Diagnostic Moléculaire en Rythmologie

Liste GENES

N350 = BHN **3270**
N351= BHN 5570

Syndrome du QTL isolé

Niveau1-N350 ; Panels de gènes analysés : KCNE1 (NM_000219.3); KCNE2 (MN_172201.1); KCNQ1 (NM_000218.2); KCNH2 (NM_000238.2); SCN5A (NM_198056.2)

Niveau2-N351 Panel de gènes analysés KCNE1 (NM_000219.3); KCNE2 (MN_172201.1); KCNQ1 (NM_000218.2); KCNH2 (NM_000238.2); SCN5A (NM_198056.2) ; AKAP9 (Yotiao) (NM_005751.4); ANK2/ANKB (MN_001148.4); CACNA1C (NM_199460.3); CALM1 (NM_006888.4); CALM2 (NM_ 13056241); CASQ2 (NM_001232.3); CAV3 (MN_033337.2); KCNJ2 (NM_000891.2); KCNJ5 (NM_000890.3) ; RYR2 (NM_001035.2); SCN4B (NM_174934.3); SNTA1 (NM_003098.2) ; TRDN (triadin) (NM_006073.3)

Syndrome du QTL dit syndromique

Syndrome d'andersen-Tawil Niveau1-N350

Gène analysé : KCNJ2 (NM_000891.2)

Syndrome de Timothy Niveau1-N350

Gène analysé : CACNA1C (NM_199460.3)

Syndrome de Jervell et Lange-Nielsen Niveau1-N350

Panel de gènes analysés: KCNQ1 (NM_000218.2) **;** KCNE1 (NM_000219.3)

FA Fibrillation atriale

Niveau1 N350 : SCN5A (NM_198056.2) ; KCNQ1 (NM_000218.2);

Niveau2 N351 : SCN5A (NM_198056.2) ; KCNQ1 (NM_000218.2); KCNH2 (NM_00238.3) ; KCNE2 (MN_172201.1); KCNJ2 (NM_000891.2); SCN1B (NM_199037.3) ; SCN2B (NM_004588.4); SCN3B (NM_018400.3) ; ABCC9 (NM_005691.5) ; NKX2-5 (NM_004387.3)

QT court Forfait N351

QT court : CACNA1C (NM_199460.3) ; KCNQ1 (NM_000218.2); KCNH2 (NM_000238.2); KCNJ2 (NM_000891.2) ; CACNB2 (NM_201596.2)

TVC Niveau1-N350-

Panel de gènes analysés: CASQ2 (NM_001232.3); RYR2 (NM_001035.2); TRDN (triadin) (NM_006073.3)

TVC Niveau2-N351

Panel de gènes analysés: CASQ2 (NM_001232.3); RYR2 (NM_001035.2); TRDN (triadin) (NM_006073.3) ; ANK2/ANKB (MN_001148.4); CALM1 (NM_006888.4); CALM2 (NM_ 13056241); KCNJ2 (NM_000891.2)

DVDA Niveau1-N351 forfait 20-100KB=BHN5570

Niveau 1 N350 ; Panel de gènes analysés : PKP2 (NM_004572.3) ; DSC2 (NM_024422.3); DSG2 (NM_001943.3); DSP (NM_004415.2); LMNA (MN_170707.2)

Niveau2 N351 ; Panel de gènes analysés : PKP2 (NM_004572.3) ; DSC2 (NM_024422.3); DSG2 (NM_001943.3); DSP (NM_004415.2); LMNA (MN_170707.2) ; DES (NM_001927.3) ; JUP (NM_002230.2) ; PLN (NM_002667.3); RYR2 (NM_001035.2; TGFB3 (NM_003239.2), TMEM43 (NM_24334.2)

Syndrome de BRUGADA

Niveau1-N350 ; Panel de gène analysé SCN5A (NM_198056.2)

Niveau2-N351 ; Panel de gènes analysés SCN5A (NM_198056.2) ; SCN10A (NM_ 006514.3) ; SCN1B (NM_199037.3) ; SCN2B (NM_004588.4); SCN3B (NM_018400.3) ; GPD1L (MN_015141.3) ; CACNA1C (NM_199460.3) ; CACNB2 (NM_201596.2) ; CACNA2D1 (NM_000722.2) ; KCNE3 (NM_005472.4) ; KCND3 (NM_004980.4) ; KCNJ8 (NM_004982.2) ; KCNE1L (NM_012282.2) ; RANGRF (NM_016492.4) ; HCN4 (NM_005477.2); SLMAP (NM_007159,2); TRPM4 (NM_017636 ;3) ; PKP2 (NM_004572.3)

ERS Early Repolarization Syndrome N350

KCNJ8 (NM_004982.2) ; CACNA1C (NM_199460.3) ; CACNB2 (NM_201596.2) ; CACNA2D1 (NM_000722.2)

Trouble de conduction

Niveau1 N350 : SCN5A (NM_198056.2) ; LMNA (MN_170707.2) ; NKX2-5 (NM_004387.3)

Niveau2 N351 : SCN5A (NM_198056.2) ; LMNA (MN_170707.2) ; NKX2-5 (NM_004387.3) ; HCN4 (NM_005477.2); SCN1B (NM_199037.3) ; TRPM4 (NM_017636 ;3),