Function to Presentation

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Introduction:

This file is used to judge empirical relationships between functional parameters and calculated penetrance derived from the SCN5A dataset. There is also a sensitivity analysis done at the end using peak current as an example.

```
require(Hmisc)
library(tableone)
library(ggplot2)
library("DBI")
library("RSQLite")
library(wCorr)
library(meta)
knitrSet(lang = 'markdown', echo = TRUE)
```

Read data from SQL database and tabulated files

```
con = dbConnect(SQLite(),
dbname="/Users/B/Dropbox/SCN5A/BrettsSandbox/paper/data/VariantSCN5A-new.db")
alltables = dbListTables(con)
my.data <- dbReadTable(con, 'VariantSCN5A')
my.data[my.data=='NA'] <- NA
d<-my.data
dbDisconnect(con)
d$resnum<-as.integer(d$resnum)</pre>
```

```
d$gnomAD[is.na(d$gnomAD)] <- 0
d$gnomAD<-as.numeric(d$gnomAD)</pre>
d$ipeak<-100*as.numeric(d$ipeak)
d$vhalfact<-as.numeric(d$vhalfact)</pre>
d$vhalfinact<-as.numeric(d$vhalfinact)</pre>
d$recovfrominact<-log10(100*as.numeric(d$recovfrominact))
d$recovfrominact_dist<-log10(100*as.numeric(d$recovfrominact_dist))
d$ilate[as.numeric(d$ilate)==0]<-NA
d$ilate_norm<-log10(d$ipeak*as.numeric(d$ilate)+0.00001)
d$ilate<-log10(100*as.numeric(d$ilate)+0.00001)
d$ilate_dist<-log10(100*as.numeric(d$ilate_dist)+0.00001)
d$total_carriers<-d$lqt3+d$brs1+d$unaff+d$gnomAD
d$weight = 1-1/(0.1+d$total_carriers) #weights
d$weightsMilder = 1-1/(1+d$total_carriers) #weights
# Adding in penetrance variables
abrs0=0.32
alqt0=0.11
beta0=1
d$LQT_penetranceBayesian<-(d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0)
d$BrS_penetranceBayesian<-(d$brs1+abrs0)/(d$total_carriers+1+alqt0)
d$all_penetranceBayesian<-(d$brs1+abrs0+d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0+abrs0)
servers<-read.csv("/Users/B/Dropbox/SCN5A/BrettsSandbox/paper/data/annotated_variants-trim.txt", sep =</pre>
provean <-read.csv("/Users/B/Dropbox/SCN5A/BrettsSandbox/paper/data/provean.txt", sep = "\t")</pre>
pph2 <-read.csv("/Users/B/Dropbox/SCN5A/BrettsSandbox/paper/data/pph2-short.txt", sep = "\t")
sift <-read.csv("/Users/B/Dropbox/SCN5A/BrettsSandbox/paper/data/SIFT.txt", sep = "\t")</pre>
d <- merge(d, servers, all = TRUE)</pre>
d <- merge(d, provean, all = TRUE)</pre>
d <- merge(d, sift, all = TRUE)</pre>
d <- merge(d, pph2, all = TRUE)</pre>
d<-d[!is.na(d$var), ]</pre>
d$eaRate<-as.numeric(d$eaRate)</pre>
d$blastpssm<-as.numeric(d$blastpssm)</pre>
d$pamscore<-as.numeric(d$pamscore)</pre>
d<-d[d$total_carriers>0, ]
```

Defining Functions

```
plt.loess <- function(fit, xlabel="X", ylabel="Y", xlims=NA, ylims=NA){
  if(sum(is.na(xlims) & is.na(ylims))>0){
    print(xlims)
    lines(min(fit$x)-1000, 0, xlim=c(min(fit$x), max(fit$x)), ylim=c(min(fit$y), max(fit$y)), ylab=ylab
} else{ if(is.na(xlims)){
    lines(min(fit$x)-1000, 0, xlim=c(min(fit$x), max(fit$x)), ylim=ylims, ylab=ylabel, xlab=xlabel)
} else{
    lines(min(fit$x)-1000, 0, xlim=xlims, ylim=ylims, ylab=ylabel, xlab=xlabel)
}
xrange <- seq(min(fit$x), max(fit$x), length.out = 100)</pre>
```

```
ps <- predict(fit, xrange, se=T)</pre>
  lines(xrange, ps$fit*100, lwd=5)
  lines(xrange, (ps$fit+1.96*ps$se.fit)*100, lty=2, lwd=5)
  lines(xrange, (ps$fit-1.96*ps$se.fit)*100, lty=2, lwd=5)
}
plt.disease <- function(func, funcName, disName, x1=c(0,200), sp=0.7){
  par(cex=1, bty='1', lwd=2)
  if (disName=="BrS_penetranceBayesian"){dis="BrS1"; pcolor="gray"}
  else if(disName=="LQT_penetranceBayesian"){dis="LQT3"; pcolor="gray"}
  else if (disName=="all_penetranceBayesian"){dis="BrS1 and LQT3"; pcolor="gray"}
  plot(d[,func], 100*(d[,disName]), pch=21,
     bg=pcolor, cex=log(d[,"total_carriers"])+1, lwd=1.5, axes=FALSE,
     cex.lab=1.5, ylab=paste("Penetrance (%",dis,")",sep=""), xlab = funcName,
     ylim = c(-1,101), xlim = xl)
  axis(side=1,lwd=3, cex.axis=1.5)
  axis(side=2,lwd=3, cex.axis=1.5)
  plt.loess(loess(d[,disName]~as.numeric(d[,func]),span = sp,
      weights = 1-1/(0.1+d[,"total_carriers"])), xlabel=funcName,
       ylims=c(0, 1))
}
```

Plots (final images made in plotting window)

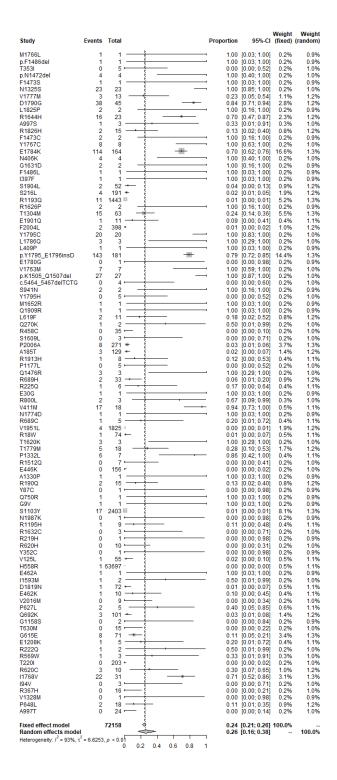
BrS1 meta analysis

```
#plots:
#forest plot of by variant
#BrS1 for variants that have peak current measured
f<-d[!is.na(d$ipeak),]
f<-f[order(f$ipeak, f$var),]
tmp<-metaprop(brs1,total_carriers,data = f,studlab = var)
forest(tmp)</pre>
```

					Weight (fixed) (Weight
c.3480delT	2 events	Total	+	0.67 (0.09; 0.99)		0.4%
c.5040_5042delTTAInsC c.5280delG c.5445_5446insT	2 2	10 10 10 10 10 10 10 10 10 10 10 10 10 1		0.67 (0.09; 0.99) 1.00 (0.03; 1.00) 1.00 (0.16; 1.00) 0.40 (0.05; 0.85)	0.1% 0.1% 0.4%	0.4% 0.4% 0.5%
D1430N D356N	9	- 1		1.00 (0.16; 1.00) 0.82 (0.48; 0.98)	0.1%	0.4%
D1490N D356N E473X G1408R G1712C G1740R G1743E G1743R	8	26	-	1.00 (0.03; 1.00) 0.29 (0.13; 0.49)	0.1% 1.9% 0.4°	0.4% 0.6% 0.4%
G1740R G1743E	10	10		1.00 (0.16; 1.00)	0.1%	0.4%
G1743R L846R 065X	12	18		0.67 (0.41; 0.87) 1.00 (0.03; 1.00) 0.33 (0.01; 0.91)	1.3% 0.1% 0.2%	0.6% 0.4% 0.4%
R104Q R104W	5 2	9	-	0.83 (0.36; 1.00) 0.67 (0.09; 0.99)	0.3%	0.5%
R1629X R1638X	2 3	6	+	0.33 (0.04; 0.78) 0.50 (0.12; 0.88)	0.1% 0.4% 0.5%	0.4% 0.5% 0.5%
R179X R222X	33	53		0.60 (0.15; 0.95) 0.62 (0.48; 0.75)	0.4% 4.1%	0.5% 0.6%
07743FL (J466FL (J466FL F15MC)	5		=	1.00 (0.48; 1.00) 0.67 (0.30; 0.93)	0.2%	0.4%
S1218 S910L	3	-		1.00 (0.29; 1.00)	0.1%	0.4%
W1191X W822X	2 4	ě	1	0.50 (0.07; 0.93) 0.67 (0.22; 0.96)	0.3%	0.5%
C5464_5467dMTCTG I1660V R282H	5	19		0.25 (0.01; 0.81) 0.26 (0.09; 0.51) 0.50 (0.16; 0.84)	1.2%	0.4% 0.6% 0.5%
G752R G1405R	8	10		0.80 (0.44; 0.97) 0.80 (0.28; 0.99)	0.5%	0.5% 0.5%
R23264 075084 0740490	1 0	24	=	0.04 (0.00; 0.21)	0.3%	0.5%
Y352C G351V	3	-		1.00 (0.03; 1.00)	0.1%	0.4%
M1766L A14278	0		-	1.00 (0.03; 1.00)	0.1%	0.4%
1230T P335L 1325R	1	12	=	0.00 [0.00; 0.25] 0.33 [0.01; 0.91] 1.00 (0.03; 1.00)	0.2% 0.2% 0.1%	0.4% 0.4% 0.4%
R376H p.K1493del	2	10	1	0.47 (0.21: 0.73) 0.67 (0.09: 0.99)	1.2%	0.6%
p.F1486del D1714G	0			0.00 (0.00; 0.98)	0.1%	0.4%
N406K D1690N	2	-		0.00 (0.00 0.60)	0.1%	0.4%
A2267 R367H	13	36 16	+	0.11 (0.03; 0.27) 0.81 (0.54; 0.96)	1.2%	0.6%
L1789Q V1378M	0	3	===	0.00 (0.00; 0.84) 0.00 (0.00; 0.71) 0.50 (0.01; 0.99)	0.1% 0.1% 0.2%	0.4% 0.4% 0.4%
G1158S C1850S	1			1.00 (0.00; 0.84)	0.1%	0.4%
8101K L8122 T3531 R889H R811H Q1832E p,N1472sel Q750R P1996S T220 P1298L E555K	1 5	20 13 15 16 16 16 17 17 18 18 18 18 18 18 18 18 18 18 18 18 18		0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	0.1%	0.4%
R811H Q1832E	3 2 2	33	-	0.09 (0.02; 0.24) 0.33 (0.04; 0.78) 0.06 (0.01: 0.24)	0.9% 0.4% 0.6%	0.5% 0.5% 0.5%
p.N1472del Q750R P1508S	0	-	=	0.00 (0.00; 0.60)	0.1%	0.4%
T2201 P1298L	4 2 0	200		0.01 (0.00; 0.04) 0.01 (0.00; 0.04) 0.00 (0.00; 0.26)	0.3% 0.7% 0.2%	0.5% 0.5% 0.4%
120100	1	10	===	0.10 (0.00; 0.45) 0.11 (0.00; 0.48) 0.00 (0.00; 0.48)	0.3% 0.3% 0.3%	0.5% 0.5% 0.4%
E1784K A551T	31 2	164	*	0.19 (0.13; 0.26) 0.67 (0.09; 0.99)	8.3% 0.2%	0.6%
KB17E A14288	1 8	15		0.00 (0.00; 0.37) 1.00 (0.03; 1.00) 0.44 (0.22; 0.66)	0.2% 0.1% 1.5%	0.4% 0.4% 0.6%
E1780G P701L P085U	1	31	1	1.00 (0.03; 1.00) 0.03 (0.00; 0.17)	0.1%	0.4%
E48K (397F	0	19	=	0.00 (0.03; 1.00) 0.00 (0.00; 0.18) 0.00 (0.00; 0.98)	0.1% 0.2% 0.1%	0.4% 0.4%
A STREAM CONTROL OF THE STREAM CONTROL OF TH		190 100 100 100 100 100 100 100 100 100	-1	1	0.2%	0.4% 0.5% 0.4%
L567Q Y1795H	6 2	6	1	0.67 (0.30, 0.93) 0.40 (0.05, 0.85)	0.7%	0.5%
0270K P1177L	1	-		0.50 (0.01; 0.99) 0.00 (0.00; 0.52)	0.1% 0.2% 0.2%	0.4% 0.4% 0.4%
G2988 R1898H T670M	0	12		0.00 (0.00; 0.46) 0.00 (0.00; 0.25) 0.00 (0.00; 0.22)	0.2%	0.4% 0.4% 0.4%
R1512W Y87C	1	10	-	1.00 (0.03:1.00)	0.1%	0.5%
8216L E446K	1 0	191 156		0.01 (0.00; 0.03)	0.3%	0.5%
L409P R1626H	0	12	= -	0.00 (0.00; 0.98)	0.2% 0.1% 0.2%	0.4% 0.4% 0.4%
R1309H D1819N G1319V	0	72		0.00 (0.00; 0.46) 0.00 (0.00; 0.05) 0.35 (0.14; 0.62)	0.2% 0.2% 1.3%	0.4% 0.4% 0.6%
R2012H Q1909R	1 0	10	=	0.10 (0.00; 0.45)	0.3%	0.5% 0.4%
R985C R526H	7 6	20	#==	0.27 (0.12; 0.48) 0.26 (0.10; 0.48)	1.7%	0.6%
p.K1505_Q1507del E1208K	0	27		0.03 (0.07; 0.70) 0.00 (0.00; 0.13) 0.00 (0.00; 0.52)	0.7% 0.2% 0.2%	0.5% 0.4% 0.4%
R27H D1275N T1779M	3	277 280 181 111 111 112 202 203 40 40 63	=	0.04 (0.01; 0.11) 0.11 (0.02; 0.28) 0.06 (0.00; 0.27)	0.9% 0.9% 0.3%	0.5% 0.5% 0.5%
L619F E1901Q	1 0	- 1		0.09 (0.00; 0.41) 0.00 (0.00; 0.28)	0.3%	0.5%
R34C G245K	0	3172		0.00 (0.00; 0.00)	0.2%	0.4%
R800L R340Q R1897W	0	25		0.00 (0.00; 0.71) 0.00 (0.00; 0.15) 0.00 (0.00; 0.12)	0.1% 0.2% 0.2%	0.4% 0.4% 0.4%
R1512Q E1053K P216W	5	40	=	0.00 [0.00; 0.41] 0.12 [0.04; 0.27]	0.2% 1.4% 1.5%	0.4%
R1913H D1790G	1	45	=	0.12 (0.00: 0.53) 0.02 (0.00: 0.12)	0.3%	0.5%
R1629Q K126E	4			0.44 (0.14; 0.79) 1.00 (0.03; 1.00)	0.7%	0.5%
T1304M T1620M H658R	2 6 7	60 11 63697		0.03 (0.00; 0.11) 0.55 (0.23; 0.03) 0.00 (0.00; 0.00)	0.6%	0.5%
R18Q E462A	0	10 63697 26 10 80 10	=	0.04 (0.00 0.20)	0.3%	0.5%
R1023H R190Q	3	80	-	0.04 (0.01; 0.11) 0.07 (0.00; 0.32)	0.9%	0.5% 0.5%
Q1476R T1620K O682W	0	101		0.00 (0.00; 0.71)	0.1%	0.4% 0.4% 0.4%
R620C N406S	- 1	10	#	0.10 (0.00; 0.45) 0.50 (0.01; 0.99)	0.3%	0.5%
V1328M E439K	1	10		1.00 (0.03; 1.00) 1.00 (0.03; 1.00)	0.1%	0.4%
G552R 8524Y 86491	0	1247		0.17 [0.00; 0.64] 0.00 [0.00; 0.00] 0.04 [0.00; 0.27]	0.3%	0.5%
A1113V A1330P	1 0	1		0.14 (0.00; 0.58) 0.00 (0.00; 0.98)	0.3%	0.5%
A6720 A672F A6470	0	1512		0.00 (0.00; 0.00) 0.00 (0.00; 0.04) 1.00 (0.03; 1.00)	0.1%	0.5%
A736V E1938K E1473C	5	-		0.62 (0.24 0.91) 0.20 (0.01; 0.72)	0.6%	0.5%
F1486L F532C C1621D	4	15	<u> </u>	0.00 (0.00; 0.98) 0.33 (0.10; 0.85)	0.1%	0.4%
K1493R L1308F	0	1512	-	0.00 [0.00; 0.84] 0.00 [0.00; 0.46] 0.01 [0.00; 0.04]	0.1% 0.2% 0.3%	0.4% 0.4% 0.5%
L1501V M1652R M1851V	9	-		0.13 (0.03; 0.34) 0.00 (0.00; 0.98) 0.00 m nn nn nn	0.9% 0.1% 0.1%	0.5% 0.4% 0.4%
N70K p.Y1795_E1796insO	15	101	-	0.08 (0.05; 0.13) 0.08 (0.05; 0.13)	0.3%	0.5%
P2006A Q673E	1 0	27	*	0.00 (0.00; 0.58) 0.00 (0.00; 0.02) 0.00 (0.00; 0.71)	0.3% 0.3% 0.1%	0.5% 0.4%
K1195H R1232W R1623Q	1 4 0	10	1	0.11 (0.00; 0.48) 1.00 (0.40; 1.00) 0.00 (0.00; 0.24)	0.3% 0.1% 0.2%	0.5% 0.4% 0.4%
R1626P R1632H 8210H	10	13		0.00 (0.00; 0.84) 0.77 (0.46; 0.95)	0.1%	0.4% 0.5%
R43Q R620H	0	10	=	0.00 (0.00; 0.25) 0.10 (0.00; 0.45)	0.2%	0.4%
81904L 8941N	1 0	50		1.00 (0.03; 1.00) 0.02 (0.00; 0.10) 0.00 (0.00; 0.84)	0.1% 0.3% 0.1%	0.4% 0.5% 0.4%
T512I T632M V232I	2 2	4		0.33 (0.01; 0.91) 0.50 (0.07; 0.93) 0.05 (0.01; 0.17)	0.3%	0.4% 0.5% 0.5%
V411M Y1767C	0	18		0.00 (0.00; 0.19)	0.2%	0.4%
R1193Q E1295K	18	144		0.01 (0.01; 0.02) 0.00 (0.00; 0.84)	5.8% 0.1%	0.6%
L618F 8262G	1 0	1221 181 181 181 181 181 181 181 181 181		0.01 [0.00; 0.98] 0.00 [0.00; 0.98]	0.3%	0.5%
V95I P468L R18W	1 0	74		0.11 [0.00; 0.48] 0.14 [0.00; 0.58] 0.00 [0.00; 0.05]	0.3%	0.5% 0.5% 0.4%
N1325S I137V IR4V	0	23		0.00 (0.00; 0.15) 0.00 (0.00; 0.84)	0.2% 0.1% 0.4%	0.4% 0.4%
Y1795C E30G	0	20	=	0.00 (0.00; 0.17)	0.2%	0.4% 0.4%
R569W T1526P	0	-		0.00 (0.00; 0.84) 0.00 (0.00; 0.71) 0.00 (0.00; 0.84)	0.1% 0.1% 0.1%	0.4% 0.4% 0.4%
N1987K S1103Y I1593M	0	240		0.00 (0.00; 0.98) 0.00 (0.00; 0.00) 0.00 (0.00; 0.00)	0.1% 0.2% 0.1%	0.4% 0.4% 0.4%
V1951M G358 V1763M	1	26 26		0.04 (0.00; 0.18) 0.03 (0.00; 0.18)	0.3%	0.5% 0.5%
F2004L F816Y	3	396		0.01 [0.00; 0.41] 0.01 [0.00; 0.02] 0.00 [0.00; 0.71]	1.0% 0.1%	0.5% 0.4%
L212P R568H P1644C	0		==	0.00 (0.00; 0.98)	0.1%	0.4%
A1330T P1090L	10	488		0.00 (0.00; 0.41) 0.02 (0.01; 0.04)	0.2%	0.4%
6014C N927K R481W	2 0 1	38		- 1.00 (0.16; 1.00) 0.00 (0.00; 0.98) 0.00 (0.00; 0.11	0.1% 0.1% 0.3%	0.4% 0.4% 0.5%
V1951L G983D H1849R	0	1825		0.00 (0.00; 0.00)	0.7% 0.2% 0.1%	0.5% 0.4% 0.4%
WARDEN W	1	23				
ANTITAD	0 0 0 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	1825		0.00 (0.00; 0.10) 0.00 (0.00; 0.71) 0.00 (0.00; 0.46) 0.00 (0.00; 0.98)	0.2% 0.1% 0.2% 0.1%	0.4% 0.4% 0.4% 0.4%
Fixed effect model		80526			0.1%	
Fixed effect model Random effects model Heterogenety: r ² = 85%, r ² =	4.2306, p	< 0.0	0 02 04 05 08	0.18 [0.14; 0.23] 1	-	100.0%

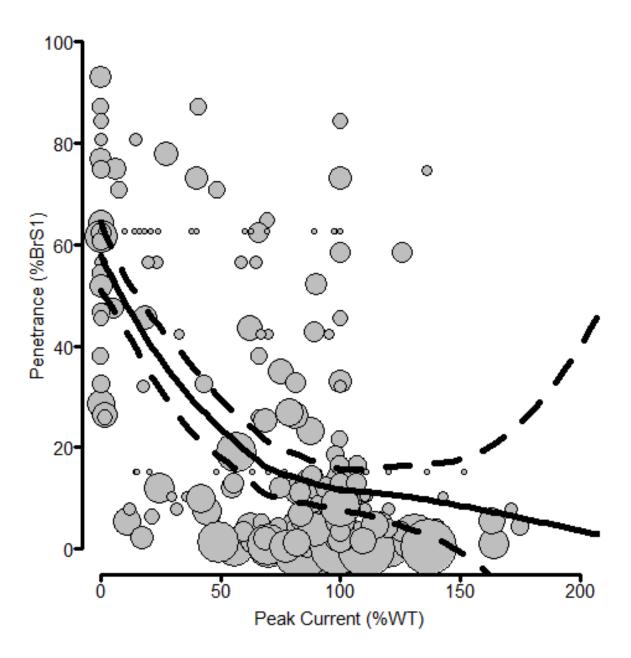
LQT3 meta analysis

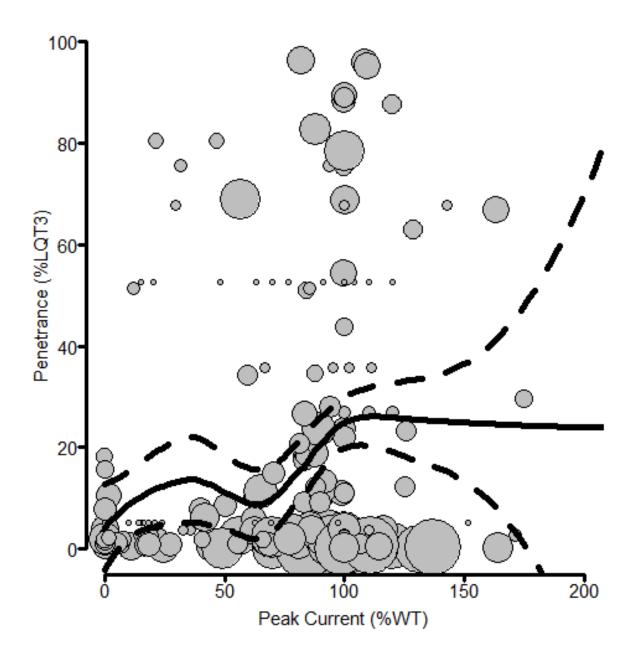
```
#LQT3 for variants that have late current measured
f<-d[!is.na(d$ilate),]
f<-f[order(-f$ilate, f$var),]
tmp<-metaprop(lqt3,total_carriers,data = f,studlab = var)
forest(tmp)</pre>
```

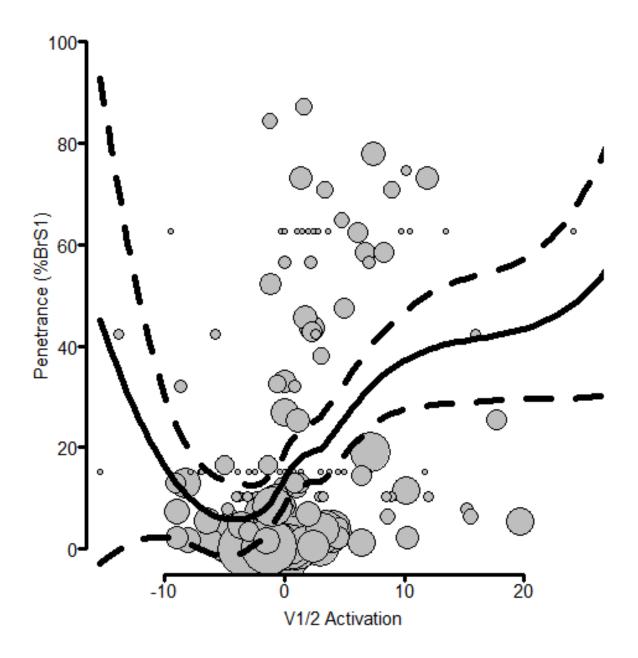


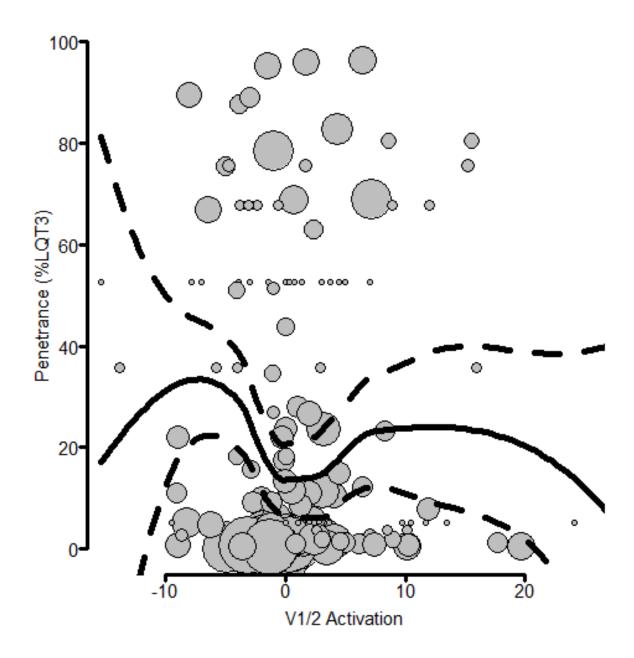
Functional Perturbation vs. Calculated Penetrance

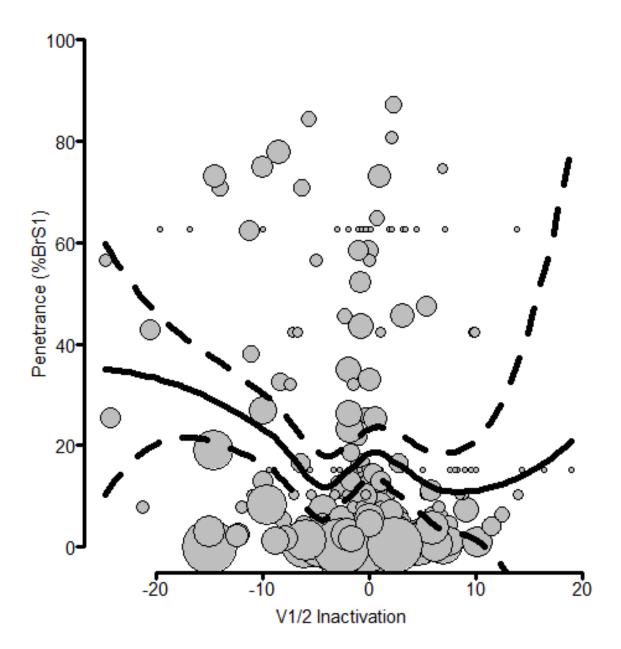
```
plt.disease("ipeak", "Peak Current (%WT)", "BrS_penetranceBayesian")
plt.disease("ipeak", "Peak Current (%WT)", "LQT_penetranceBayesian")
plt.disease("vhalfact", "V1/2 Activation", "BrS_penetranceBayesian", xl = c(-15,25))
plt.disease("vhalfact","V1/2 Activation", "LQT_penetranceBayesian", x1 = c(-15,25))
plt.disease("vhalfinact", "V1/2 Inactivation", "BrS_penetranceBayesian", x1 = c(-25,20))
plt.disease("vhalfinact","V1/2 Inactivation", "LQT_penetranceBayesian", x1 = c(-25,20))
plt.disease("recovfrominact", "Recovery from Inactivation (log[%WT])", "BrS_penetranceBayesian", xl = c(
plt.disease("recovfrominact", "Recovery from Inactivation (log[%WT])", "LQT_penetranceBayesian", xl = c(
plt.disease("ilate","Late Current (log[%WT])", "BrS_penetranceBayesian", x1 = c(1.5,4))
plt.disease("ilate", "Late Current (log[%WT])", "LQT_penetranceBayesian", xl = c(1.5,4))
plt.disease("ilate_norm", "Late Current (log[%WT]) Normalized", "BrS_penetranceBayesian", x1 = c(1.5,4))
plt.disease("ilate norm", "Late Current (log[%WT]) Normalized", "LQT penetranceBayesian", x1 = c(1.5,4))
plt.disease("ipeak", "Peak Current (%WT)", "all penetranceBayesian")
plt.disease('eaRate', 'Evolution Rate', 'all_penetranceBayesian', xl=c(0,4))
plt.disease('pph2_prob', 'PolyPhen-2', 'all_penetranceBayesian', xl=c(0,1))
plt.disease('SIFT.Score','SIFT', 'all_penetranceBayesian', xl=c(0,1))
plt.disease('provean_score','PROVEAN', 'all_penetranceBayesian', xl=c(-13,5))
plt.disease('CADD_RAW','CADD', 'all_penetranceBayesian', xl=c(0,8))
plt.disease('Condel_n','ConDel', 'all_penetranceBayesian', xl=c(0,1))
plt.disease('blastpssm','BLAST-PSSM', 'all_penetranceBayesian', xl=c(-7,7))
plt.disease('pamscore', 'PAM Score', 'all_penetranceBayesian', xl=c(-15,2))
plt.disease('aasimilaritymat','AA Similarity Matrix', 'all_penetranceBayesian', xl=c(-0.5,0.6))
```

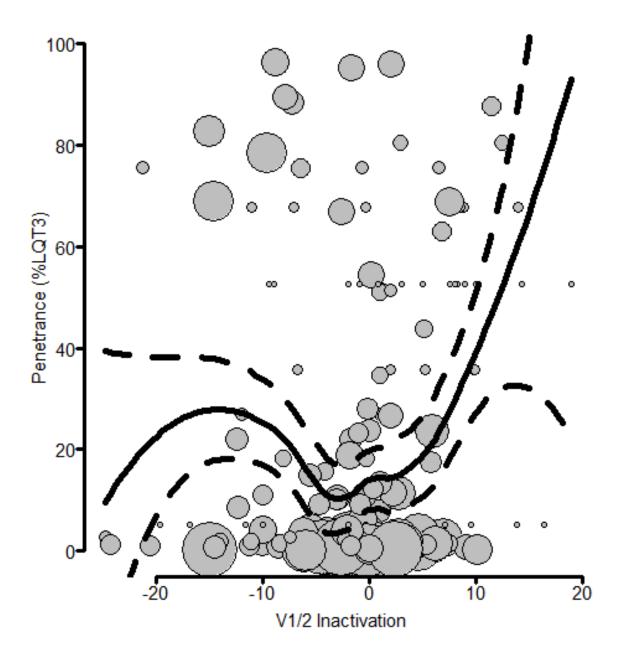


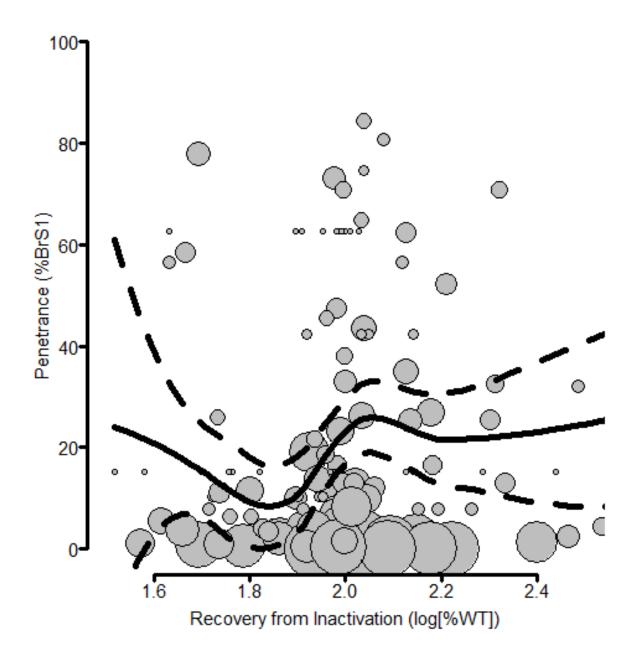


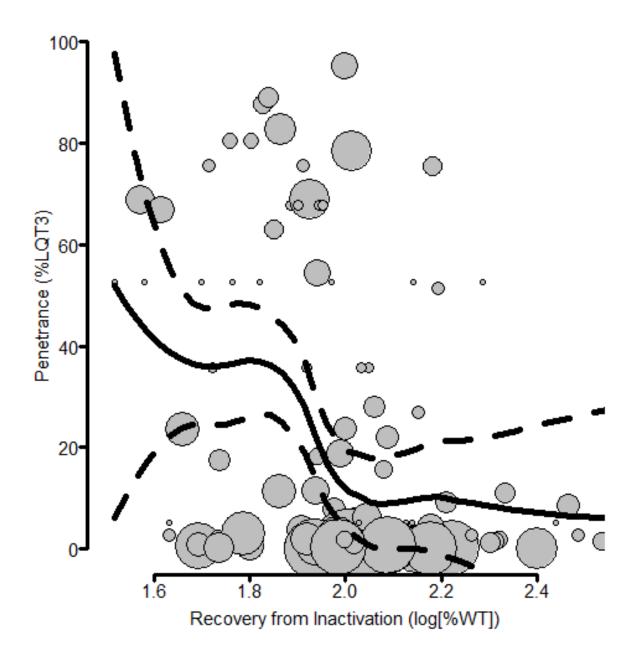


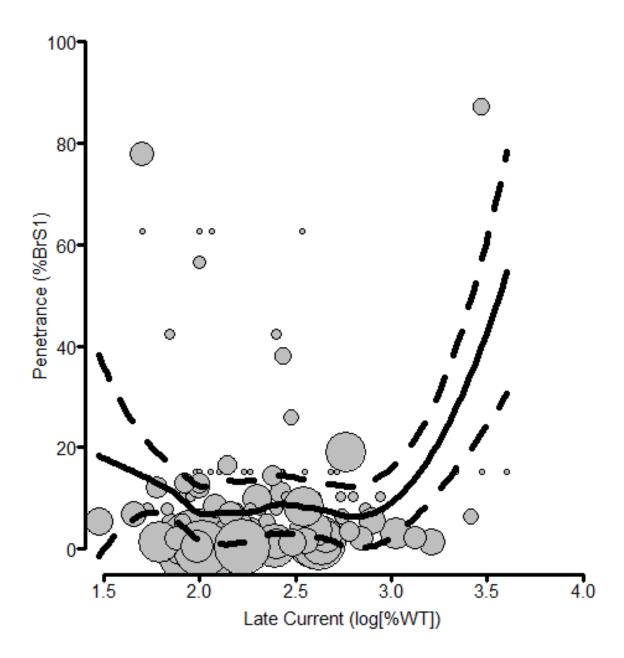


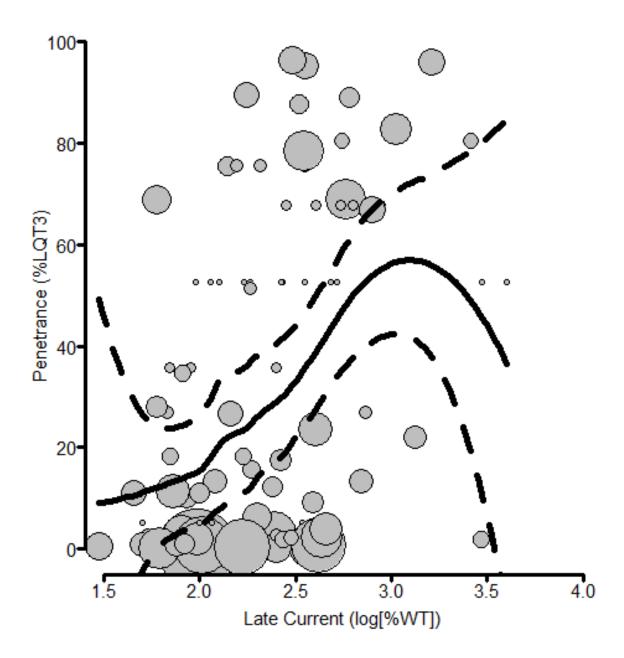


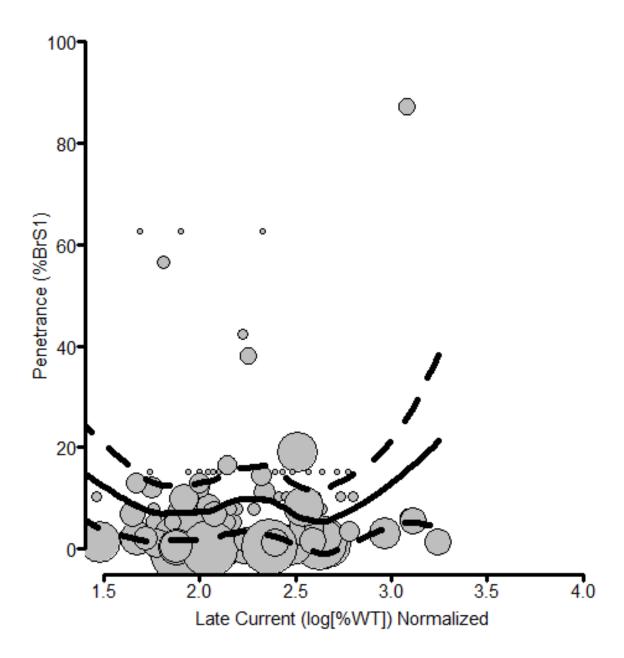


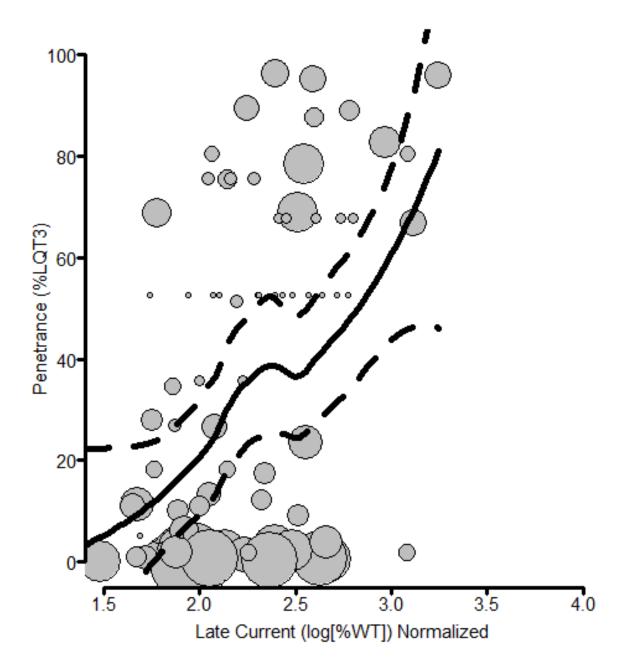


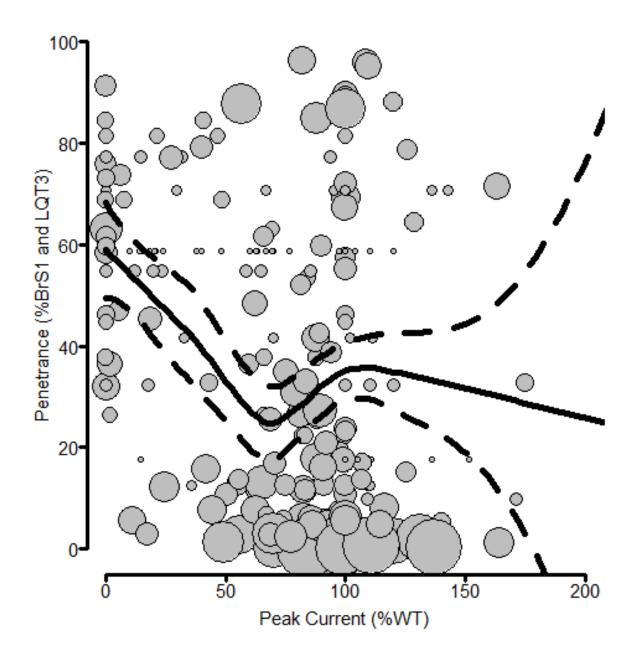


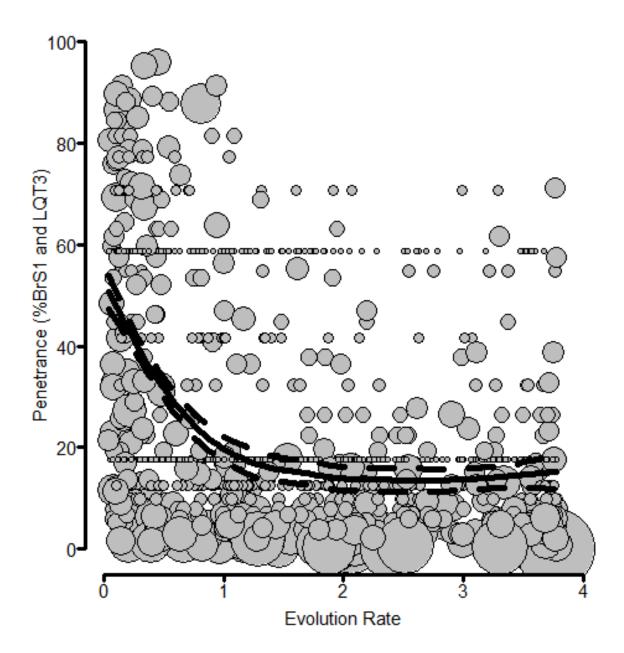


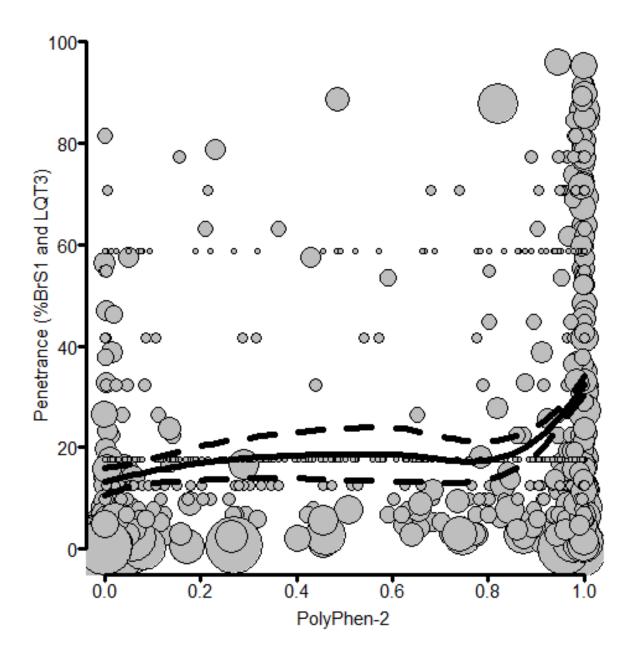


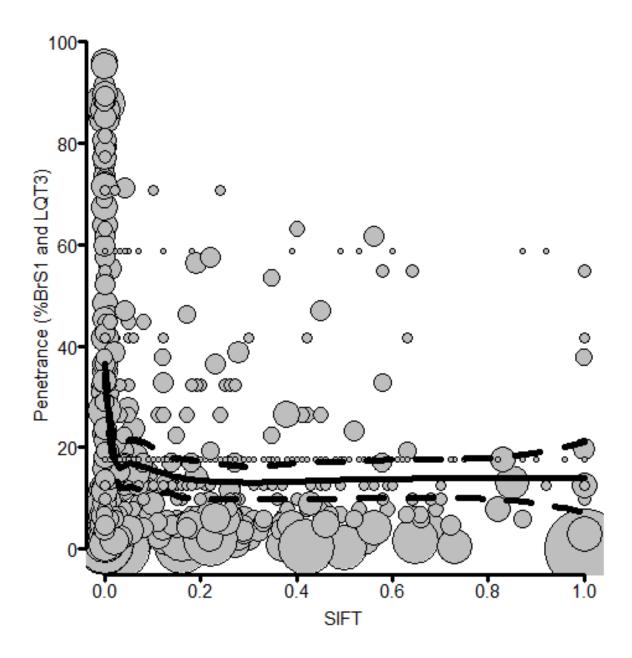


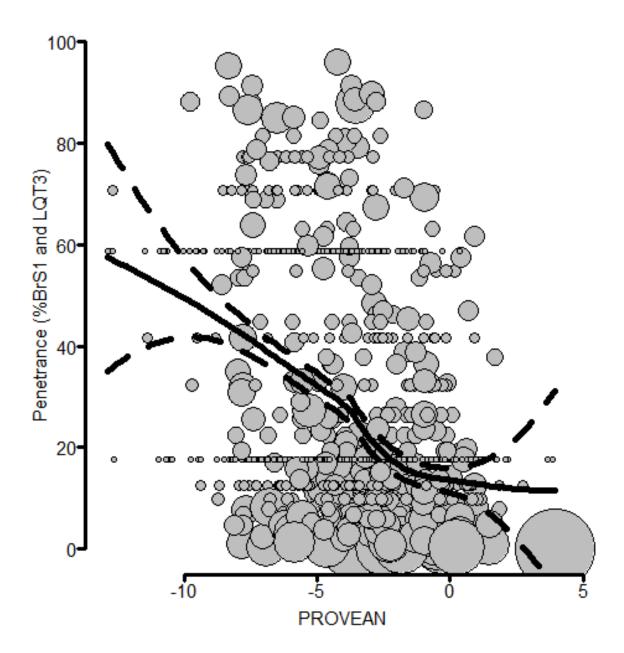


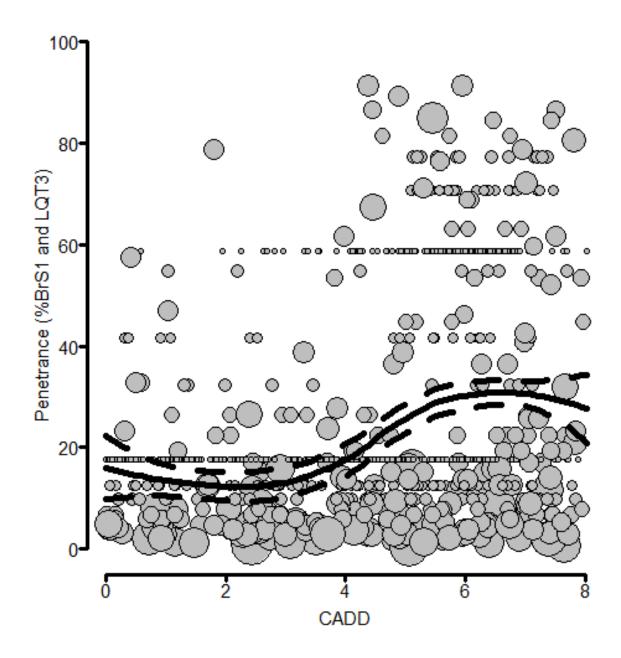


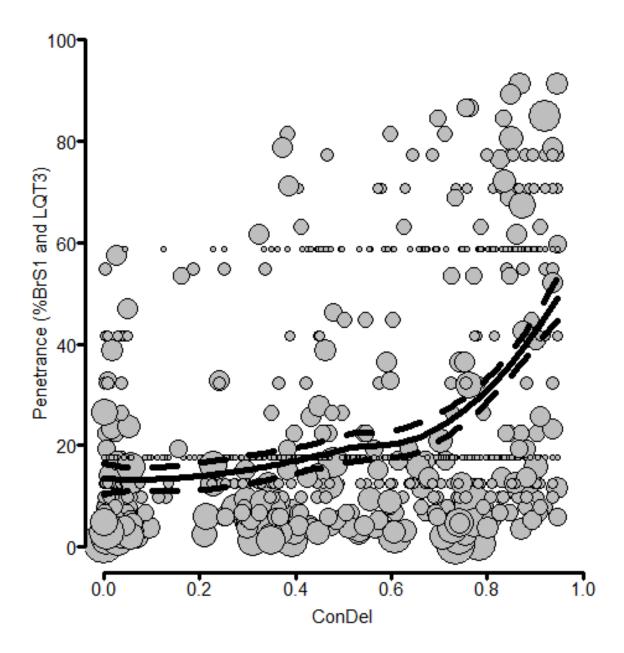


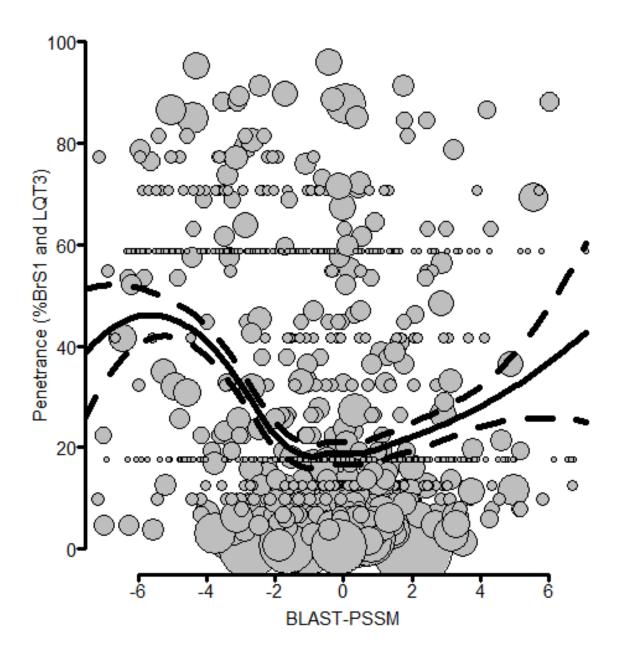


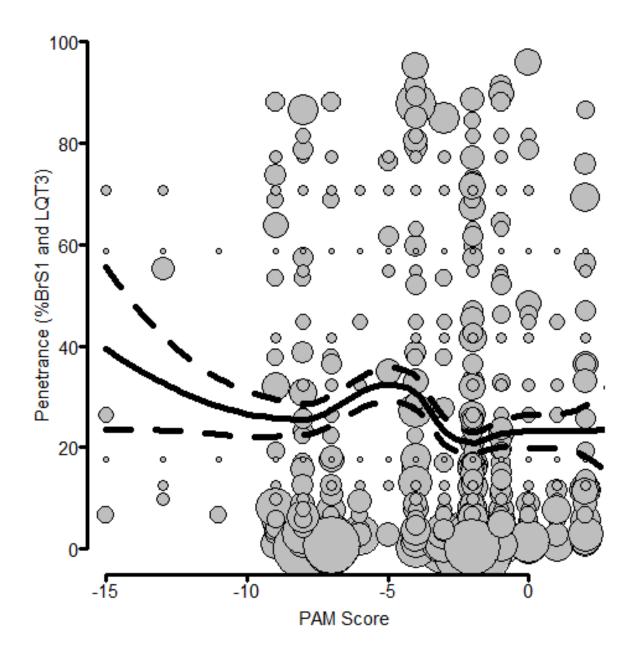


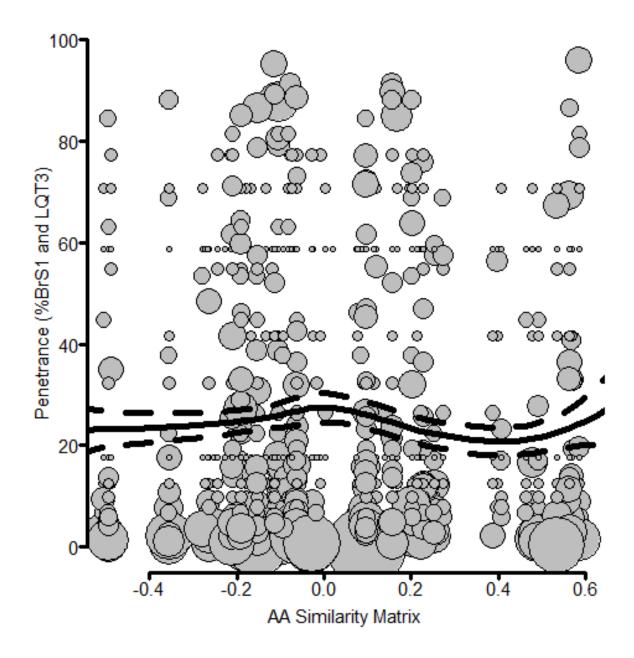








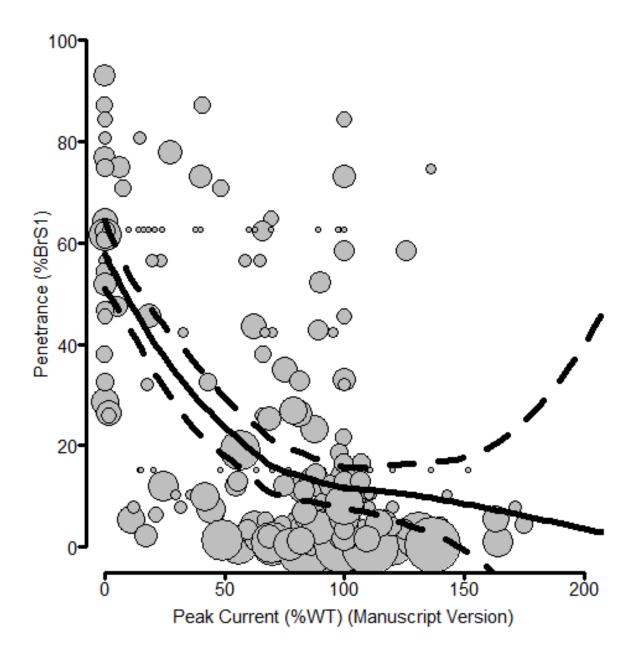


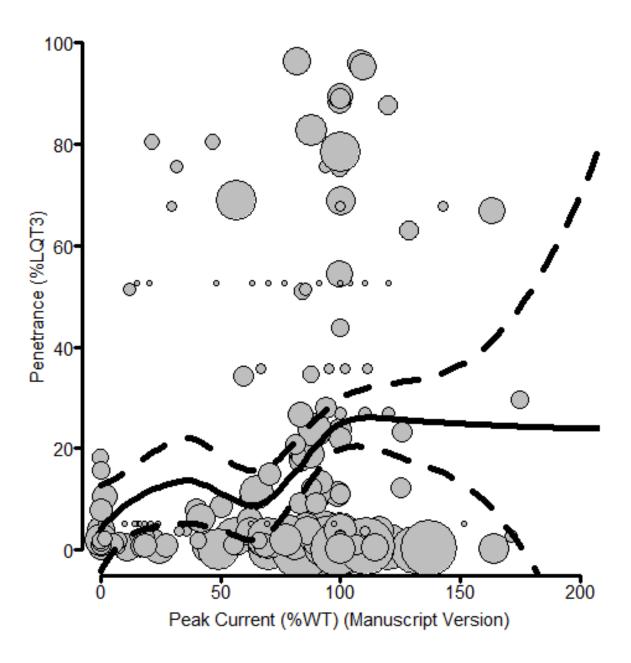


Sensitivity Analysis

Empirical Bayes (used in manuscript)

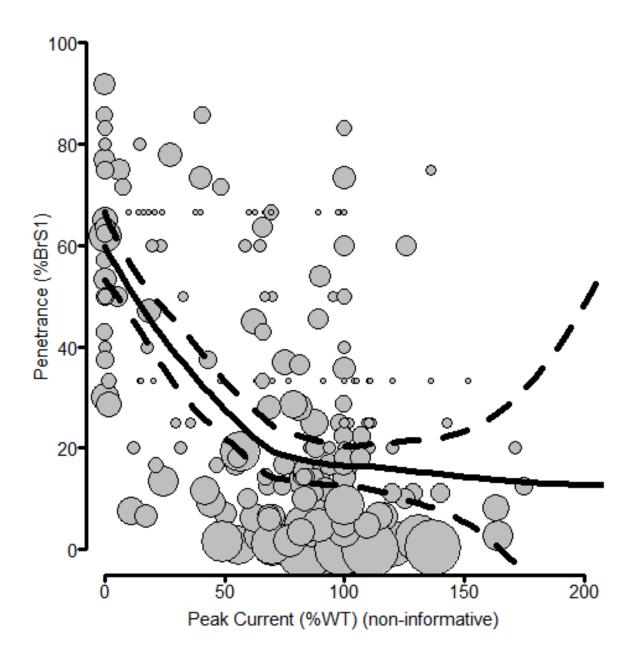
```
# Penetrance calculation used (Manuscript Version)
abrs0=0.32
alqt0=0.11
beta0=1
d$LQT_penetranceBayesian<-(d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0)
d$BrS_penetranceBayesian<-(d$brs1+abrs0)/(d$total_carriers+1+alqt0)
d$all_penetranceBayesian<-(d$brs1+abrs0+d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0+abrs0)</pre>
```

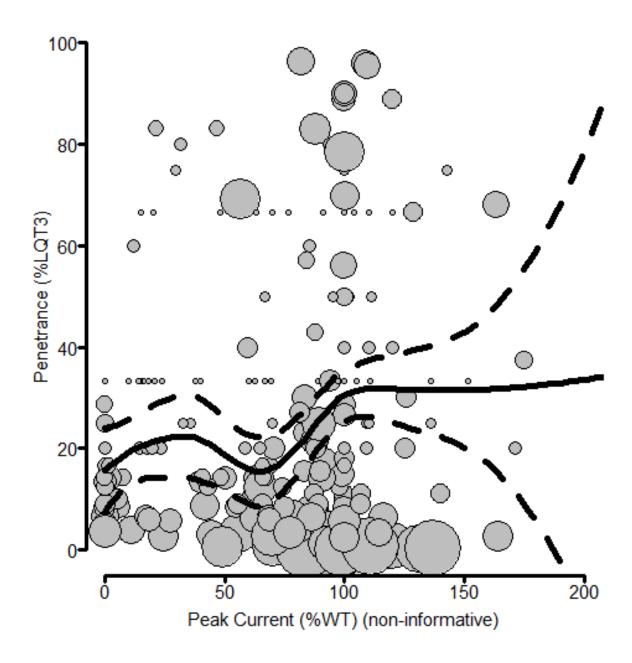




Uninformative Prior

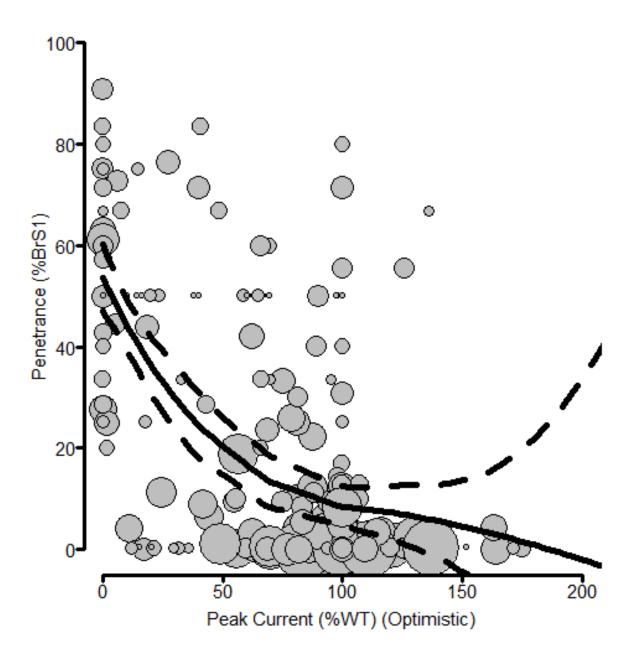
```
# Changing penetrance calculation to uninformative prior
abrs0=1
alqt0=1
beta0=1
d$LQT_penetranceBayesian<-(d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0)
d$BrS_penetranceBayesian<-(d$brs1+abrs0)/(d$total_carriers+1+alqt0)
d$all_penetranceBayesian<-(d$brs1+abrs0+d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0+abrs0)
plt.disease("ipeak","Peak Current (%WT) (non-informative)", "BrS_penetranceBayesian")
plt.disease("ipeak","Peak Current (%WT) (non-informative)", "LQT_penetranceBayesian")</pre>
```

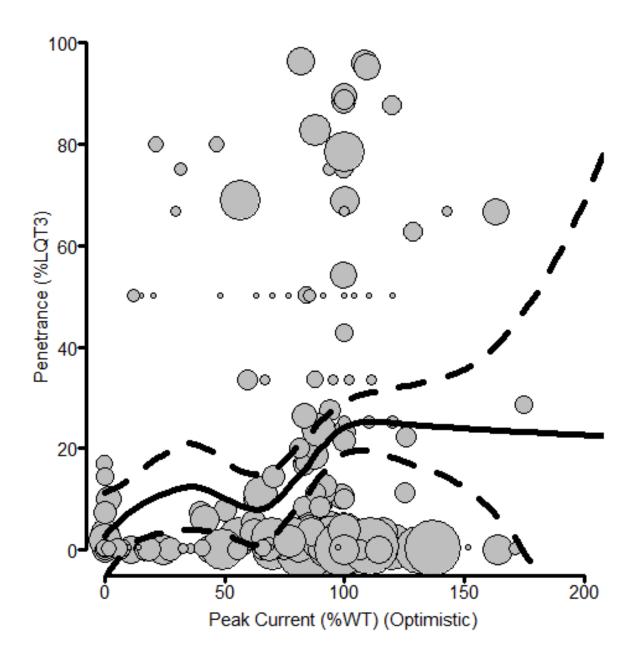




Optimistic Prior

```
# Changing penetrance calculation to optimistic (no affected carriers)
abrs0=0.01
alqt0=0.01
beta0=1
d$LQT_penetranceBayesian<-(d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0)
d$BrS_penetranceBayesian<-(d$brs1+abrs0)/(d$total_carriers+1+alqt0)
d$all_penetranceBayesian<-(d$brs1+abrs0+d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0+abrs0)
plt.disease("ipeak","Peak Current (%WT) (Optimistic)", "BrS_penetranceBayesian")
plt.disease("ipeak","Peak Current (%WT) (Optimistic)", "LQT_penetranceBayesian")</pre>
```





Pessimistic Prior

```
# Changing penetrance calculation to pessimistic (one affected carrier)
abrs0=1
alqt0=1
beta0=0.01
d$LQT_penetranceBayesian<-(d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0)
d$BrS_penetranceBayesian<-(d$brs1+abrs0)/(d$total_carriers+1+alqt0)
d$all_penetranceBayesian<-(d$brs1+abrs0+d$lqt3+alqt0)/(d$total_carriers+beta0+alqt0+abrs0)
plt.disease("ipeak","Peak Current (%WT) (Pessimistic)", "BrS_penetranceBayesian")
plt.disease("ipeak","Peak Current (%WT) (Pessimistic)", "LQT_penetranceBayesian")</pre>
```

