



Population_genetics.csv

Gene	Res	WT	Alt	multiallele	C	AF
RAG1	3	Ala	NA	NA	1	0
RAG1	4	Ser	NA	NA	1	0
RAG1	5	Phe	Leu	NA	0	7.96e-6
RAG1	6	Pro	Ser	NA	0	3.98e-6
RAG1	7	Pro	NA	NA	1	0

Percentage_of_mutated_variatns.csv

Gene	Status	value	Total	Percentage
RAG1	Unmutated	584	1043	56
RAG1	Mutated	459	1043	44
RAG2	Unmutated	295	527	56
RAG2	Mutated	232	527	44

Ratio_of_mutation.csv

Gene	Amino	Mutant	WT	Rate_mutated
RAG1	Ala	28	40	0.411764706

MRF.csv

Gene	Res	WT	Alt	multiallele	C	MRF
RAG1	2	Ala	NA	NA	1	0
RAG1	3	Ala	NA	NA	1	0
RAG1	4	Ser	NA	NA	1	0.041
RAG1	5	Phe	Leu	NA	0	0.013
RAG1	6	Pro	Ser	NA	0	0.028
RAG1	7	Pro	NA	NA	1	0.028

Disease database
Clinical RAG
deficiency

Disease database
HMGD

Pathogenicity
prediction
tool
(PHRED-CADD)

Functional
assay
data

Protein
structure

Rare PID
cohort
genomes

Visualisation
thresholds

Fig 1 & E1
MRF & conservation

Fig 4
False scores in core
regions rescued
by conservation

Fig 2
MRF vesus
allele freq

Table E2
Clinical relevance
of top candidates

Fig 7
PHRED-scaled CADD score
versus MRF score
against HGMD data

Fig 6
Mutation likelihood
versus pathogenicity
prediction

Fig 3 & E2
MRF score
categories &
variants assayed
to date

Table 1
MRF, Assayed, Recombination activity (%)
0.03, I56T, 3.5+/-0.2
0.14, G99S, 113.2+/-3.7

Fig E3
MRF mapped
on structure

Fig 5
MRF in RAG deficiency
versus unrelated PIDs