Heterogeneity of Clinical Syndromes Related to Loss of Function Mutations in *KCNJ2*

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XS WKH PLQRULW\ RI GLVHDVH
R .LU LQ DGGLWLRQ WR 3,3 ELQGLQJ
HQGRSODVPLF UHWLFXOXP H[SRUW VHTX
PHFKDQLVP IRU .LU ORVV G0 DRN&N HQ

mutations and

various inherited arrhythmia syndromes, such as Andersen-Tawil Syndrome (ATS) Short QT Syndrome 3 and Catecholaminergic ventricular arrhythmias and craniofacial dysmorphic features. Cardiac presentations on ECG reveal prolongation of the QT interval, pre-necessary for Kir2.1 to open [1] and ATS has been shown to be a result of

KCNJ2 mutations that DvHVVRFLDWHG ELQGLQJ RU DQ DOORVWHULF
FRQIRUPDWLRQDO FKDQJH OHDGLQJ WR GHFUHDVHG .LU FXUUHQW > @ 2WKHU
$76 FDX.&1J PXWDWLRQV LQFOXGH GLVWXUEDQFHV LQ WKH SRUH
VHOHFWLYLW\ \PLVIROGHG RU VHTXHVWHUHG SURWHLQV EXW WKHVH PDNH

E\ H[HUFLVH RU VWUHVV $GGLWLRQDOO\ LQ WHUPV RI IXQFWLRQDO FKDUDFWHUL]DWLRQ PRVW .LU PXWDWLRQV DVVRFLDWHG ZLWK $76 H[KLELW GRPLQDQW QHJDWLYH .LU FXUUHQW ZKHQ FR H[SUHVVHG ZLWK :7 .LU ZKLOH WKH .LU PXWDWLRQV DVVRFLDWHG ZLWK &397 GLVSOD\HG PDUNHGO\ GHFUHDVHG RXWZDUG [illegible]

H PHFKDQLVPV XQGHUO\LQJ [illegible] ZLWK WKHVH PXWDWLRQV DUH QRW FOHDU DW WKLV WLPH KRZHYHU WKH\ DUH DUHD RI DFWLYH UHVHDUFK IRU RXU ODERUDWRU\ DQG RWKHUV 3KRVSKDWLG\OLQRVLWRO ELVSKRVSKDWH 3,3 LV D ZHOO HVWDEOLVKHG UHJXODWRU RI LQZDUG UHFWLI\LQJ SRWDVVLXP FKDQQHOV > @ 0DQ\ RI WKH .LU PXWDWLRQV DVVRFLDWHG ZLWK $76 DQG WKH 5 4 .&1- PXWDWLRQ DVVRFLDWHG &D97 [illegible] RI WKH FKDQQHO ZLWK 3,3 > @ $Q 1 WHUPLQXV PXWDWLRQ WKDW DOWHUV .LU VHQVLWLYLW\ WR 3,3 KDV EHHQ VKRZQ WR H[JJHUDWH WKH LQKLELWLRQ RI .LU E\ D GLYDOHQW FDWLRQ EDUL\XP > @ K\SRWKHVLV IRU DGUHQHUJLF PRGXODWHG FKDQQHO IXQFWLRQ PD\ EH HQKDQFHG LQKLELWLRQ LQ WKH SUHVHQFH RI LQFUHDVHG FHOOXODU FDOFLXP >

,PSRUWDQW TXHVWLRQV UHPDLQ LQ WKH SHQRW\SH JHQRW\SH FRUUHODWLRQ IRU FOLQLFDO V\QGURPHV DVVRFLDWHG &DWR [illegible] SKHQRW\SLQJ DV ZHOO DV IXQFWLRQDO VWXGLHV ZKLFK IRFXV RQ WKH