

Datum 28.04.2011

Choroba

[3-Methylcrotonyl-CoA carboxylázový deficit](#)

A289V (MCCC1), D532H (MCCC1), E99Q (MCCC2), I437V (MCCC2), L437P (MCCC1), R193C (MCCC2), R385S, S173L, V339M (MCCC2)

[Acrodermatitis enteropathica \(SLC39A4\)](#)

1223_1227delCCGGG, L48X

[Alpha-1 antitrypsinový deficit \(SERPINA1\)](#)

S allele, Z allele

[Amyotrophická laterální skleróza \(ALS2\)](#)

1867delCT

[Argininosuccinát-lyázový deficit \(ASL\)](#)

D87G, IVS5+1G>A, Q116X, Q354X, R193Q, R385C, V178M

[Autoimunní polyglandulární syndrom, typ I \(AIRE\)](#)

64-69del6, 967-979del13, A502fsX519, C311Y, C311fsX376, C446G, C449fsX502, E298K, F77S, G218fsX284, G228W, H415fsX422, IVS11+1G>A, IVS3+2T>C, IVS3-2A>T, IVS7+1G>A, IVS8+5G>T, K83E, L28P, L29P, L323fsX372, L397fsX478, L417fsX478, L93R, L97P, M1L, M388fsX422, P252L, P370fsX370, P539L, Q358X, R139X, R15C, R203X, R257X, R303P, R433/C434fsX, S135fsX147, T16M, V80L, W78R, X546C, Y85C, Y90C

[Bartterův syndrom typ 4A \(BSND\)](#)

G47R

[Beta-ketothiolázový deficit \(ACAT1\)](#)

149delC, G152A, G183R, IVS11+2T>C, IVS8+1G>T, Q272X, R208X, T297M

[Beta-thalassemie \(HBB\)](#)

-28A>G, -29A>G, -87C>G, -88C>T, 17A>T, 41/42-TTCT, Hb, IVS1+110G>A, IVS1+1G>A, IVS1+5G>T, IVS1+6T>C, IVS2+1G>A, IVS2+654C>T, IVS2+745C>G, IVS2+849A>C, IVS2+849A>G, Malay, cd24T>A, cd39C>T, cd44-C, cd8-AA, cd8/9+G

[Biotinidázový deficit \(BTD\)](#)

A171T, C33FfsX36, D444H, Q456H, R538C

[Bloomův syndrom \(BLM\)](#)

C1055S, Q645X, Q975fsX, R836fsX, R899X, S186X, W428X, W567X, W803fsX, blmAsh

[Canavanova choroba \(ASPA\)](#)

245insA, 433-2A>G, 827delGT, A305E, C218X, E285A, F295S, G274R, M195R, P280S, Y109X, Y231X

[Carnitinová deficiencie, primární systémová \(SLC22A5\)](#)

N32S, P46S, R169W, R254X, T219fsX284, T440M, T468R, W283C, Y211C, Y4X

[Cerebrotendinózní xanthomatóza \(CYP27A1\)](#)

A216P, D354G, E195X, E408X, G145G, G472A, IVS2+1G>A, IVS4+1G>A, IVS6+1G>A, IVS6-1G>T, IVS7+1G>A, IVS7+5G>T, K284X, P384L, P401R, P441S, Q159X, Q461X, R127Q, R127W,

R137W, R231X, R270X, R395C, R405Q, R405W, R474W, T339M, W260X

[Citrullinemie typ I \(ASS1\)](#)

952delG, E191K, G390R, IVS6-2A>G, K310Q, Q380X, R272C, R304W, R307C, R86H, R95S, S180N, V269M, V345G, Y190D

[Crigler-Najjarův syndrom \(UGT1A1\)](#)

1043delA, 1186delG, 1220delA, 1223insG, 397_402delCAACAA, 517delC, 652insT, 801delC, 878_890del, 973delG, A292V, A368T, A401P, A478D, C177R, C280X, E463A, F170del, G377V, G493R, H376R, H39D, I294T, I370V, IVS1+1G>C, IVS3-2A>G, IVS4+1G>T, IVS4-1G>A, K428E, K437X, L131P, L175Q, L233R, L443P, M204V, N279Y, N400D, P34Q, P387R, Q185P, Q239fsX256, Q283X, Q331R, Q331X, Q357R, Q357X, R209W, R336W, R341X, R403C, S375F, S381R, S488F, V160E, V225G, W335X, W40R, W483X (TAG), W483X (TGA), Y192X, Y486D, Y74X

[Cystická fibrosa \(CFTR, MiM ID 602421\)](#)

deltaF508, CFTRdele2,3(21kb), G551D, N1303K, 1812-1G>A, G542X, 1949del84, 1898+1G>A, 2043delG, R347P, 2055del9>A, 2105del13ins5, W1282X, 1677delTA, 2108delA, 1078delT, 2184delA, R1162X, 2307insA, 2789+5G>A, 2869insG, 3120+1G>A, deltaI507, 1717-1G>A, 3120G>A, 3171delC, 3272-26A>G, 3659delC, 3667ins4, 3791delC, 3849+10kbC>T, 3876delA, 3905insT, 394delTT, 405+1G>A, 405+3A>C, 444delA, 574delA, 621+1G>T, 663delT, 711+1G>T, 712-1G>T, 846delT, 935delA, 936delTA, A455E, A561E, C524X, D1152H, G1349D, G178R, G330X, G622D, G85E, I148T, K710X, L206W, L558S, M1101K, P205S, P574H, P750L, Q1100P, Q1238X, Q359K/T360K, Q493X, R1158X, R117H, R334W, R352Q, R553X, R560T, R709X, S1196X, S1251N, S364P, S549N, S549R (A>C), S549R (T>G), V232D, V520F/I, W1089X, W1204X, deltaF311

[Deficit A\(CoA\)dehydrogenasy m. kyselin se středním řetězcem \(ACADM\)](#)

K304E, Y42H

[Deficit A\(CoA\)dehydrogenasy m.kyselin s krátkým řetězcem \(ACADS\)](#)

A199V, I390M, M370V, Q365H, R107C, R139C, R380W, R46W, S353L, T169P, W177R

[Deficit A\(CoA\)dehydrogenasy m.kyselin s velmi dlouhým řetězcem\(ACADVL\)](#)

V283A

[Diabetes, permanentní neonatální](#)

E382K (ABCC8), IVS8+2T>G (GCK), N72S (ABCC8), P45L (ABCC8), R397L (GCK)

[Dihydropyrimidin dehydrogenázový deficit \(DPYD\)](#)

A777S, E386X, H978R, I560S, IVS11+1G>T, IVS14+1G>A, M182K, P86L, R235W, R886H, V335L

[Dubin-Johnsonův syndrom \(ABCC2\)](#)

I1173F, R1150H

[Dysautonomie familiární \(IKBKAP\)](#)

IVS20+6T>C, R696P

Ehlers-Danlosův syndrom -hypermobilní typ (TNXB)

2116_2117dupGT, 3551_3552delAA

Ehlers-Danlosův syndrom Typ VII (ADAMTS2)

Q225X, W795X

Ehlers-Danlosův syndrom typ VI (PLOD1)

1362delC, 153dupC, 1702insC, 467-2delA, 975+2_975+3insTT, A667T, G678R, H706R, Q327X, Q49X, R319X, R670X, W446G, W612C, Y142X, Y511X

Ethylmalonová acidurie (ACADS)

511C>T, 625G>A

Faktor XI - deficit (F11)

C128X, E117X, F283L, IVS14+1G>A

Fanconio anemie (FANCC)

IVS4+4A>T, 322delG, R548X, Q13X, R185X, L554P

GM1-gangliosidosa (GLB1)

R59H

Galactosemie (GALT)

E203K, F171S, IVS2-2A>G, K285N, L195P, L218L (c.652C>T), N314D, Q188R, S135L, Y209C

Galaktokinázový deficit (GALK1)

G349S, P28T, Q382X, R256W, T344M

Gaucherova choroba (GBA)

84GG, D409H, IVS2+1G>A, N370S, R463C, V394L

Glutarová acidemie, typ 1 (GCDH)

A293T, A421V, R227P, R402W, V400M

HMG-CoA lyázový deficit (HMGCL)

504_505delCT, E37X, R41Q

Hemochromatóza

C282Y (HFE), G320V (HFE2), H63D (HFE), M172K (TFR2), S65C (HFE), Y250X (TFR2)

Hemoglobin C (HBB)

Hemoglobin C

Hemoglobinopatie E (HBB)

Hemoglobin E

Hepatorenální gykonenosa, typ 1A-von Gierke (G6PC)

378_379dupTA, 79delC, G188R, G270V, Q242X, Q347X, R83C, R83H, deltaF327

Homocystinurie klasická (CBS)

1566delG, 1591delTTTCG, 1622ins4, 298fsX329, A114V, A155V, A226T, A355P, C109R, C165Y, D376N, D444N, D47E, E128D, E144K, E176K, E239K, E302K, G116R, G139R, G148R, G151R, G307S, G347S, G85R, H232D, I278T, I435T, IVS11-2A>C, IVS12+1G>A, IVS8+1G>A, IVS9+1G>T, K102N, K384E, L101P, L539S, M126V, P145L, P290L, P49L, P78R, P88S, R121C, R266G, R266K, R336C (C>T), R336C (delCC/insTT), R336H, R369C, R369H, R379W, R491C, R58W, S217F, S349N, S466L, T191M, T257M, T353M, V168A, V320A, V354M, V371M, W43X

Homocystinurie, typ cbIE (MTRR)

1622_1623dupTA, 1726delTTG, 1953-6_1953-2del5, R3W, R525X

Hurlerův syndrom (IDUA)

1044delCGACAA, 1695del11, 1814_1815delTT, Q70X, W402X

Karboxylázová deficiencie mnohočetná (HLCS)

780delG, D571N, G581S, L237P, R508W, R665X, V550M

Kortikosteron methyl-oxidázový deficit (CYP11B2)

104_108delTGCTG, G435S, R181W, V386A

Krabbeho choroba (GALC)

1424delA, A625T, G270D, R168C, T513M

Lipoprotein lipázový familiární deficit (LPL)

G188E

MTHFR deficit (MTHFR)

1541_1542delAG, A116T, A1298C (E429A), C193Y, C677T (A222V), E470X, E586K, G149V, G387D, I153M, IVS1-1G>T, IVS4+1G>A, IVS4-2A>G, K584X, L323P, L333P, M338T, N324S, P251L, P572L, R157Q, R183X, R325C, R357C, R377C, R51P, R52Q, R535W, R567X, R594Q, R6X, S440L, T227M, W339G, X657S

Methylmalonová acidemie

503delC (MMAA), E117X (MUT), G717V (MUT), N219Y (MUT), R108C (MUT), R145X (MMAA), R369C (MUT)

Mucolipidosa II (GNPTAB)

1581delC, 3503_3504delTC, 616_619delACAG, Q104X, Q845X, R1189X, R1205X

Mucolipidosa III

347_349_delACA (GNPTG), 499dupC (GNPTG), IVS17+6T>G (GNPTAB), K4Q (GNPTAB)

Mucolipidosa IV (MCOLN1)

IVS3-2A>G

Nemoc javorového syru

E372X (BCKDHB), G278S (BCKDHB), R183P (BCKDHB), Y438N (BCKDHA)

Nephrotický syndrom, rezistentní na steroidy (NPHS2)

1036delC, 436delA, R138Q

Niemann-Pickova choroba

E20X (NPC2), G992W (NPC1), H421Y (SMPD1), I1061T (NPC1), L302P (SMPD1), P330SfsX382 (SMPD1), R496L (SMPD1), deltaR608 (SMPD1)

Phenylketonurie (PAH)

A403V, E280K, F39L, I65T, IVS10-11G>A, L48S, P281L, R158Q, R243X, R261Q, R408Q, R408W, V245A, Y414C

Polycystická choroba ledvin (PKHD1)

T36M, R496X, I2944fs, I2957T, I3177T, P805L, Q3392X, R3482C, D3230fs, I222V, V3471G

Pompeho choroba (GAA)

2741AG>CAGG, D645E, G309R

Prekallikreínová deficiencie (KLKB1)

C529Y, W383X

Propionová acidemie

1172_1173insT (PCCB), 1218del14ins12 (PCCB), R399Q (PCCA), R410W (PCCB), T428I (PCCB)

Prothrombinový deficit (F2)

C181Y, D161Y, E352K, R263C, R2W, R314C, R457Q, R538C

Pseudovitaminová D-deficientní křivice (CYP27B1)

3398dupCCCACCC, 958delG, IVS3+1G>A, R389H

Rh-null syndrom (RHAG)

V270I

Sandhoffova choroba (HEXB)

76delA, IVS2+1G>A, S62L

Sick sinus syndrom (SCN5A)

G1408R, P1298L, R1632H, T220I

Spherocytosa

1278insTATC, 5703+16C>T (ANK1), 613delC, A142T (EPB42), C458Y, D175Y (EPB42), G269S, I335F, IVS12+1G>C, IVS2+1G>C, IVS5-1G>T, IVS6+1G>A (EPB42), IVS9+1G>A, IVS9-1G>T, R170Q, R170W, R178H/L, R310Q (EPB42), R317C (EPB42), R499H, R504C, S210F, V192L, V463I (ANK1), W119X (EPB42), W329X, deltaTTC910-912

Spinální muskulární atrofie (SMN1)

dele Ex7

Srpkovitá anemie (HBB)

Hemoglobin S

Středomořská horečka (MEFV)

A744S, K695R, M680I, M694I, M694V, R408Q, R653H, R761H, V726A

Tay-Sachsova choroba (HEXA)

1278insTATC, 5703+16C>T (ANK1), 613delC, A142T (EPB42), C458Y, D175Y (EPB42), G269S, I335F, IVS12+1G>C, IVS2+1G>C, IVS5-1G>T, IVS6+1G>A (EPB42), IVS9+1G>A, IVS9-1G>T, R170Q, R170W, R178H/L, R310Q (EPB42), R317C (EPB42), R499H, R504C, S210F, V192L, V463I (ANK1), W119X (EPB42), W329X, deltaTTC910-912

Tay-Sachsova pseudodeficiencie (HEXA)

R247W, R249W

Thrombocytopenie, kongenitální amegakaryocytová (MPL)

R102P, R43X

Tyrosinemie (FAH)

G337S, P261L, Q64H, W262X

Von Willebrandova choroba typ 2 Normandy (VWF)

C1060R, C1225G, C788R, C788Y, C804F, D879N, E1078K, E787K, G785E, H817Q, M771V, P812L, Q1053H, R763G, R782W, R816Q, R816W, R854Q, T791M, Y795C

Von Willebrandova choroba typ 3 (VWF)

1384delG, 1657dupT, 191delG, 2016_2019delCTCT, 2157delA, 2269_2270delCT, 276delT, 3258_3259insT, 3736_3737dupCC, 374del14, 4092_4093delAC, 4324dupAGTGTGGA, 6182delT, 7139dupT, 7172_7173insT, 7674dupC, 7683delT, 892dupG, C1071F, C2174G, C2362F, C2671Y, C2739Y, C2754W, C2804Y, D47H, E620X, IVS28+1G>A, IVS29+10C>T, IVS40-1G>C, IVS45+7C>T, IVS50+3G>T, IVS7+1G>A, IVS9-1G>A, L1267X, Q1346X, Q218X, Q2544X, Q565X, R1315C, R1853X, R2434X, R2535X, R273W, R324X, R365X, R373X, S71X, V1314F/3940delG, W222X, W377C, Y1456X, Y1542X, Y357X, Y610X

Ztráta sluchu nesyndromická (DFNB1 a DFNB9)

167delT (GJB2), 235delC (GJB2), 35delG (GJB2), L90P (GJB2), Q829X (OTOF), V37I (GJB2)

Ztráta sluchu nesyndromická (DFNB59)

113dupT, 509_512delCACT, 726delT, 988delG, L244R, R167X