# **Final results jSON format structure**

This is the desired Json format after QC and Phenomization for use in the Simulation process.

**submitter{}**

{user\_email, user\_name, user\_team}

**documents[]**

[{document\_name,is\_vcf}]

>> Additional information on the case, which might be provided by the user.

>> Can be a provided vcf file.

**geneList**

[has\_mask, feature\_score, gene\_symbol, combined\_score, syndrome\_name, gestalt\_score, gene\_id, gene\_omim\_id]

>> gene information added after mapping gene to syndromes

>> Can contain extra information from Simulation step : [ cadd\_phred\_score, pheno\_score, boqa\_score, cadd\_raw\_score]

**selected\_syndromes**

[has\_mask, omim\_id, syndrome\_name]

>>Syndrome as selected by the user in the Face2Gene interface.

**detected\_syndromes**

[combined\_score, feature\_score, gestalt\_score, has\_mask, omim\_id:|[], syndrome\_name]

>> Syndromes detected by the Face2Gene algorithm based on the provided phenotypic information as specified in features and the provided image.

**features[]**

>> List of phenotypic features specified using HPO terminology.

**ranks []**

[{feature\_score, value\_pheno, disease-name\_pheno,omim\_id, combined\_score , value\_boqa, disease-name\_boqa, syndrome\_name , gene-id , gestalt\_score , confirmed , gene-symbol}]

>> Ranks generated after Phenomization

**algo\_deploy\_version**

Algorithm version used in the generation of the jSON File. This might affect the detected syndromes and masks. This might change if the dumps are updated for single cases.

**case\_id**

Unique id identifying the case on Face2Gene.

**vcf []**

Name of Vcf file when available

**processing []**

List of the scripts used to produce output file and background\_sample

**genomic\_entries []**

[<entry\_filename>]

References the filename of the genomic entry for the case. There might be more than one genomic entry per case.

**genomicData[]**

[ Mutations {result, Build, HGVS-code, additional info, Inheritance Mode}

, Test Information {Gene Name, Genotype, Notation, Mutation Type, Molecular Test}

>> information extracted from genomic\_entries in preprocessing step