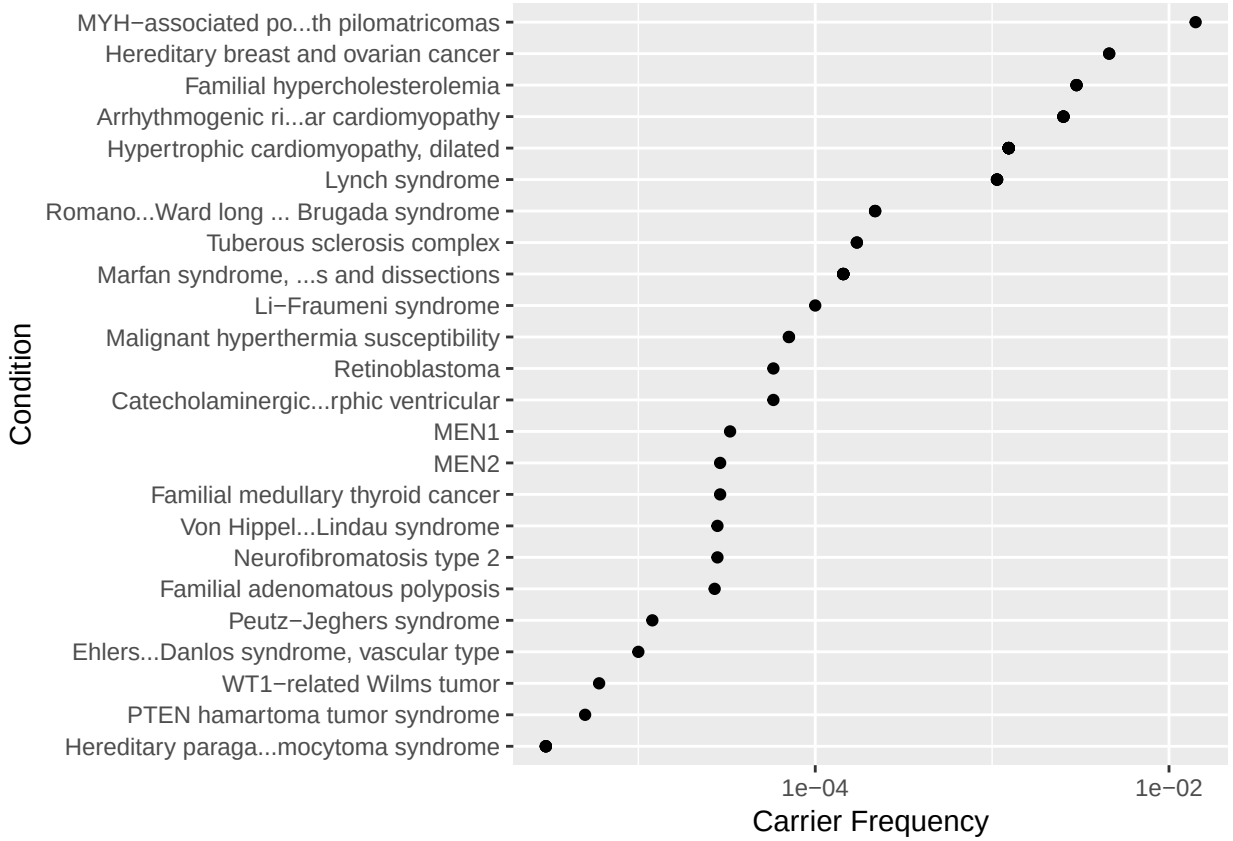
Characterizing the ACMG56 Model

Introduction

This analysis is a summary of a model developed in May 2016. The model was developed using data about typical carrier frequencies for the ACMG56 conditions (Ding, Burnett, and Chesher) plus internal Invitae data regarding the prevalence of SNVs, indels, and CNVs in the pathogenic variant spectrum for each gene.

```
d <- load.model('../data/exome_acmg56_model.cleaned.csv')
d <- calculate.sensitivity(d,0.98,0.93,0.7)
plot.condition.carrier.freqs(d)
```



The above plot shows the carrier frequency data as collected by Ding *etal*. ## References

Ding, Lucy-Enid, Leslie Burnett, and Douglas Chesher"The Impact of Reporting Incidental Findings from Exome and Whole-Genome Sequencing: Predicted Frequencies Based on Modeling." *Gm* 17 (3). Pathology North, New South Wales Health Pathology, Royal North Shore Hospital, Sydney, Australia.: 197–204. doi:10.1038/gim.2014.94.