

pRCC germline

Catogories	Filters
Lower bound	
-ClinVAR (5.5% in 1KG)	Pathogenic evidence (Curated for cancer?) Filter based on MAF? Classic mutations
Best estimation	Lower bound + High impacts
-High impact (1.2% in 1KG)	HIGH impact (stop loss/gain etc.) TSL 1 MAF < 0.5% (in all superpop> in both ExAC and 1KG?)
Upper bound	Best estimation + High missense mut.
- Missense (3-5%? in 1KG)	Missense mutations with high PolyPhen (0.9?)/SIFT TSL 1-3 MAF cut-offs

“Powerful” Ideas for Candisp2

- Masking SNPs known to confounding factors
- Limit searching scope:
 - Focus on mutations shown significant different MAFs in two races
 - Filtering by prior knowledge (gene set)
 - Filtering by functional impacts
- Paired patients in our patients recruitment