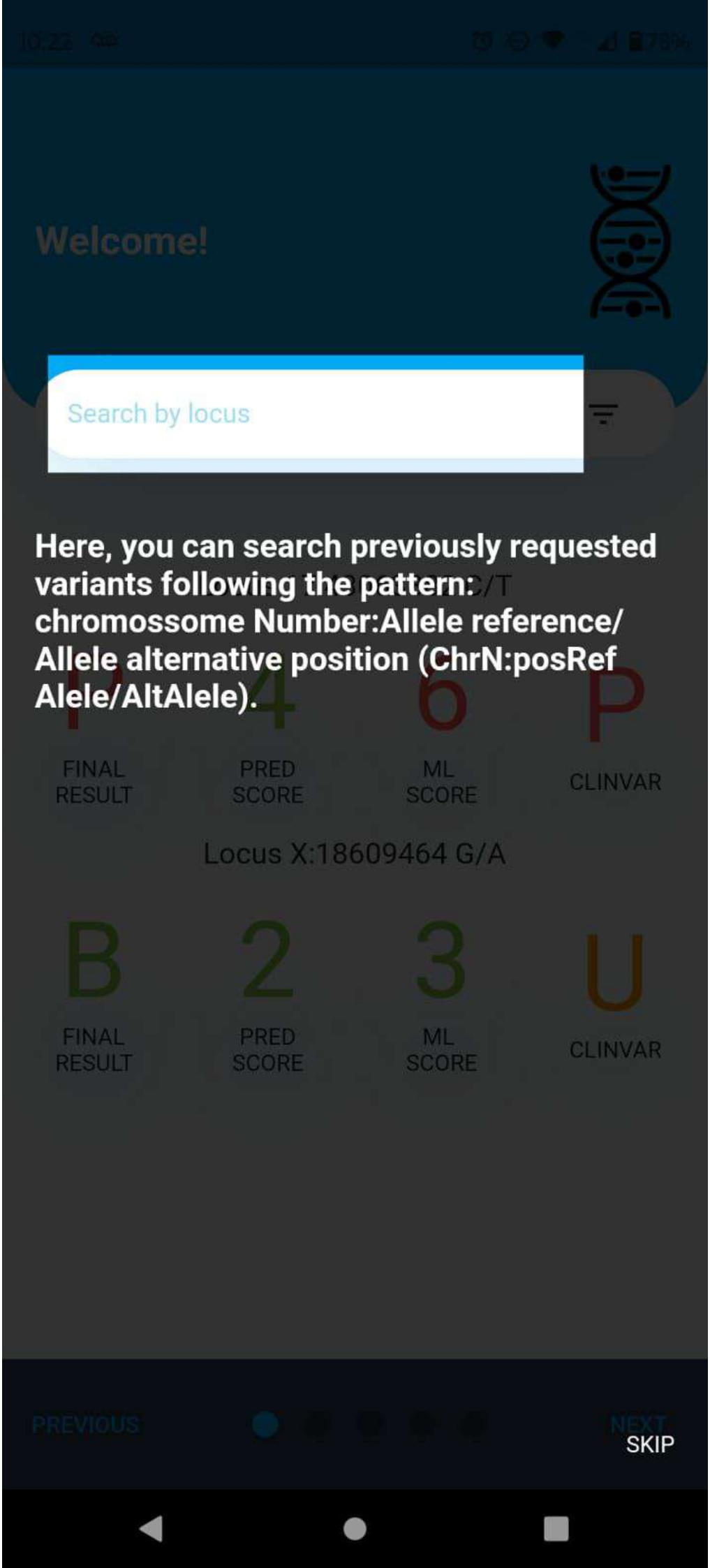
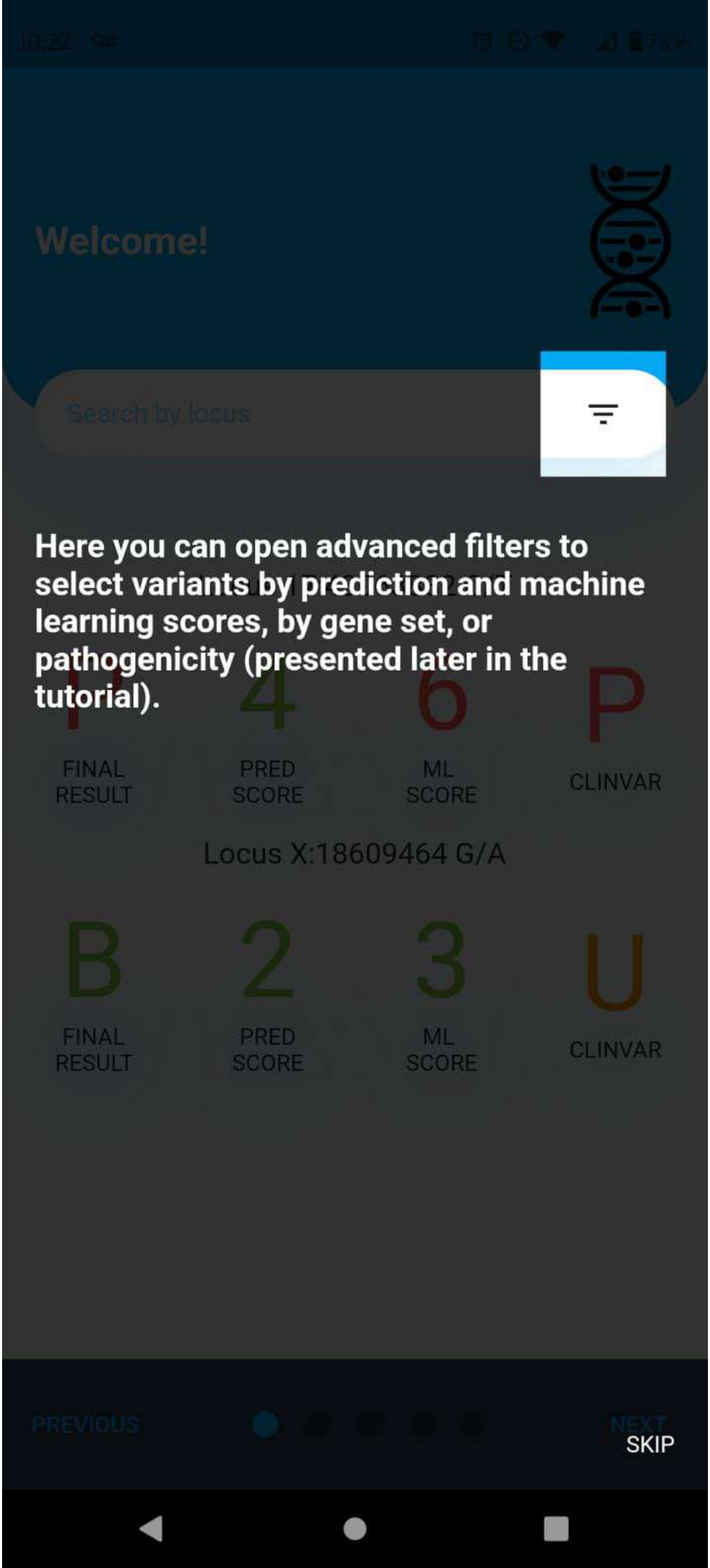
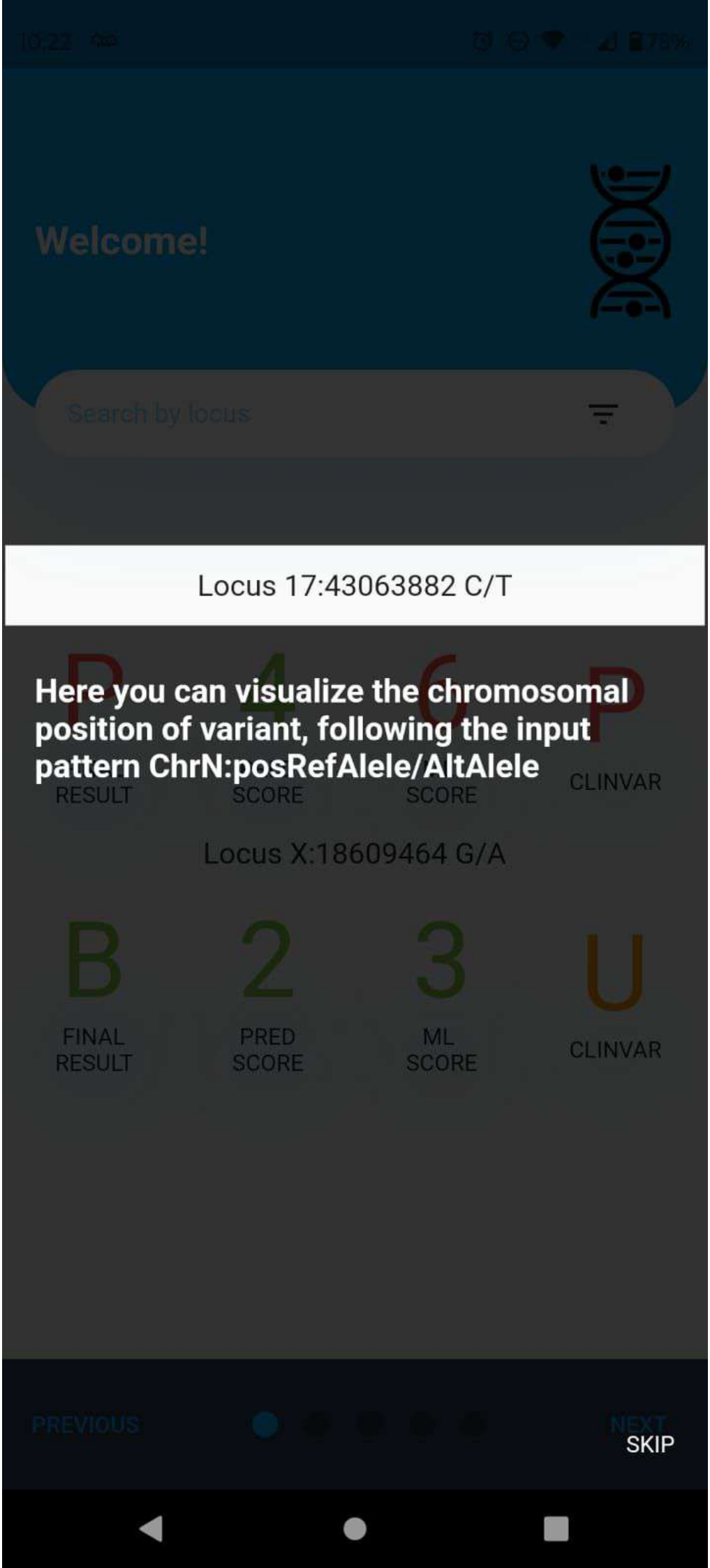
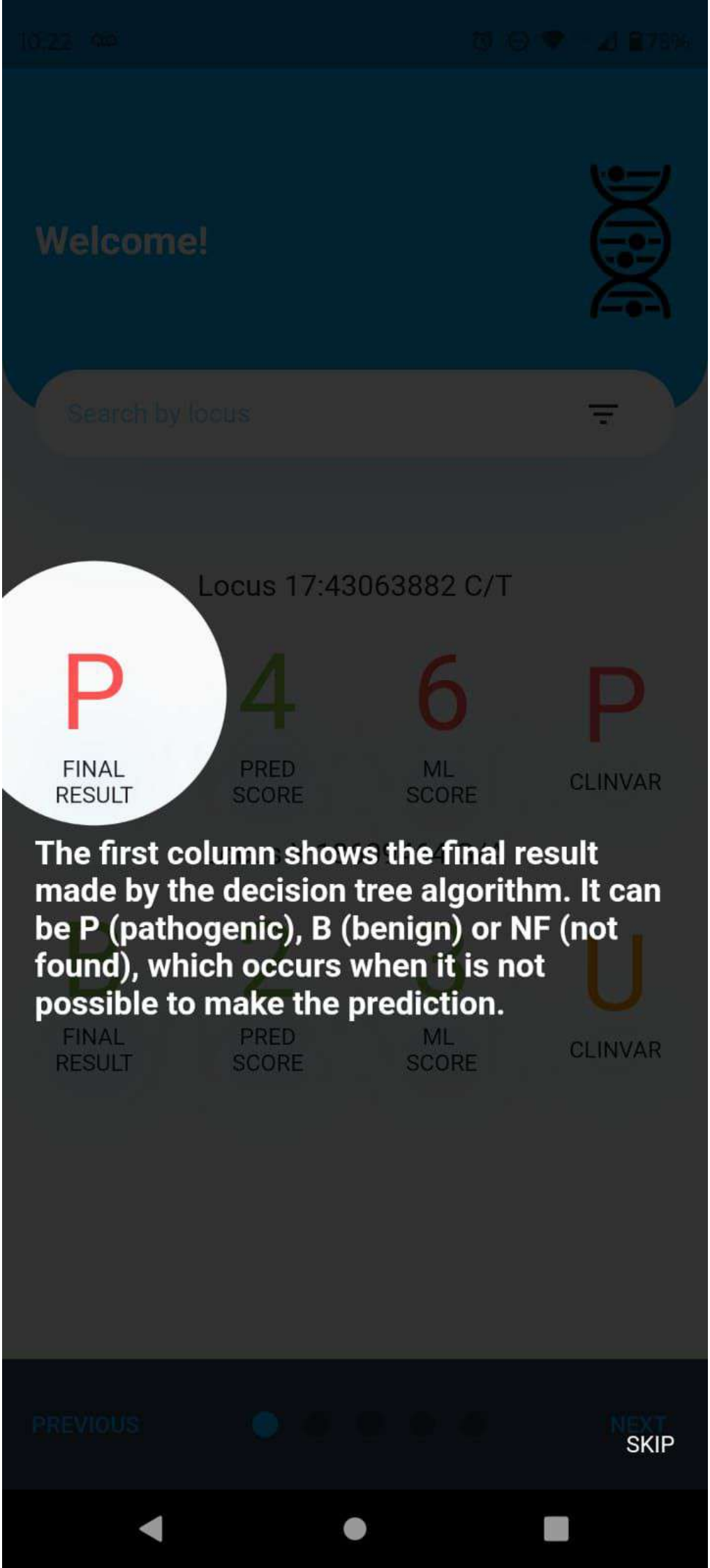
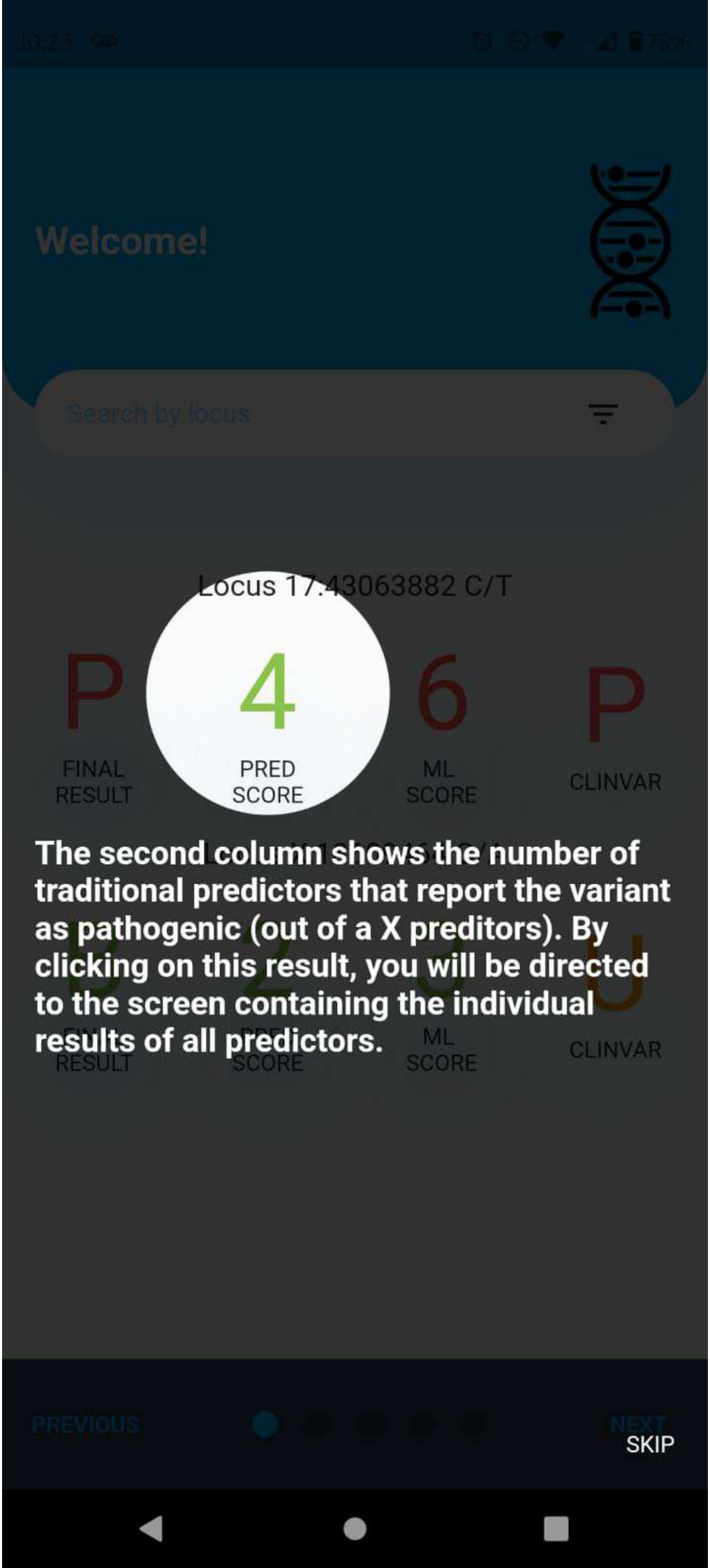
10:20
76%
Welcome!
Search by locus
Locus 17:43063882 C/T
P
FINAL RESULT
4
PRED SCORE
6
ML SCORE
P
CLINVAR
Locus X:18609464 G/A
B
FINAL RESULT
2
PRED SCORE
3
ML SCORE
U
CLINVAR
PREVIOUS
NEXT

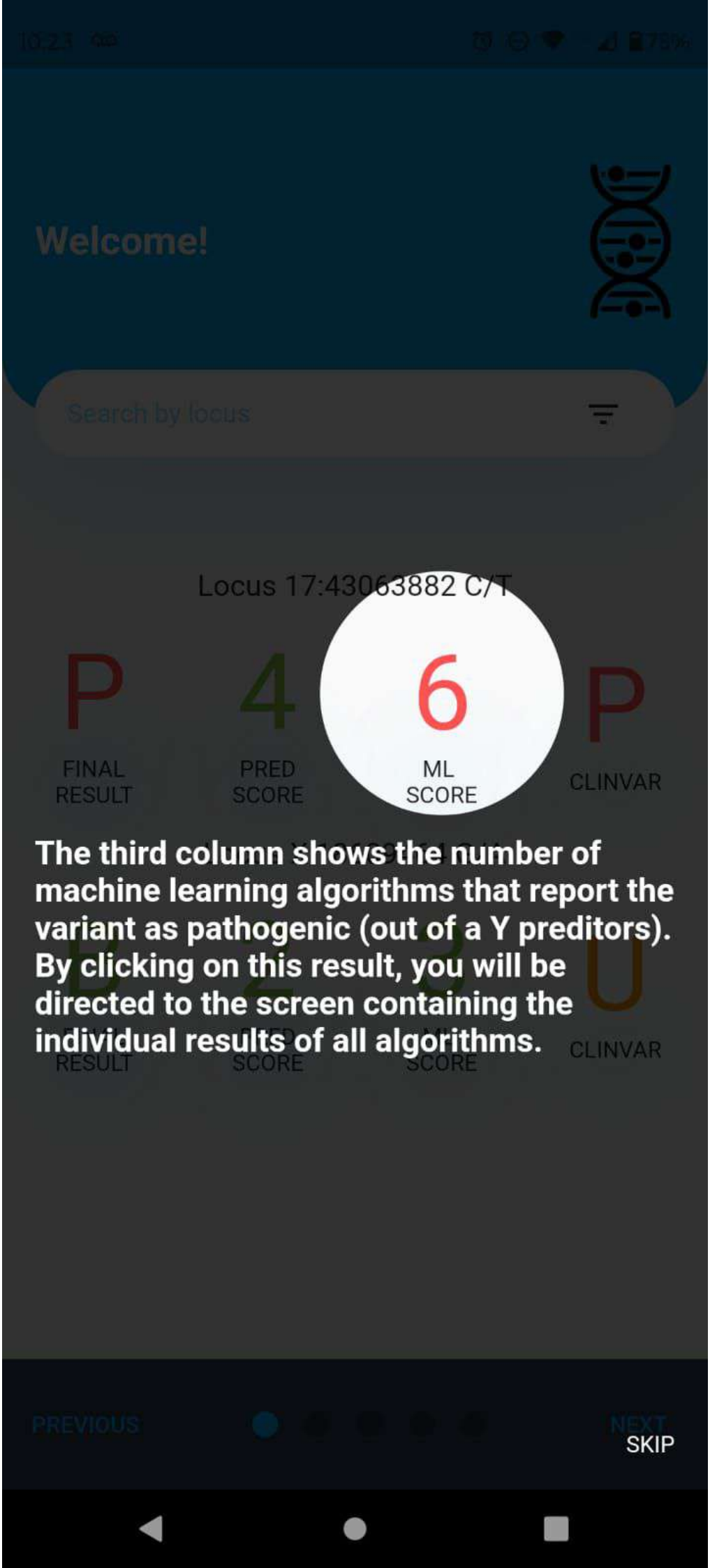

Welcome!
Search by locus
Here, you can search previously requested variants following the pattern: chromossome Number:Allele reference/ Allele alternative position (ChrN:posRef Alele/AltAlele).
P
4
6
P
FINAL RESULT
PRED SCORE
ML SCORE
CLINVAR
Locus X:18609464 G/A
B
2
3
U
FINAL RESULT
PRED SCORE
ML SCORE
CLINVAR
PREVIOUS
NEXT
SKIP

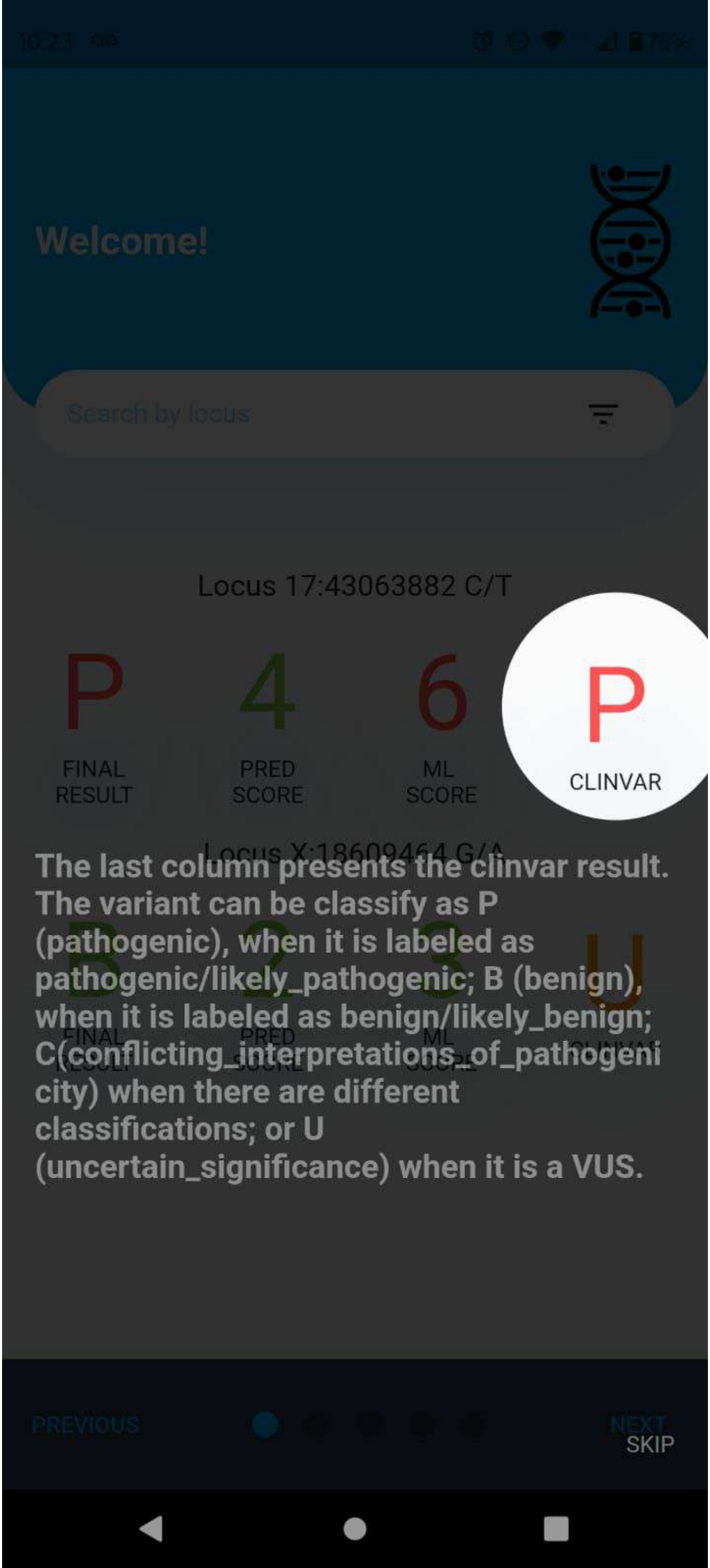

Welcome!
Search by locus
Here you can open advanced filters to select variants by prediction and machine learning scores, by gene set, or pathogenicity (presented later in the tutorial).
P
4
6
P
FINAL RESULT
PRED SCORE
ML SCORE
CLINVAR
Locus X:18609464 G/A
B
2
3
U
FINAL RESULT
PRED SCORE
ML SCORE
CLINVAR
PREVIOUS
NEXT
SKIP

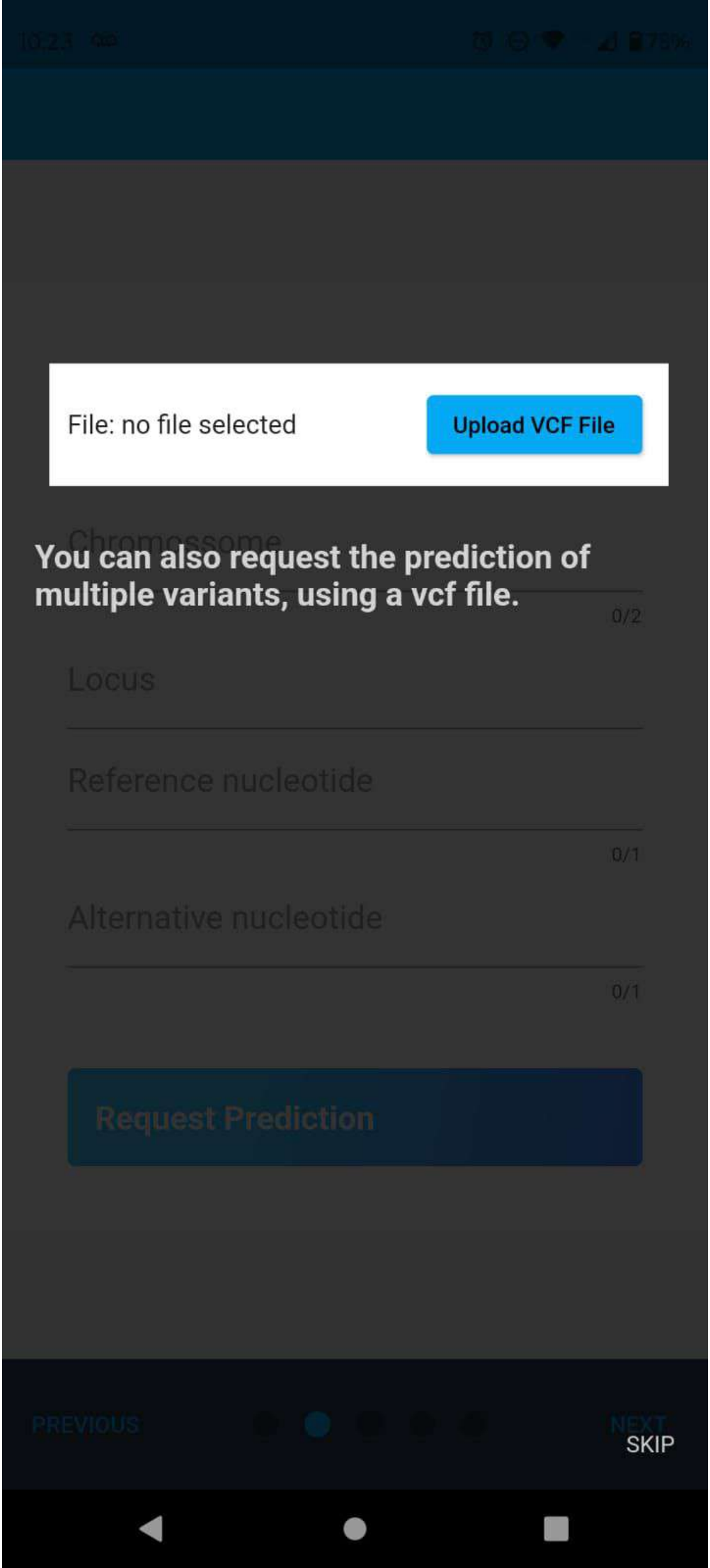

Welcome!
Search by locus
Locus 17:43063882 C/T
Here you can visualize the chromosomal position of variant, following the input pattern ChrN:posRefAlele/AltAlele
RESULT
SCORE
SCORE
CLINVAR
Locus X:18609464 G/A
B
2
3
U
FINAL RESULT
PRED SCORE
ML SCORE
CLINVAR
PREVIOUS
NEXT
SKIP

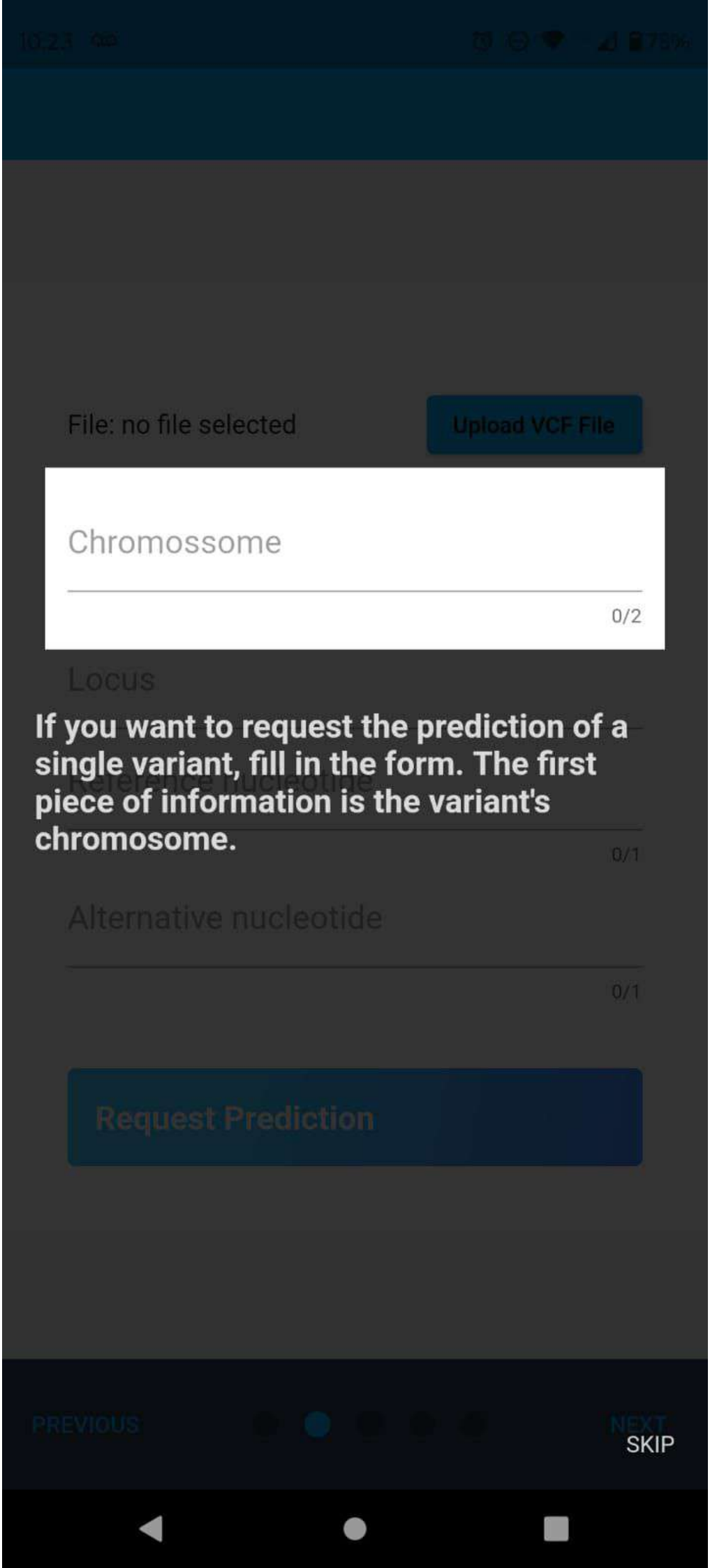

Welcome!
Search by locus
Locus 17:43063882 C/T
P
FINAL RESULT
4
PRED SCORE
6
ML SCORE
P
CLINVAR
The first column shows the final result made by the decision tree algorithm. It can be P (pathogenic), B (benign) or NF (not found), which occurs when it is not possible to make the prediction.
FINAL RESULT
PRED SCORE
ML SCORE
CLINVAR
PREVIOUS
NEXT
SKIP

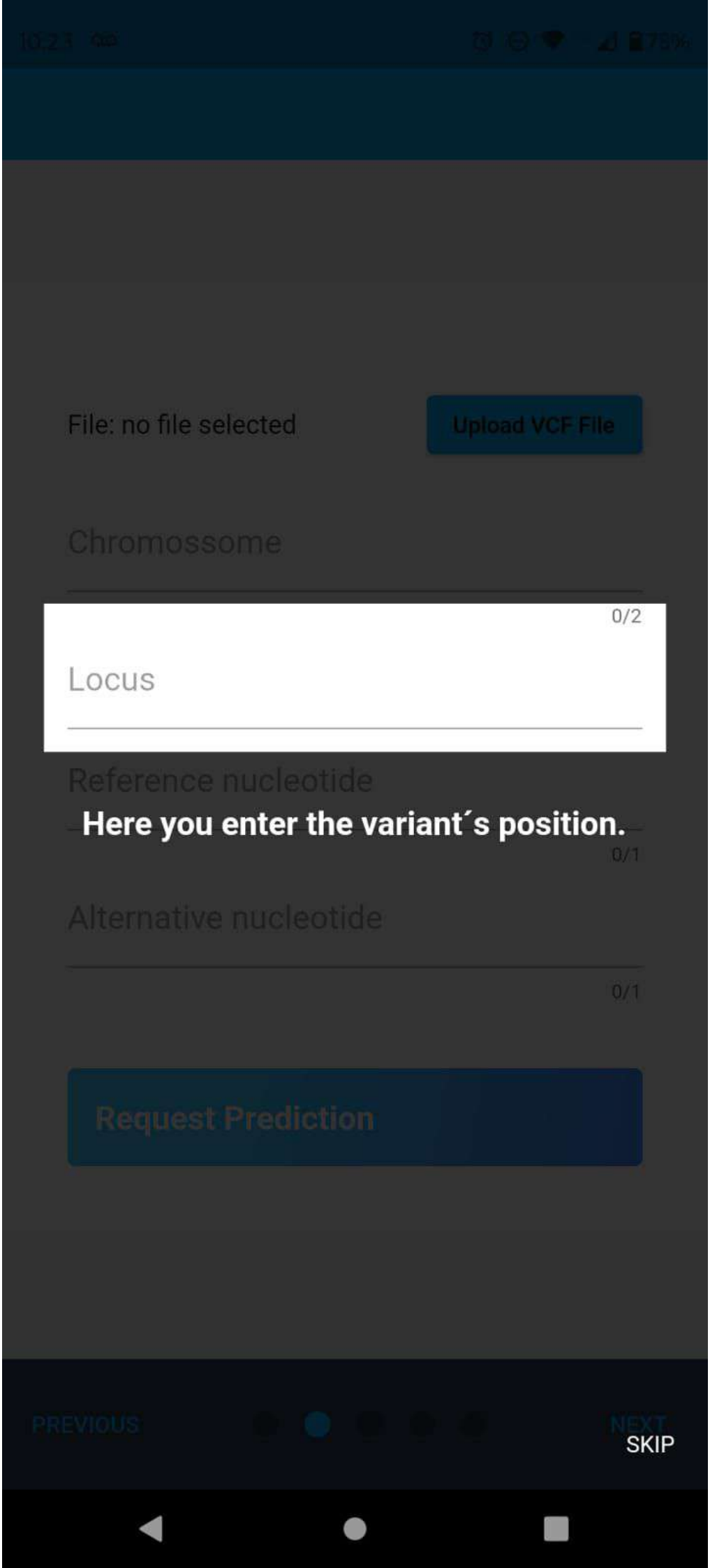

Welcome!
Search by locus
Locus 17:43063882 C/T
P
FINAL RESULT
4
PRED SCORE
6
ML SCORE
P
CLINVAR
The second column shows the number of traditional predictors that report the variant as pathogenic (out of a X preditors). By clicking on this result, you will be directed to the screen containing the individual results of all predictors.
PREVIOUS
NEXT
SKIP

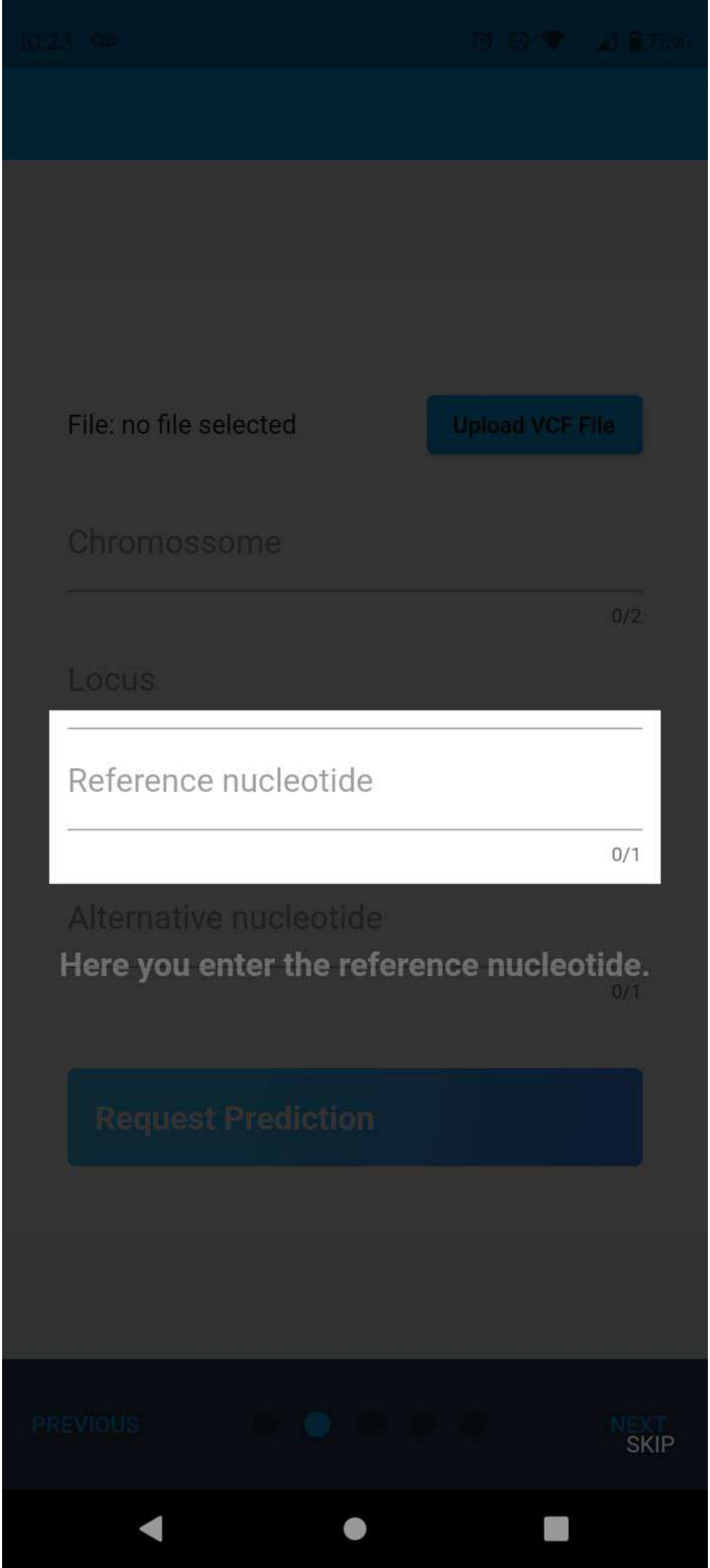

Welcome!
Search by locus
Locus 17:43063882 C/T
P
FINAL RESULT
4
PRED SCORE
6
ML SCORE
P
CLINVAR
The third column shows the number of machine learning algorithms that report the variant as pathogenic (out of a Y preditors). By clicking on this result, you will be directed to the screen containing the individual results of all algorithms.
PREVIOUS
NEXT
SKIP

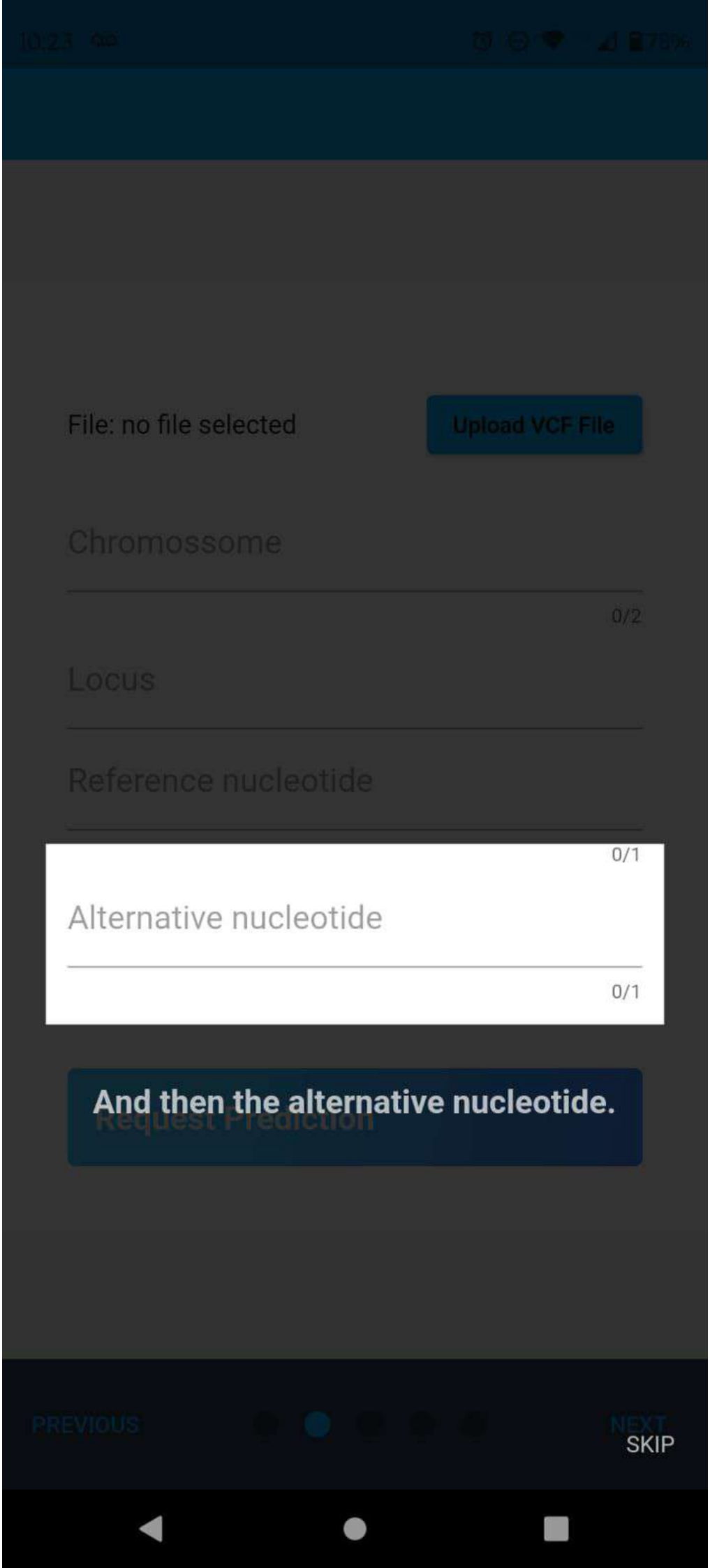

Welcome!
Search by locus
Locus 17:43063882 C/T
P
FINAL RESULT
4
PRED SCORE
6
ML SCORE
P
CLINVAR
The last column presents the clinvar result. The variant can be classify as P (pathogenic), when it is labeled as pathogenic/likely_pathogenic; B (benign), when it is labeled as benign/likely_benign; C(conflicting_interpretations_of_pathogenicity) when there are different classifications; or U (uncertain_significance) when it is a VUS.
PREVIOUS
NEXT
SKIP

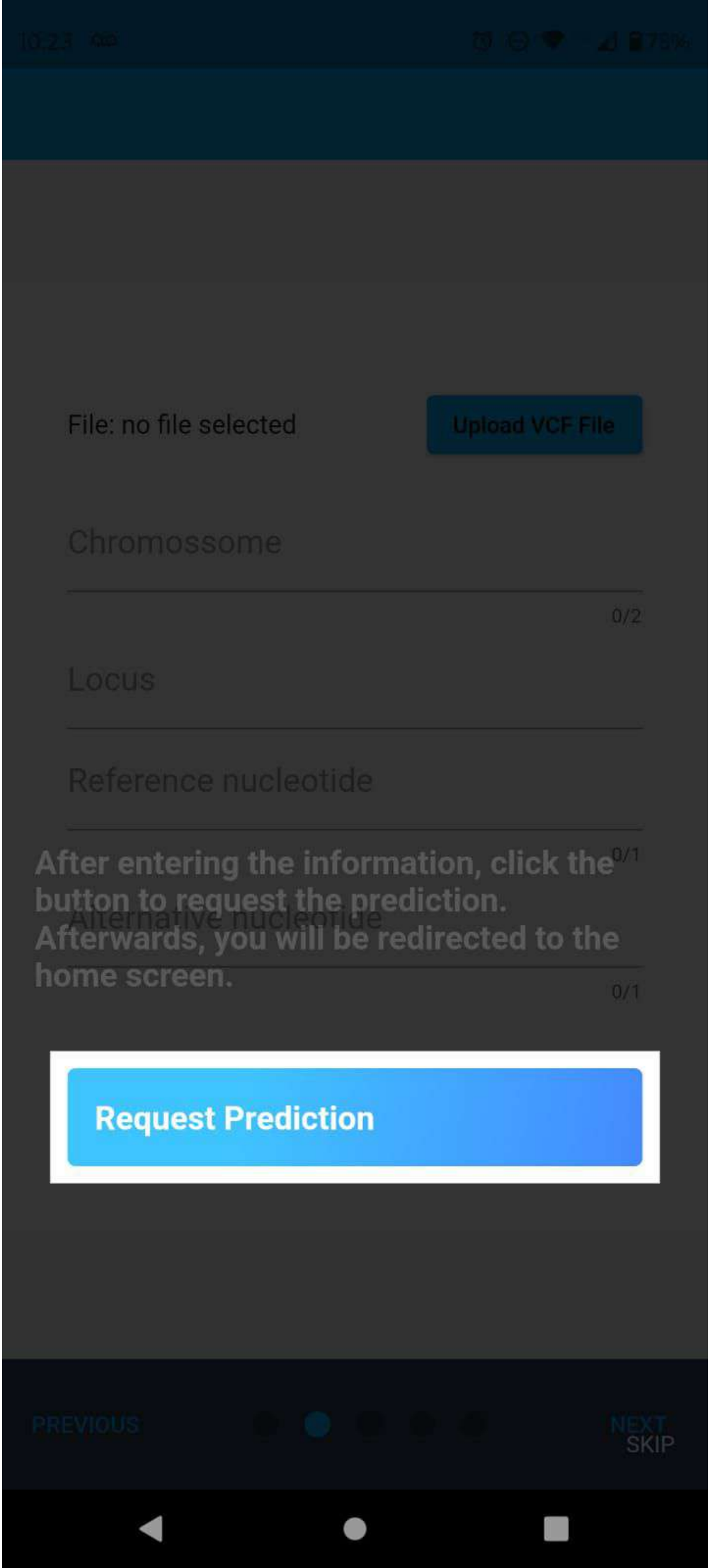

File: no file selected
Upload VCF File
You can also request the prediction of multiple variants, using a vcf file.
Chromosome
0/2
Locus
Reference nucleotide
0/1
Alternative nucleotide
0/1
Request Prediction
PREVIOUS
NEXT
SKIP

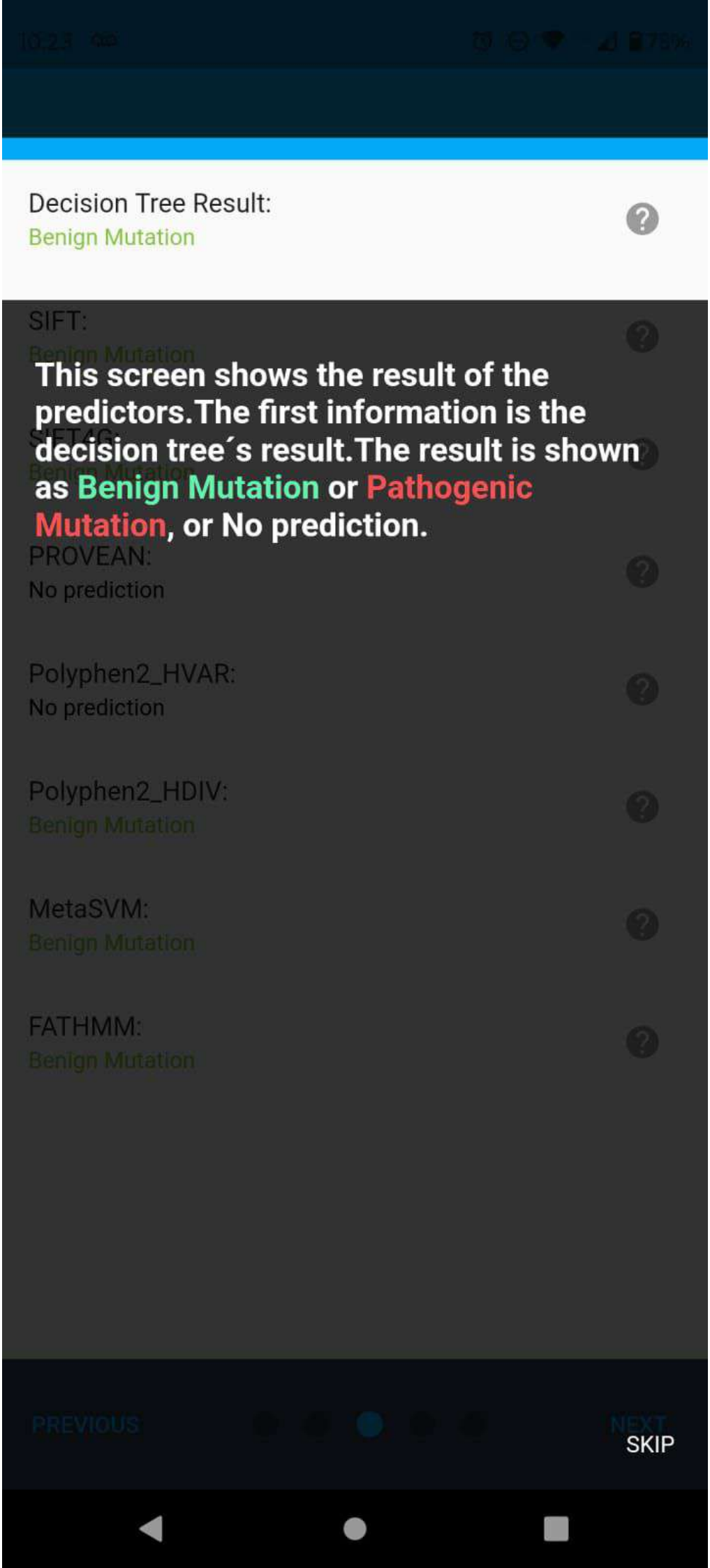

File: no file selected
Upload VCF File
Chromossome
0/2
Locus
If you want to request the prediction of a single variant, fill in the form. The first piece of information is the variant's chromosome.
0/1
Alternative nucleotide
0/1
Request Prediction
PREVIOUS
NEXT
SKIP

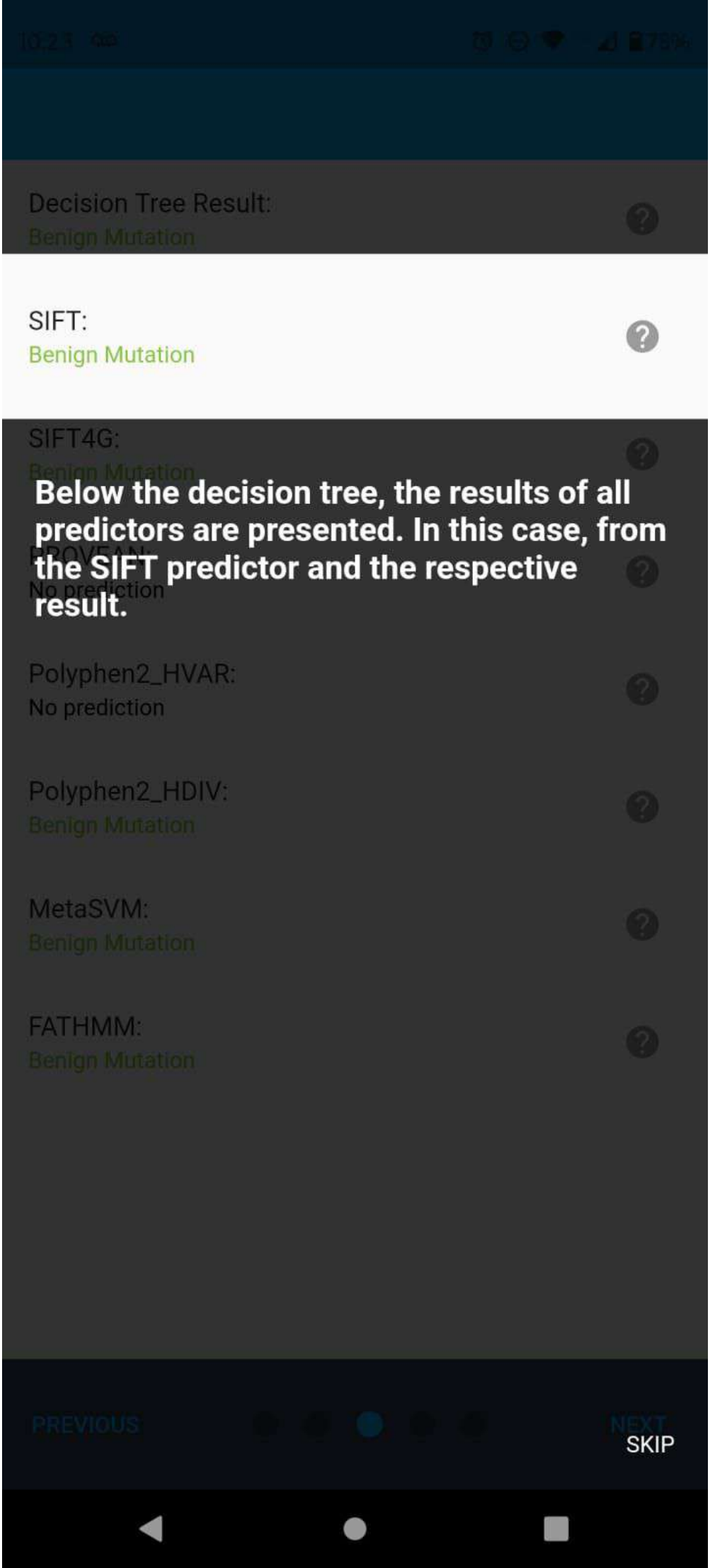

File: no file selected
Upload VCF File
Chromossome
0/2
Locus
Reference nucleotide
Here you enter the variant´s position.
0/1
Alternative nucleotide
0/1
Request Prediction
PREVIOUS
NEXT
SKIP

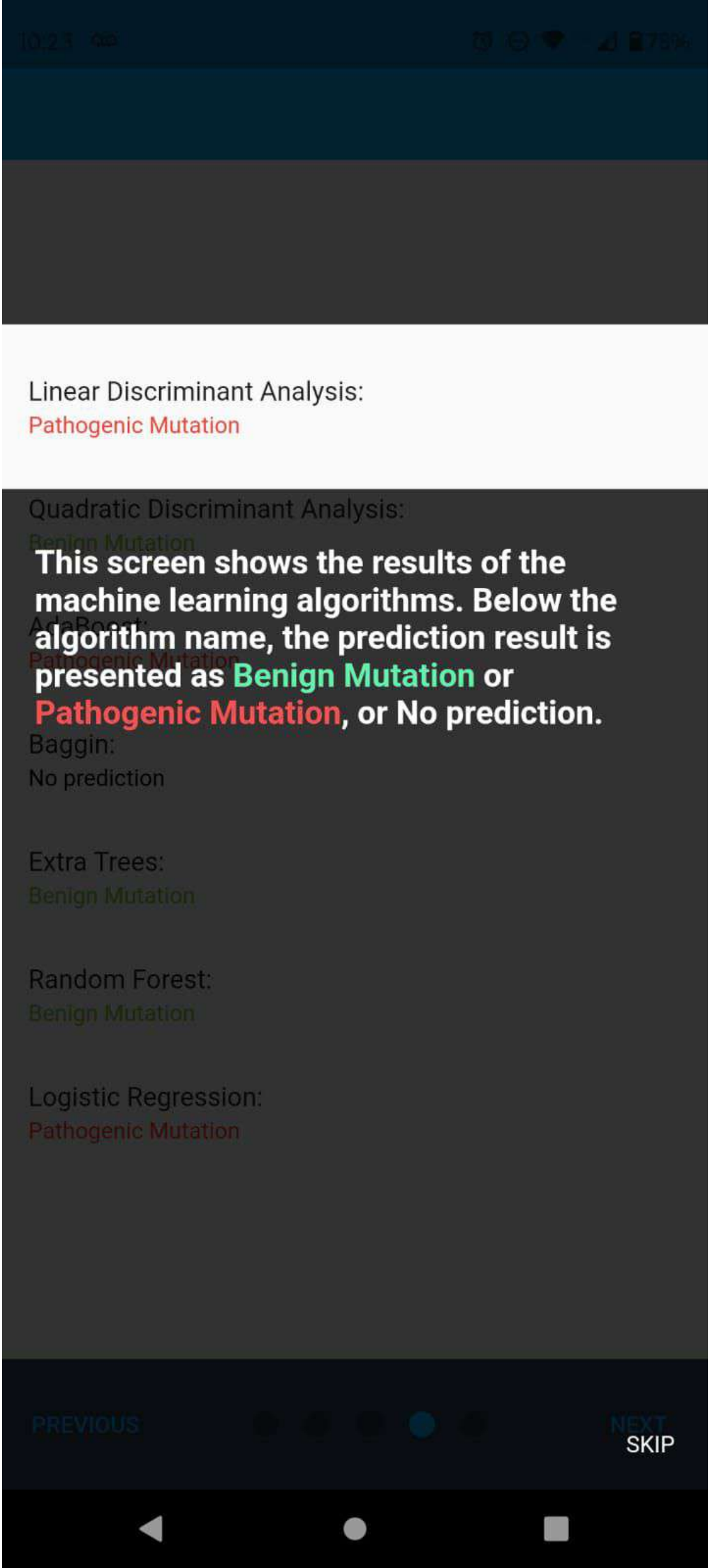

File: no file selected
Upload VCF File
Chromossome
0/2
Locus
Reference nucleotide
0/1
Alternative nucleotide
Here you enter the reference nucleotide.
0/1
Request Prediction
PREVIOUS
NEXT
SKIP

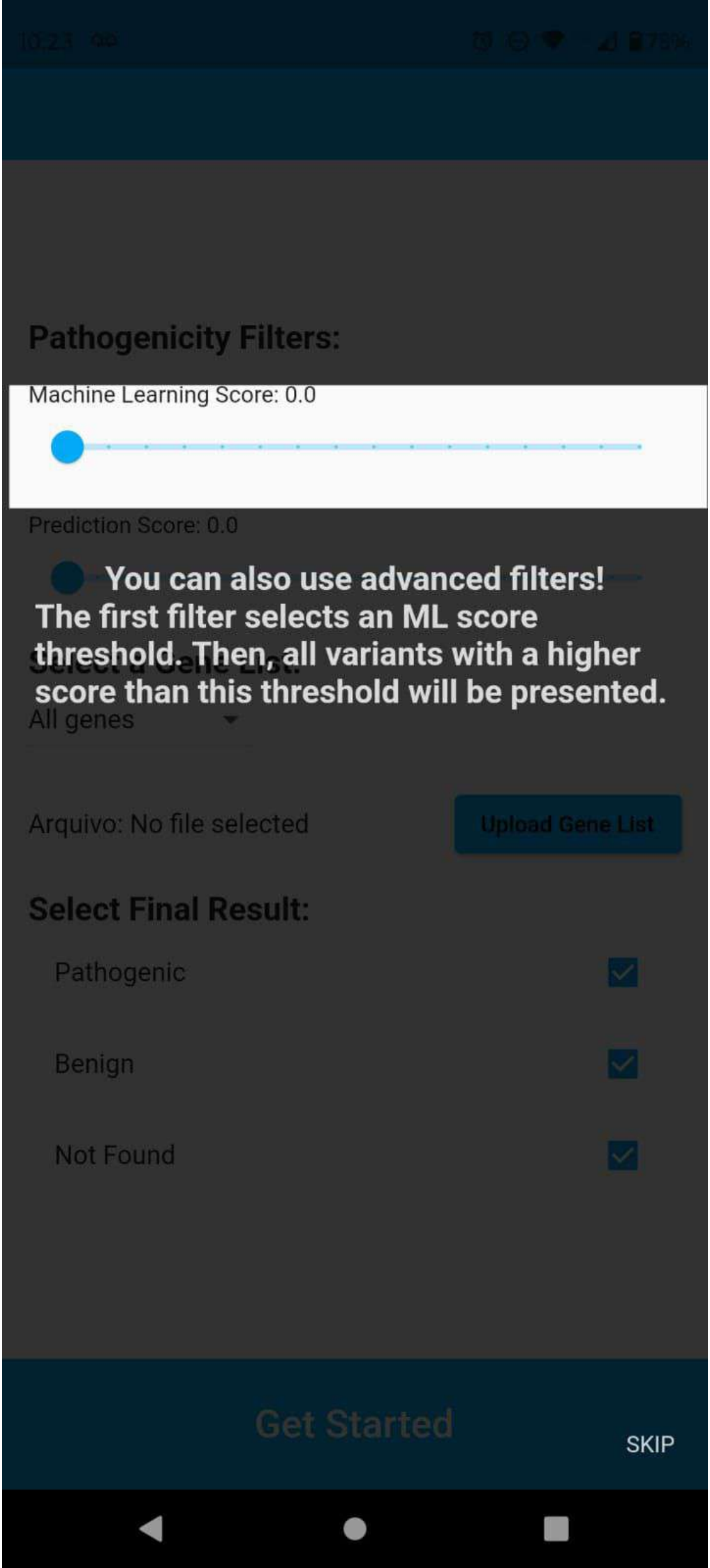

File: no file selected
Upload VCF File
Chromossome
0/2
Locus
Reference nucleotide
0/1
Alternative nucleotide
0/1
And then the alternative nucleotide.
Request Prediction
PREVIOUS
NEXT
SKIP

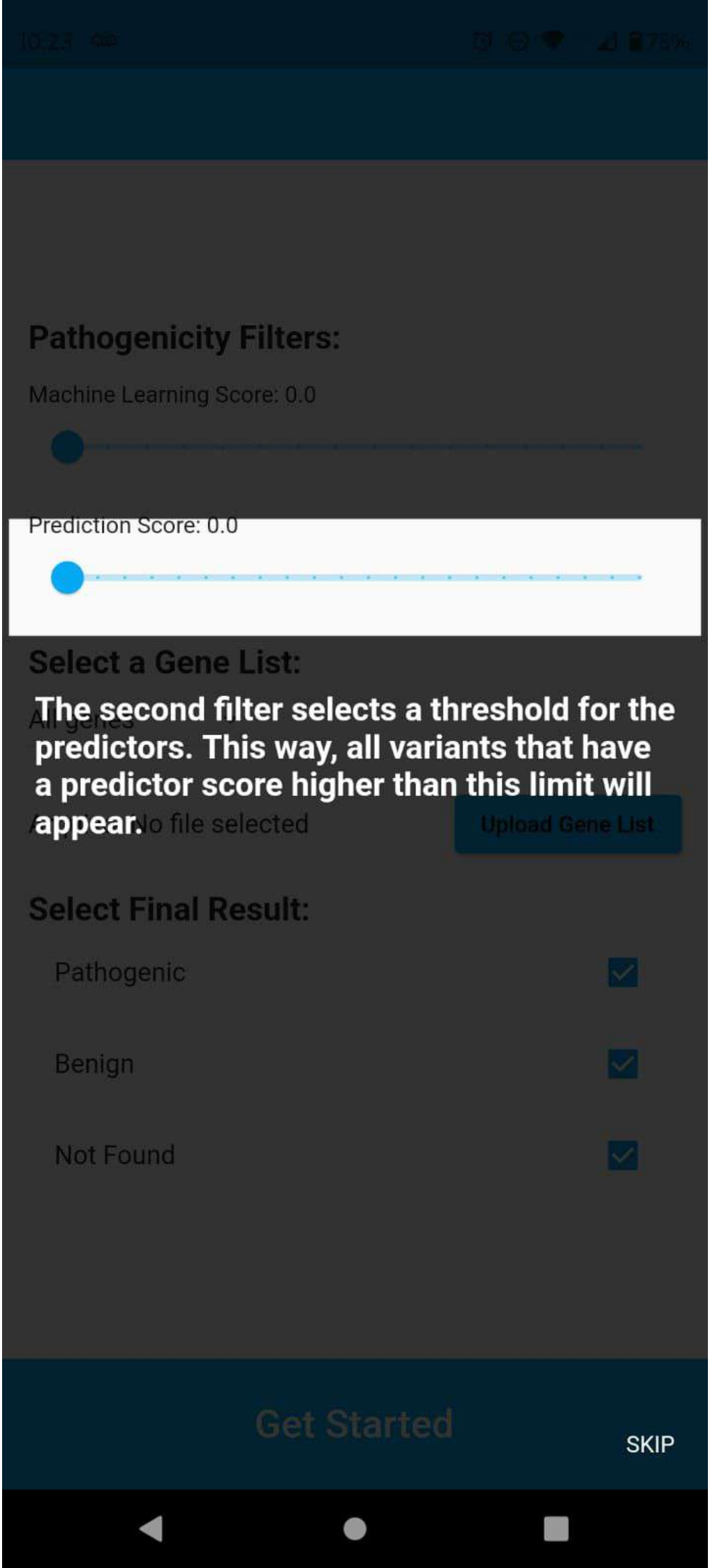

File: no file selected
Upload VCF File
Chromossome
0/2
Locus
Reference nucleotide
0/1
After entering the information, click the button to request the prediction. Afterwards, you will be redirected to the home screen.
Alternative nucleotide
0/1
Request Prediction
PREVIOUS
NEXT
SKIP

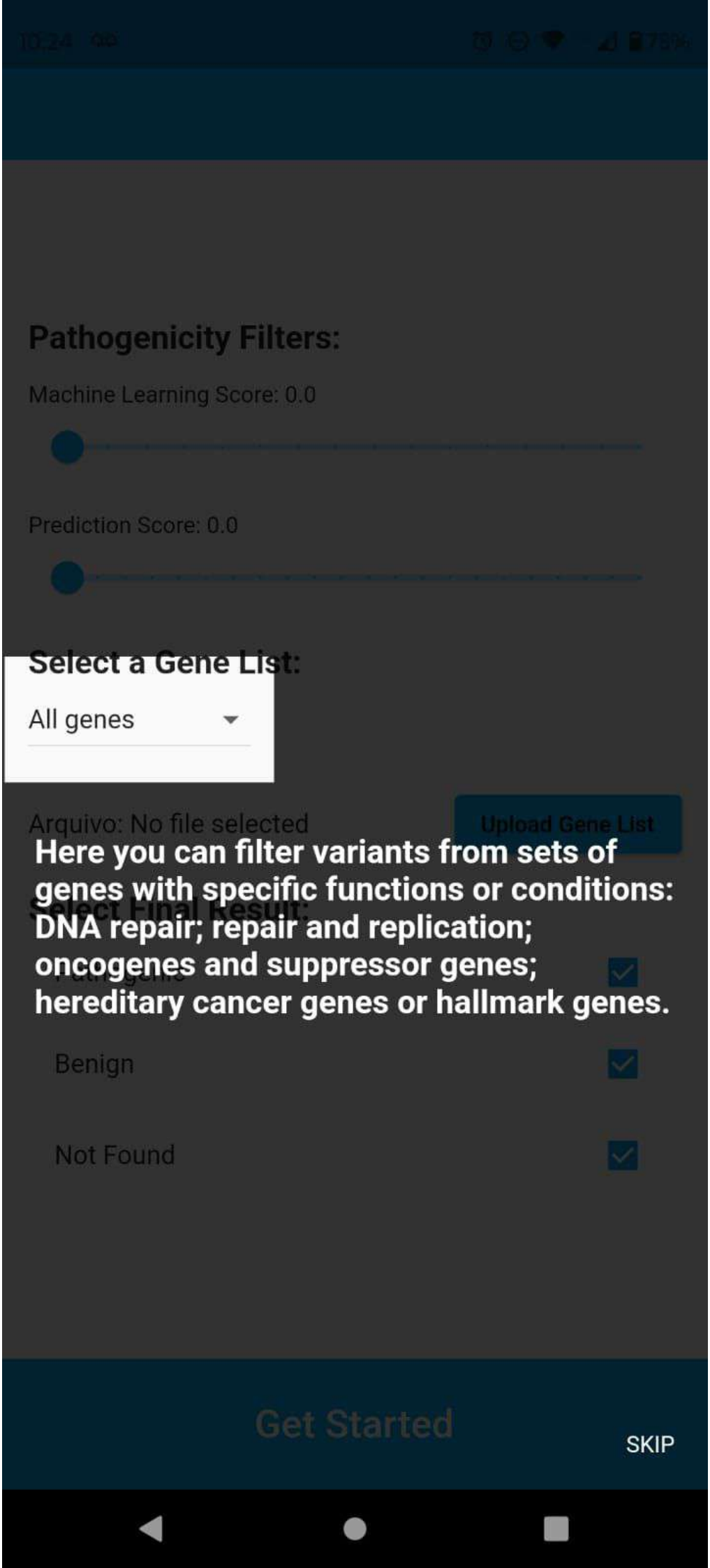

Decision Tree Result:
Benign Mutation
SIFT:
Benign Mutation
SIFT4G:
Benign Mutation
PROVEAN:
No prediction
Polyphen2_HVAR:
No prediction
Polyphen2_HDIV:
Benign Mutation
MetaSVM:
Benign Mutation
FATHMM:
Benign Mutation
This screen shows the result of the predictors.The first information is the decision tree´s result.The result is shown as Benign Mutation or Pathogenic Mutation, or No prediction.
PREVIOUS
NEXT
SKIP

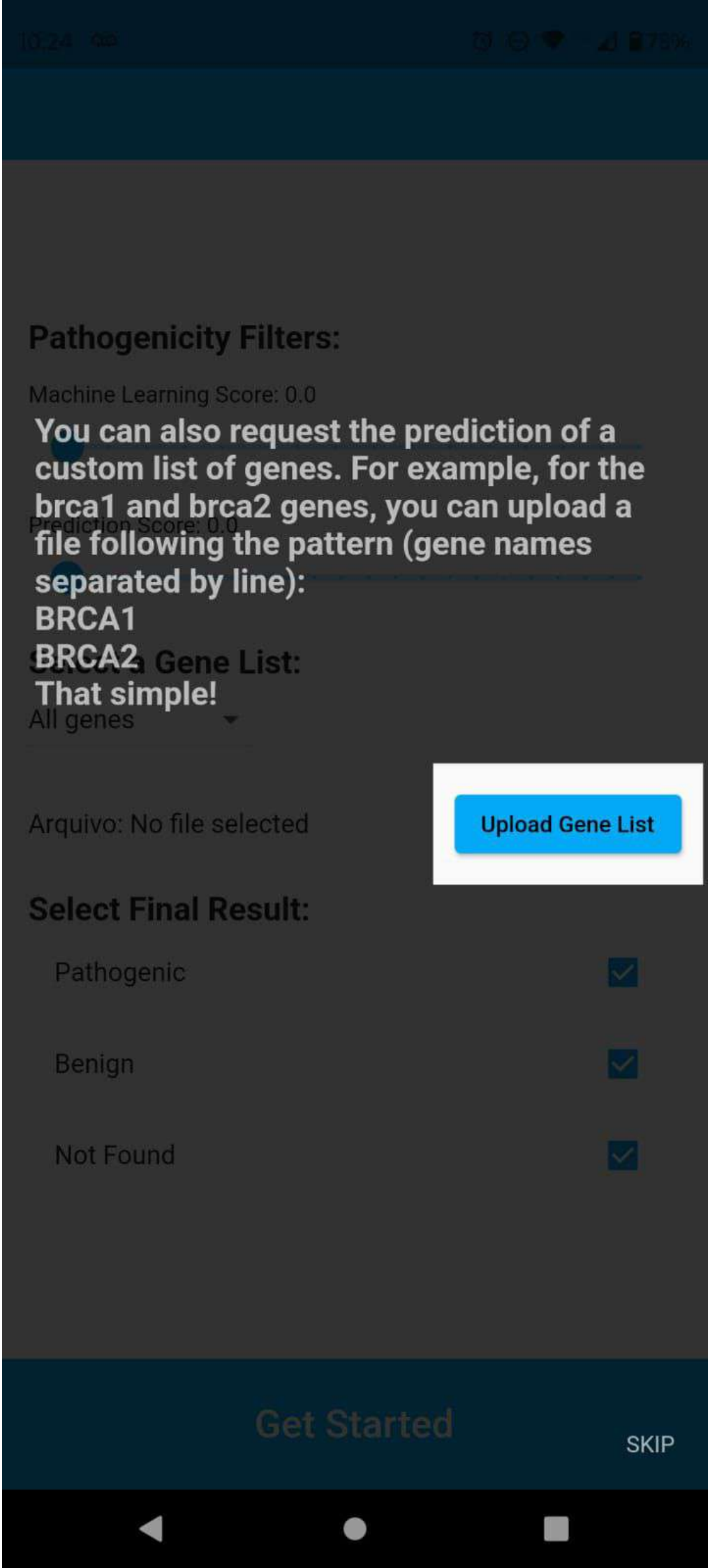

Decision Tree Result:
Benign Mutation
SIFT:
Benign Mutation
SIFT4G:
Benign Mutation
Below the decision tree, the results of all predictors are presented. In this case, from the SIFT predictor and the respective result.
No prediction
Polyphen2_HVAR:
No prediction
Polyphen2_HDIV:
Benign Mutation
MetaSVM:
Benign Mutation
FATHMM:
Benign Mutation
PREVIOUS
NEXT
SKIP

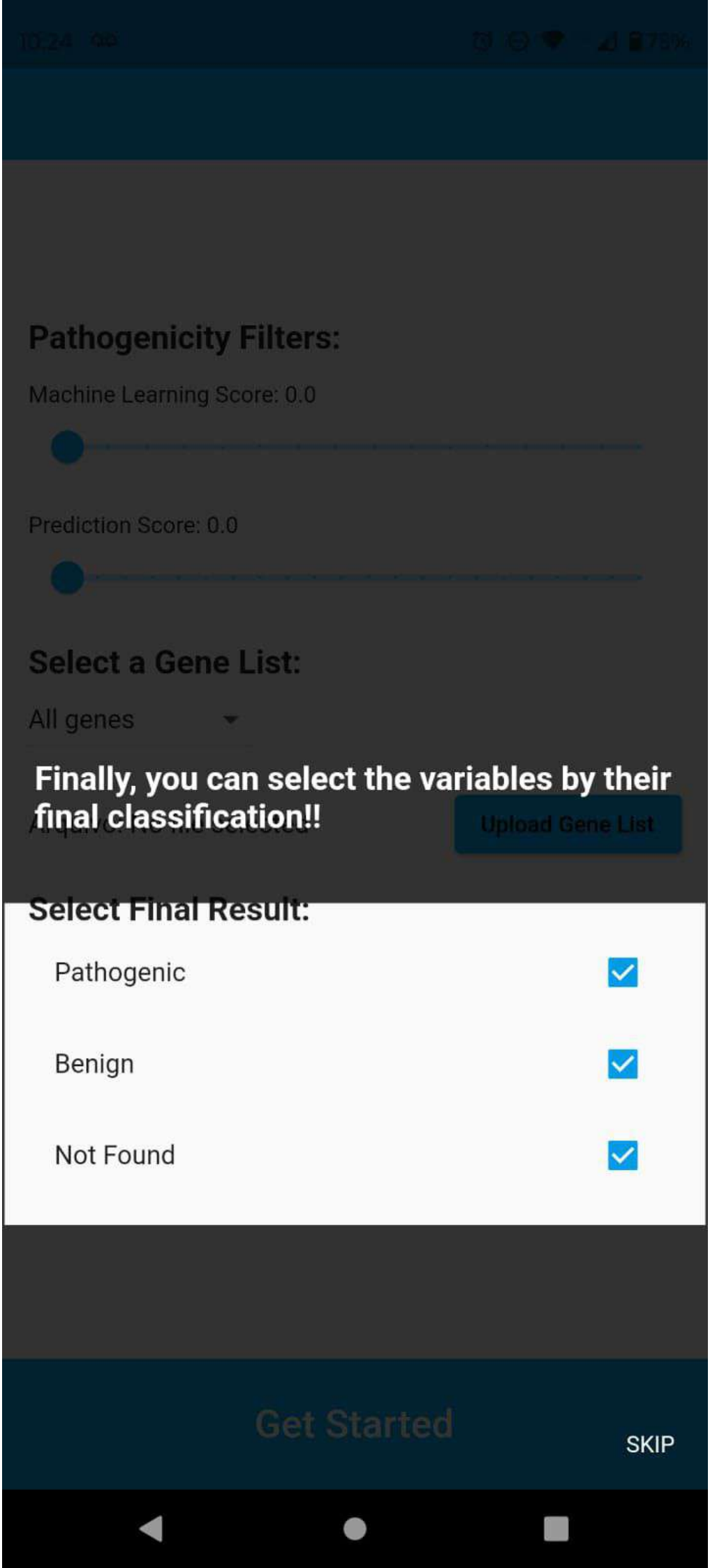

Linear Discriminant Analysis:
Pathogenic Mutation
Quadratic Discriminant Analysis:
Benign Mutation
This screen shows the results of the machine learning algorithms. Below the algorithm name, the prediction result is presented as Benign Mutation or Pathogenic Mutation, or No prediction.
Baggin:
No prediction
Extra Trees:
Benign Mutation
Random Forest:
Benign Mutation
Logistic Regression:
Pathogenic Mutation
PREVIOUS
NEXT
SKIP


Pathogenicity Filters:
Machine Learning Score: 0.0
Prediction Score: 0.0
You can also use advanced filters!
The first filter selects an ML score threshold. Then, all variants with a higher score than this threshold will be presented.
All genes
Arquivo: No file selected
Upload Gene List
Select Final Result:
Pathogenic
Benign
Not Found
Get Started
SKIP


Pathogenicity Filters:
Machine Learning Score: 0.0
Prediction Score: 0.0
Select a Gene List:
The second filter selects a threshold for the predictors. This way, all variants that have a predictor score higher than this limit will appear.
Upload Gene List
Select Final Result:
Pathogenic
Benign
Not Found
Get Started
SKIP


Pathogenicity Filters:
Machine Learning Score: 0.0
Prediction Score: 0.0
Select a Gene List:
All genes
Arquivo: No file selected
Upload Gene List
Here you can filter variants from sets of genes with specific functions or conditions: DNA repair; repair and replication; oncogenes and suppressor genes; hereditary cancer genes or hallmark genes.
Benign
Not Found
Get Started
SKIP


Pathogenicity Filters:
Machine Learning Score: 0.0
You can also request the prediction of a custom list of genes. For example, for the brca1 and brca2 genes, you can upload a file following the pattern (gene names separated by line):
BRCA1
BRCA2
That simple!
Prediction Score: 0.0
Select a Gene List:
All genes
Arquivo: No file selected
Upload Gene List
Select Final Result:
Pathogenic
Benign
Not Found
Get Started
SKIP


Pathogenicity Filters:
Machine Learning Score: 0.0
Prediction Score: 0.0
Select a Gene List:
All genes
Finally, you can select the variables by their final classification!!
Upload Gene List
Select Final Result:
Pathogenic
Benign
Not Found
Get Started
SKIP