/W, p 150 Philadelphia, 10/2008</a>
W95S	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
A97P	classic	<a href="#">Kimura (2002) Br J Dermatol 147, 545</a>
R100K	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
R100T	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>

R112C	Classic	<a href="#">Ishii (1992) Hum Genet 89, 29</a>
F113_R118del	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
F113S	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
L120P/A121T	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
L120_N122delinsH	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
A121P	Classic	<a href="#">Kotanko (2004) J Am Soc Nephrol 15, 1323</a>
S126G	Classic	<a href="#">Branton (2002) Medicine (Baltimore) 81, 122</a>
G128E	Classic	<a href="#">Blanch (1996) Hum Mutat 8, 38</a>
L131P	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
G132R	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
Y134S	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
A135V	Classic	<a href="#">Shabbeer (2005) Hum Mutat 25, 299</a>
D136H	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
G138E	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
G138R	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
T141I	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
C142R	Classic	<a href="#">Topaloglu (1999) Mol Med 5, 806</a>
C142Y	Classic	<a href="#">Okumiya (1995) Hum Genet 95, 557</a>
A143P	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
G144V	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
G144D	Classic	<a href="#">Li (2014) J Dermatol 41, 456</a>
S148N	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
S148R	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
D155H	Classic	<a href="#">Dobrovolny (2005) J Mol Med 83, 647</a>
A156V	Classic	<a href="#">Okumiya (1995) Hum Genet 95, 557</a>
A156T	Classic	<a href="#">Eng (1994) Hum Mutat 3, 103</a>
W162R	Classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
G163V	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
D165V	Classic	<a href="#">Davies (1994) Hum Mol Genet 3, 667</a>
L166G	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
L166V	Classic	<a href="#">Okumiya (1995) Hum Genet 95, 557</a>
L167P	Classic	<a href="#">Morrone (2003) J Med Genet 40, e103</a>
K168R	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>

F169S	Classic	<a href="#">Cooper (2000) Hum Genet 107 535</a>
D170V	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
D170H	Classic	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
G171D	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
G171R	Classic	<a href="#">Dobrovoly (2005) J Mol Med 83, 647</a>
G171C	Classic	<a href="#">Nagasaki (2008) J Dermat Sci</a>
C172Y	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
C172F	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
C172R	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
C172G	Classic	<a href="#">Yasuda (2003) Hum Mutat 22, 486</a>
C174R	Classic	<a href="#">Meng (2010) Zhonghua Yi Xue Za Zhi 90, 551</a>
G183D	Classic	<a href="#">Topaloglu (1999) Mol Med 5, 806</a>
G183V	Classic	<a href="#">Dobrovoly ASHG Symp., Abstract 633/W, p 150 Philadelphia, 10/2008</a>
M187T	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
M187V	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
L191Q	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
L191P	Classic	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
V199M	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
S201F	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
S201Y	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
C202Y	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
C202W	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
P205_Y207DEL	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
P205R	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
P205T	Classic	<a href="#">Blanch (1996) Hum Mutat 8, 38</a>
Y207S	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
Y207H	Classic	<a href="#">Dobrovoly ASHG Symp., Abstract 633/W, p 150 Philadelphia, 10/2008</a>
Y216D	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
Y216C	Classic	<a href="#">Filoni (2010) Biochim Biophys Acta 1802, 247</a>
I219N	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
C223R	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
C223Y	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
N224S	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>

N224D	Classic	<a href="#">Guffon (1998) Hum Mutat S1, S288</a>
W226R	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
R227Q	Classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
A230T	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
D231N	Classic	<a href="#">Redonnet-Vernhet (1996) J Med Genet 33, 682</a>
D231G	Classic	<a href="#">Park (2009) Exp Mol Med 41, 1</a>
D231V	Classic	<a href="#">Shimotori (2008) Hum Mutat 29, 331</a>
D234E	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
D234Y	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
S235C	Classic	<a href="#">Topaloglu (1999) Mol Med 5, 806</a>
S235F	Classic	<a href="#">Shimotori (2008) Hum Mutat 29, 331</a>
W236R	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
W236L	Classic	<a href="#">Topaloglu (1999) Mol Med 5, 806</a>
W236C	Classic	<a href="#">Davies (1996) Eur J Hum Genet 4, 219</a>
I239T	Classic	<a href="#">Kotanko (2004) J Am Soc Nephrol 15, 1323</a>
I242N	Classic	<a href="#">Takata (1997) Brain Dev 19, 111</a>
L243F	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
L243W	Classic	<a href="#">Rozenfeld (2006) Clin Genet 69, 344;</a>
D244N	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
D244H	Classic	<a href="#">Topaloglu (1999) Mol Med 5, 806</a>
S247P	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
S247_F248InsWTS	Classic	<a href="#">Guffon (1998) Hum Mutat S1, S288</a>
G258V	Classic	<a href="#">Shimotori (2008) Hum Mutat 29, 331</a>
P259R	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
G260A	Classic	<a href="#">Okumiya (1995) Biochem Biophys Res Commun 214, 1219</a>
G261D	Classic	<a href="#">Takata (1997) Brain Dev 19, 111</a>
N263S	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
D264V	Classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
D264Y	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
D266N	classic	<a href="#">Lee (2000) Clin Genet 58, 228</a>
D266H	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
D266V	Classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
D266E	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>

D266A	Classic	<a href="#">Tian (2013) Zhonghua Yi Xue Yi Chuan Xue Za Zhi 30, 185</a>
M267R	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
V269A	Classic	<a href="#">Davies (1993) Hum Mol Genet 2, 1051</a>
V269M	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
I270T	classic	<a href="#">Ries (2005) Pediatrics 115, 344</a>
G271C	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
G271S	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
G271V	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
N272K	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
N272S	Classic	<a href="#">Verovnik (2004) Eur J Hum Genet 12, 678</a>
L275F	Classic	<a href="#">Dobrovolny ASHG Symp., Abstract 633/W, p 150 Philadelphia, 10/2008</a>
S276G	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
Q279R	Classic	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
Q279H	Classic	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
Q279K	Classic	<a href="#">Dominissini (2004) Hum Heredity 57, 138</a>
Q280K	Classic	<a href="#">Dobrovolny (2005) J Mol Med 83, 647</a>
V281_T282delinsA	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
T282A	Classic	<a href="#">Shimotori (2008) Hum Mutat 29, 331</a>
T282N	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
Q283P	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
M284T	Classic	<a href="#">Blanch (1996) Hum Mutat 8, 38</a>
A285P	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
A285D	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
W287G	Classic	<a href="#">Davies (1996) Eur J Hum Genet 4, 219</a>
W287C	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
A288D	Classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
A288P	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
I289F	Classic	<a href="#">Topaloglu (1999) Mol Med 5, 806</a>
I289S	Classic	<a href="#">Erdos et al (2008) Mol Genet Metab 95, 224</a>
M290I	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
A292P	Classic	<a href="#">Wang (2005) Zhonghua Yi Xue Yi Chuan Xue Za Zhi 22, 489</a>
P293L	Classic	<a href="#">Sirrs (2010) Mol Genet Metab 99, 367</a>
P293A	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>

P293T	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
S297C	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
S297F	Classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
N298S	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
N298K	Classic	<a href="#">Blanch (1996) Hum Mutat 8, 38</a>
N298H	Classic	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
R301G	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
R301P	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>
I303N	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
K308N	Classic	<a href="#">Shimotori (2008) Hum Mutat 29, 331</a>
L310F	Classic	<a href="#">Calado (2004) Hum Genet 115 347</a>
L310R	Classic	<a href="#">Meng (2010) Zhonghua yi Xue Za Zhi 90,551</a>
L311R	Classic	<a href="#">Suzuki (2011) Clin Exp Nephrol 15, 916</a>
Q312H	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
V316E	Classic	<a href="#">Davies (1994) Hum Mol Genet 3, 667</a>
I317T	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
I319T	Classic	<a href="#">Sirrs (2010) Mol Genet Metab 99, 367</a>
N320I	classic	<a href="#">Illsinger (2003) Eur J Pediatr 162, 522</a>
N320K	classic	<a href="#">Okumiya (1995) Biochem Biophys Res Commun 214, 1219</a>
N320Y	Classic	<a href="#">Ashton-Prolla (2000) J Investig Med 48, 227</a>
Q321E	Classic	<a href="#">Topaloglu (1999) Mol Med 5, 806</a>
Q321R	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
D322E	Classic	<a href="#">Lee (2010) J Hum Genet 55, 512</a>
G325D	classic	<a href="#">Ries (2005) Pediatrics 115, 344</a>
Q327K	Classic	<a href="#">Davies (1993) Hum Mol Genet 2, 1051</a>
G328R	Classic	<a href="#">Ishii (1992) Hum Genet 89, 29</a>
G328A	Classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
G328V	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
E338K	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
E341D	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
E341K	Classic	<a href="#">Beyer (1999) Clin Chim Acta 280, 81</a>
R342Q	Classic	<a href="#">Ploos van Amstel (1994) Hum Mol Genet 3, 503;</a>
A348P	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>

A350P	Classic	<a href="#">Schafer (2005) Hum Mutat 25, 412;</a>
A352D	classic	<a href="#">Morrone (2003) J Med Genet 40, e103</a>
N355K	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
R356W	Classic	<a href="#">Bernstein (1989) J Clin Invest 83, 1390</a>
Q357X	Classic	<a href="#">Cooper (2000) Hum Genet 107 535</a>
E358G	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
E358del	Classic	<a href="#">Blanch (1996) Hum Mutat 8, 38</a>
E358K	Classic	<a href="#">Miyazaki (1998) Hum Mutat S1, S139;</a>
E358A	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
G360S	classic	<a href="#">Dobrovolny (2005) J Mol Med 83, 647</a>
G360C	Classic	<a href="#">Chien (2011) Mol Genet Metab 102, 57</a>
G361A	Classic	<a href="#">Sirrs (2010) Mol Genet Metab 99, 367</a>
G361R	Classic	<a href="#">Davies (1993) Hum Mol Genet 2, 1051</a>
P362L	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
R363C	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
L372Q	Classic	<a href="#">Utsumi (2009) J Neurol Sci 283, 83</a>
L372R	Classic	<a href="#">Dobrovolny (2008) Am Soc Hum Genet Meet Abs . 150</a>
G373D	classic	<a href="#">Germain (2001) Hum Mutat 17, 353;</a>
G373S	Classic	<a href="#">Okumiya (1995) Biochem Biophys Res Commun 214, 1219</a>
A377D	Classic	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
C378Y	Classic	<a href="#">Topaloglu (1999) Mol Med 5, 806</a>
C382Y	Classic	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
F383del	Classic	<a href="#">Carilou (1996) Hum Genet 97, 468</a>
I384N	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
T385P	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
Q386P	Classic	<a href="#">Shabbeer (2006) Hum Genomics 2, 297</a>
E398K	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
W399X	Classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
L403S	Classic	<a href="#">Shimotori (2008) Hum Mutat 29, 331</a>
I407K	classic	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
P409A	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>
P409S	Classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
T410K	Classic	<a href="#">Ashley (2001) J Hum Genet 46, 192</a>



T410P	Classic	<a href="#">Shimotori (2008) Hum Mutat 29, 331</a>
G411D	Classic	<a href="#">Guffon (2003) J Med Genet 40, e38</a>
L414S	Classic	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
L415P	Classic	<a href="#">Rozenfeld (2006) Clin Genet 69, 344</a>

**Total****54****GLA Mutations consistently associated with Non-classic (Variant, later-onset) Fabry disease (total number: 54)**

Abbreviations	Literature Phenotype	Reference 1
L19P	Non-classic	<a href="#">Teraguchi (2004) Jpn Heart J 45, 685</a>
A20P	Non-classic	<a href="#">Nakao (1995) N Engl J Med 333, 288</a>
W24R	Non-classic	<a href="#">Al-Thihli (2012) Gene 497, 320</a>
G35R	Non-classic	<a href="#">Davies (1994) Hum Mol Genet 3, 667</a>
L36S	Non-classic	<a href="#">Erdos et al (2008) Mol Genet Metab 95, 224</a>
P40A	Non-classic	<a href="#">Koskenvuo (2009) Clin Physiol Funct Imaging 29, 177</a>
M42L	Non-classic	<a href="#">Rosenthal (2004) Am J Kidney Dis 44, e85</a>
W47G	Non-classic	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
M51I	Non-classic	<a href="#">Spada (2006) Am J Hum Genet 79, 31</a>
E66G	Non-classic	<a href="#">Spada (2006) Am J Hum Genet 79, 31</a>
M72V	Non-classic	<a href="#">Okumiya (1998) Hum Mutat S1, S213</a>
A73V	Non-classic	<a href="#">Spada (2006) Am J Hum Genet 79, 31</a>
M76T	Non-classic	<a href="#">Shimotori (2008) Hum Mutat 29, 331</a>
I91T	Non-classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
D92G	Non-classic	<a href="#">Huerva (2008) Cornea 27, 970</a>
A97V	Non-classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
G104V	Non-classic	<a href="#">Lin (2009) Circ Cardiovasc Genet 2, 450</a>
L106R	Non-classic	<a href="#">Wattanasirichaigoon (2006) J Med Assoc Thai 89, 1528</a>
R112H	Non-classic	<a href="#">Eng (1994) Hum Mol Genet 3, 1795</a>
R118C	Non-classic	<a href="#">Spada (2006) Am J Hum Genet 79, 31</a>
N139T	Non-classic	<a href="#">Havndrup (2006) Circulation (abstract #917)</a>
N139S	Non-classic	<a href="#">Havndrup (2010) Eur J Heart Fail 12, 535</a>
A143T	Non-classic	<a href="#">Spada (2006) Am J Hum Genet 79, 31</a>
D165H	Non-classic	<a href="#">Erdos et al (2008) Mol Genet Metab 95, 224</a>
G183A	Non-classic	<a href="#">Filoni (2010) Biochim Biophys Acta 1802, 247</a>
Y184N	Non-classic	<a href="#">Tuttolomondo (2012) Clin Biochem epub, epub</a>
G195V	Non-classic	<a href="#">Nakagawa (2011) Int Heart J 52, 308</a>
P210L	Non-classic	<a href="#">Saito (2013) PLoS One 8, e84267</a>
P210S	Non-classic	<a href="#">Saito (2013) PLoS One 8, e84267</a>
N215S	Non-classic	<a href="#">Dobrovlny (2005) J Mol Med 83, 647; Eng (1993) Am J Hum Genet 53, 1186; Davies (1996) Eur J Hum Genet 4, 219</a>
R220P	Non-classic	<a href="#">Fukutomi (2013) J Cardiol epub, epub</a>

F229L	Non-classic	<a href="#">Kawano (2007) Am J Cardiol 99, 261</a>
S238N	Non-classic	<a href="#">Montserrat (2007) J Am Coll Cardiol 50, 2399</a>
S247C	Non-classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
V254del	Non-classic	<a href="#">Ichinose (2005) Clin Exp Nephrol 9, 228</a>
P259L	Non-classic	<a href="#">Topaloglu (1999) Mol Med 5, 806</a>
P265R	Non-classic	<a href="#">Germain (1996) Hum Genet 98, 719</a>
D266Y	Non-classic	<a href="#">Erdos et al (2008) Mol Genet Metab 95, 224</a>
F273L	Non-classic	<a href="#">Zhang (2007) J Nephrol 20(6):716-26</a>
Q279E	Non-classic	<a href="#">Ishii (1992) Hum Genet 89, 29</a>
A291T	Non-classic	<a href="#">Zhang (2007) J Nephrol 20, 716</a>
M296I	Non-classic	<a href="#">Nakao (1995) N Engl J Med 333, 288</a>
M296L	Non-classic	<a href="#">Lin (2009) Circ Cardiovasc Genet 2, 450</a>
M296V	Non-classic	<a href="#">von Scheidt (1991) N Engl J Med 324, 395</a>
Q312R	Non-classic	<a href="#">Shimotori (2008) Hum Mutat 29, 331</a>
W340R	Non-classic	<a href="#">Davies (1996) Eur J Hum Genet 4, 219</a>
P343L	Non-classic	<a href="#">Mills (2005) J Inherit Metab Dis 28, 35</a>
R356Q	Non-classic	<a href="#">Chien (2011) Mol Genet Metab 102, 57</a>
C382W	Non-classic	<a href="#">Galanos (2002) Intern Med J 32, 575</a>
K391T	Non-classic	<a href="#">Lin (2009) Circ Cardiovasc Genet 2, 450</a>
G395A	Non-classic	<a href="#">Bono (2011) BMC Res Notes 4, 253</a>
T400_S401dup	Non-classic	<a href="#">Kroepfl (2002) J Inherit Metab Dis 25, 695</a>
R404del	Non-classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
T410A	Non-classic	<a href="#">Yang (2003) Clin Genet 63, 205</a>

Total:

5

**Mutations associated with different with Fabry disease phenotypes (total number 5)**

Abbreviations	Literature Phenotype	Reference 1
E66Q	Classic	<a href="#">Ishii (1992) Hum Genet 89, 29</a>
	Non-classic	<a href="#">Fujii (2009) Am J Nephrol 30, 527</a>
	polymorphism	<a href="#">Lee (2010) J Hum Genet 55, 512</a>
F113L	Non-classic	<a href="#">Eng (1997) Mol Med 3, 174</a>
	Classic	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
R301Q	Non-classic	<a href="#">Sakuraba (1990) Am J Hum Genet 47, 784</a>
	Non-classic	<a href="#">Ishii (1992) Hum Genet 89, 29</a>
	Non-classic	<a href="#">Germain (2002) Mol Med 8, 306</a>
	classic	<a href="#">Germain (1999) Biochem Biophys Res Commun 257, 708</a>
D313Y	Classic	<a href="#">Eng (1993) Am J Hum Genet 53, 1186</a>
	pseudodeficiency	<a href="#">Froissart Mol Genet Metab 2003 80, 307</a>
R363H	Non-classic	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
	Classic	<a href="#">Shabbeer (2002) Mol Genet Metab 76, 23</a>

**Total:****275****Mutations without Fabry phenotype reported (total number: 275)**

Abbreviations	Literature Phenotype	Reference 1
M1L	unknown	Amicus
M1V	unknown	<a href="#">Kwan et al. ASHG Symp. San Diego 10/2007</a>
L3P	unknown	Amicus
A13P	unknown	<a href="#">Booth (2008) MPS Abstract 17</a>
A13T	unknown	Amicus
A15G	unknown	<a href="#">Koskenvuo (2009) Clin Physiol Funct Imaging 29, 177</a>
A15P	unknown	<a href="#">Turaça (2012) J Hum Genet 57, 347</a>
A15T	unknown	<a href="#">Ebrahim (2012) J Inherit Metab Dis 35, 325</a>
L16H	unknown	<a href="#">Kobayashi (2007) J Inherit Metab Dis 30(Suppl 1), 105</a>
F18S	unknown	<a href="#">Vedder (2008) Mol Genet Metab</a>
L19Q	unknown	Amicus
A20D	unknown	Amicus
A20V	unknown	Amicus
L21P	unknown	Amicus
L21R	unknown	Amicus
L21F	unknown	<a href="#">Heo et al. ASHG Abstract 2009</a>
W24G	unknown	<a href="#">Kleinert (2009) Transpl Int 22, 287</a>
W24C	unknown	Amicus
L32P	unknown	<a href="#">Madsen (1995) Hum Mutat 5, 277</a>
D33Y	unknown	<a href="#">Kobayashi (2007) J Inherit Metab Dis 30(Suppl 1), 105</a>
D33G	unknown	Amicus
L36F	unknown	<a href="#">Pereira (2007) Braz J Med Biol Res 40, 1599</a>
L36W	unknown	Amicus
A37T	unknown	<a href="#">Sadick (2007) Heart Lung Circ 16, 200</a>
A37V	unknown	<a href="#">Kobayashi (2007) J Inherit Metab Dis 30(Suppl 1), 105</a>
P40R	unknown	Amicus
M42R	unknown	<a href="#">Kwan et al. ASHG Symp. San Diego 10/2007</a>
M42K	unknown	Amicus
G43S	unknown	<a href="#">Kwan et al. ASHG Symp. San Diego 10/2007</a>

L45P	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
L45R	unknown	mutation was made because of the reported L45R/H46S
H46P	unknown	<a href="#">Hwu (2009) Hum Mutat 30, 1397</a>
W47L	unknown	<a href="#">Cooper (2000) Hum Genet 107 535</a>
W47C	unknown	Amicus
W47R	unknown	Amicus
E48D	unknown	<a href="#">Kwan et al. ASHG Symp. San Diego 10/2007</a>
E48Q	unknown	Amicus
R49C	unknown	<a href="#">Shin (2008) Pharmacogenet Genomics 18,773</a>
R49_F50InsP	unknown	Amicus
C52W	unknown	Amicus
N53D	unknown	Amicus
N53L	unknown	Amicus
L54F	unknown	Amicus
L54P	unknown	Shire FOS registry
D55V	unknown	Amicus
C56S	unknown	Amicus
Q57L	unknown	Amicus
D55V/Q57L	unknown	Amicus
P60S	unknown	<a href="#">Johnson (2013) Ann Lab Med 33, 274</a>
P60L	unknown	<a href="#">Vedder (2007) J Inherit Metab Dis 30, 68</a>
P60T	unknown	Amicus
C63R	unknown	Amicus
C63Y	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
E66K	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
M72R	unknown	<a href="#">Slee (2000) Ned Tijdschr Geneesk 144, 2412</a>
A73E	unknown	<a href="#">Nishino (2012) Ren Fail 34, 566</a>
M76R	unknown	Amicus
W81C	unknown	<a href="#">Kwan et al. ASHG Symp. San Diego 10/2007</a>
W81R	unknown	<a href="#">Burlina (2008) J Neurol 255, 738</a>
D83N	unknown	Amicus

G85D	unknown	<a href="#">Madsen (1995) Hum Mutat 5, 277</a>
G85M	unknown	Shire FOS registry
G85S	unknown	Amicus
Y86H	unknown	Amicus
Y88D	unknown	<a href="#">Cooper (2000) Hum Genet 107 535</a>
L89F	unknown	Amicus
C90R	unknown	<a href="#">Booth (2008) MPS Abstract 17</a>
C90Y	unknown	Amicus
D93Y	unknown	<a href="#">Ebrahim (2012) J Inherit Metab Dis 35, 325</a>
M96I	unknown	Amicus
S102L	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
E103Q	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
Q107L	unknown	<a href="#">Das (2008) Abstract SSIEM</a>
A108T	unknown	<a href="#">Tai (2012) Clin Chim Acta 413, 422</a>
D109G	unknown	<a href="#">Herrera (2014) Clin Nephrol 81, 112</a>
R112S	unknown	<a href="#">Shabbeer (2005) Hum Mutat 25, 299</a>
R112G	unknown	Amicus
I117S	unknown	<a href="#">Booth (2008) MPS Abstract 17</a>
L120P	unknown	Amicus
L120S	unknown	<a href="#">Chinese PhD thesis abstract found on the internet</a>
L120V	unknown	<a href="#">Chien (2009) Hum Genet 125 336</a>
A121T	unknown	Amicus
Y123C	unknown	Amicus
V124D	unknown	<a href="#">Kwan et al, ASHG Symp. San Diego 10/2007</a>
H125P	unknown	<a href="#">Auray-Blais (2008) Mol Genet Metab 93, 331</a>
H125L/G128E	unknown	Amicus
H125L	unknown	Amicus
L129P	unknown	<a href="#">Whybra (2001) J Inherit Metab Dis 24, 715</a>
K130R	unknown	Amicus
G132E	unknown	<a href="#">Kwan et al, ASHG Symp. San Diego 10/2007</a>
G132A	unknown	Amicus

Y134H	unknown	<a href="#">Finoli (2007) Acta Paediatrica (abstract #5)</a>
Y134D	unknown	Amicus
D136Y	unknown	<a href="#">Vedder (2008) Mol Genet Metab</a>
D136E	unknown	Amicus
K140T	unknown	WORLD 2012 presentation, found in newborn screening
T141N	unknown	Amicus
C142W	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
P146S	unknown	<a href="#">Ploos van Amstel (1994) Hum Mol Genet 3, 503</a>
G147R	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
G147E	unknown	Shire FOS registry
Y152C	unknown	<a href="#">Kobayashi (2007) J Inherit Metab Dis 30(Suppl 1), 105</a>
Y152_D153InsD	unknown	Amicus
D153del	unknown	<a href="#">Shabbeer (2005) Hum Mutat 25, 299</a>
A156D	unknown	Amicus
W162C	unknown	<a href="#">Germain (1996) Hum Genet 98, 719</a>
W162G	unknown	Shire FOS registry
W162L	unknown	Amicus
V164G	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
D165Y	unknown	Shire FOS registry
D165G	unknown	Amicus
L167Q	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
K168N	unknown	<a href="#">Kwan et al, ASHG Symp. San Diego 10/2007</a>
D170N	unknown	Amicus
D170G	unknown	Amicus
C172W	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
C172S	unknown	<a href="#">Booth (2008) MPS Abstract 17</a>
C174G	unknown	<a href="#">Abstract EJHG 2008</a>
D175E	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
Y184C	unknown	Shire FOS registry
K185E	unknown	Amicus
H186P	unknown	<a href="#">Valentina et al 2011 ESGH poster</a>



M187I	unknown	Amicus
M187R	unknown	Amicus
M187_S188DUP	unknown	Amicus
T194I	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
I198T	unknown	Amicus
V199G	unknown	Amicus
Y200C	unknown	Amicus
C202R	unknown	Amicus
C202R/N215S	unknown	Amicus
E203K	unknown	Amicus
E203V	unknown	Amicus
E203D	unknown	Amicus
W204C	unknown	Amicus
P205L	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
P205S	unknown	Shire FOS registry
L206P	unknown	<a href="#">Chinese PhD thesis abstract found on the internet</a>
Y207C	unknown	<a href="#">Kwan et al, ASHG Symp. San Diego 10/2007</a>
K213M	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
P214S	unknown	Amicus
P214L	unknown	Amicus
N215D	unknown	Shire FOS registry
N215S/D313Y	unknown	Amicus
N215S/G271S	unknown	Amicus
N215S/G271S/D313Y	unknown	Amicus
I219T	unknown	<a href="#">Chien (2011) Mol Genet Metab 102, 57</a>
R220Q	unknown	<a href="#">Scott et al 2012 ASHG poster</a>
C223G	unknown	<a href="#">Germain (1999) Biochem Biophys Res Commun 257, 708</a>
H225D	unknown	Shire FOS registry
H225R	unknown	<a href="#">Politei (2005) Rev Neurol 41, 506</a>
W226C	unknown	<a href="#">Rodriguez-Mari (2003) Hum Mutat 22, 258</a>
R227P	unknown	Amicus

N228S	unknown	Amicus
I232T	unknown	<a href="#">Tai (2012) Clin Chim Acta 413, 422</a>
S235Y	unknown	Amicus
S238R	unknown	<a href="#">SSIEM 2010</a>
I239del	unknown	<a href="#">Valbuena (2008) Virchows Arch 453, 329</a>
I242F	unknown	<a href="#">Kwan et al, ASHG Symp. San Diego 10/2007</a>
W245G	unknown	Amicus
N249K	unknown	<a href="#">Duro (2014) Gene 535, 365</a>
Q250P	unknown	<a href="#">Kobayashi (2007) J Inherit Metab Dis 30(Suppl 1), 105</a>
R252T	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
I253T	unknown	<a href="#">Finoli (2007) Acta Paediatrica (abstract #5)</a>
I253S	unknown	Amicus
A257P	unknown	<a href="#">Cooper (2000) Hum Genet 107 535</a>
A257D	unknown	Amicus
A257G	unknown	Amicus
G258R	unknown	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
G260E	unknown	<a href="#">Mignani (2008) Nephrol Dial Transplant 23, 1628</a>
G261V	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
W262C	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
W262L	unknown	<a href="#">Oneli-Mungan, SSIEM 2010</a>
D264A	unknown	Amicus
D264Y/V269M	unknown	Amicus
P265L	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
P265S	unknown	Amicus
M267T	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
L268S	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
V269E	unknown	Amicus
G271D	unknown	Amicus
G271S/D313Y	unknown	Amicus
G274V	unknown	Amicus
G274S	unknown	<a href="#">Mignani (2008) Nephrol Dial Transplant 23, 1628</a>

S276N	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
W277C	unknown	Amicus
W277G	unknown	Amicus
Q280H	unknown	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
T282I	unknown	Amicus
Q283R	unknown	Amicus
M284V	unknown	Amicus
M290L	unknown	<a href="#">Ferri (2012) Clin Genet 81, 224</a>
A292T	unknown	<a href="#">Spanu (2007) Hum Genet 121 295</a>
A292V	unknown	<a href="#">Turaça (2012) J Hum Genet 57, 347</a>
P293S	unknown	<a href="#">Cooper (2000) Hum Genet 107 535</a>
P293H	unknown	Shire FOS registry
L294S	unknown	Amicus
F295C	unknown	<a href="#">Kwan et al, ASHG Symp. San Diego 10/2007</a>
M296T	unknown	Amicus
D299G	unknown	<a href="#">Kwan et al, ASHG Symp. San Diego 10/2007</a>
D299E	unknown	<a href="#">Vedder (2007) J Inherit Metab Dis 30, 68</a>
L300F	unknown	<a href="#">Shabbeer (2005) Hum Mutat 25, 299</a>
L300H	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
L300P	unknown	<a href="#">Shin (2008) Pharmacogenet Genomics 18, 773</a>
R301L	unknown	Amicus
S304N	unknown	<a href="#">Scott (2013) J Pediatr 163, 498</a>
S304T	unknown	<a href="#">Lee (2014) Mol Genet Metab 111, 507</a>
A307T	unknown	<a href="#">Abstracts of the 11<sup>th</sup> ICIEM (2009) Mol Genet Metab 98, 1</a>
A309P	unknown	Shire FOS registry
L311P	unknown	Amicus
L311F	unknown	<a href="#">Laaksonen (2008) Clin Neurophysiol 119, 1365</a>
L311V	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
D313G	unknown	<a href="#">Booth (2008) MPS Abstract 17</a>
V316G	unknown	Amicus
V316I	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>

I317N	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
I317S	unknown	<a href="#">Turaça (2012) J Hum Genet 57, 347</a>
I319F	unknown	<a href="#">Brennan (2014) J Inherit Metab Dis 37, 103</a>
Q321L	unknown	Shire FOS registry
Q321H	unknown	Amicus
Q321H/D322N	unknown	Amicus
D322N	unknown	Amicus
G325R	unknown	Amicus
G325S	unknown	Shire FOS registry
Q327E	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
G328E	unknown	<a href="#">Kwan et al, ASHG Symp. San Diego 10/2007</a>
G328W	unknown	Amicus
Q333R	unknown	Amicus
G334E	unknown	<a href="#">Scott et al 2012 ASHG poster</a>
V339E	unknown	<a href="#">Kobayashi (2007) J Inherit Metab Dis 30(Suppl 1), 105</a>
V339G	unknown	Amicus
R342L	unknown	<a href="#">Vedder (2008) Mol Genet Metab</a>
R342P	unknown	Amicus
L344P	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
S345P	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
W349R	unknown	<a href="#">Chinese PhD thesis abstract found on the internet</a>
W349S	unknown	Amicus
A352P	unknown	<a href="#">Abstracts of the 11<sup>th</sup> ICIEM (2009) Mol Genet Metab 98, 1</a>
A352V	unknown	Amicus
A352_M353InsT	unknown	Amicus
I354K	unknown	<a href="#">Azibi (2002) ASHG Annual Meeting Abstracts 2170</a>
R356G	unknown	<a href="#">Chinese PhD thesis abstract found on the internet</a>
E358D	unknown	Amicus
I359T	unknown	<a href="#">Kobayashi (2007) J Inherit Metab Dis 30(Suppl 1), 105</a>
G360D	unknown	<a href="#">Mignani (2008) Nephrol Dial Transplant 23, 1628</a>
G361E	unknown	Amicus

P362T	unknown	<a href="#">Liao (2014) Clin Chim Acta 431C, 80</a>
R363P	unknown	Shire FOS registry
A368T	unknown	Amicus
L372P	unknown	<a href="#">Booth (2008) MPS Abstract 17</a>
G373R	unknown	<a href="#">Kwan et al. ASHG Symp. San Diego 10/2007</a>
G375_V376del	unknown	Amicus
A377_P380dup	unknown	<a href="#">Turaça (2012) J Hum Genet 57, 347</a>
C378R	unknown	Shin (2001) ASHG Annual Meeting Abstracts 1799
T385A	unknown	Amicus
P389R	unknown	<a href="#">Kwan et al. ASHG Symp. San Diego 10/2007</a>
P389A	unknown	Amicus
P389L	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
V390frX8	unknown	<a href="#">Schafer (2005) Hum Mutat 25, 412</a>
G395E	unknown	<a href="#">Johnson (2013) Ann Lab Med 33, 274</a>
L394_G395InsVal	unknown	Amicus
F396Y	unknown	<a href="#">Garman (2004) J Mol Biol 19, 319</a>
S405R	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
H406R	unknown	Amicus
I407V	unknown	Amicus
I407R	unknown	<a href="#">Booth (2008) MPS Abstract 17</a>
P409T	unknown	<a href="#">Blaydon (2001) Hum Mutat 18, 459</a>
T410I	unknown	<a href="#">Booth (2008) MPS Abstract 17</a>
T412N	unknown	Amicus
L415F	unknown	<a href="#">Lukas (2013) PLoS Genet 9, e1003632</a>
Q416X	unknown	Amicus
L417R	unknown	Amicus
E418G	unknown	<a href="#">Rolfs (2013) Stroke 44, 340</a>
M421V	unknown	Amicus
X430Q	unknown	Amicus