Deep Phenotype for Deep Learning (DPDL)

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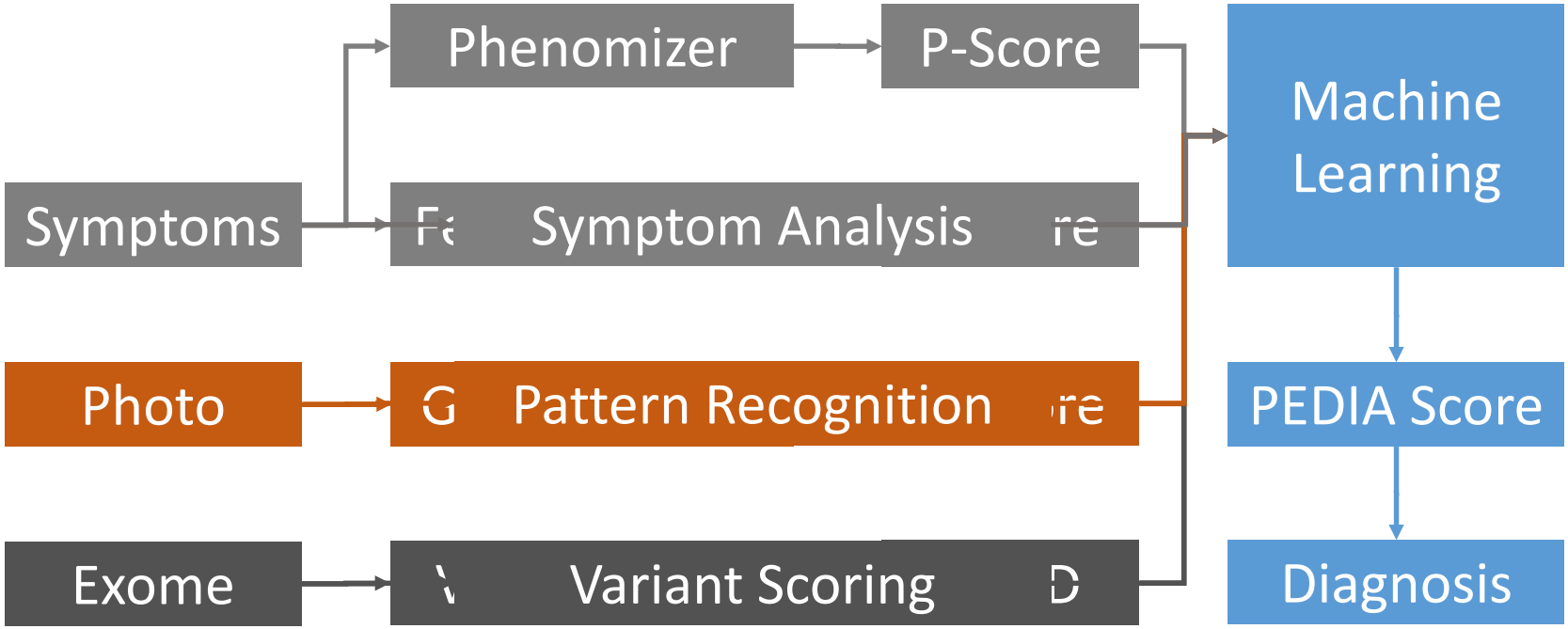
IGSB

Introduction

- A database which integrates phenotype-genotype information for clustering analysis in the phenotype space
- An analysis platform which performs genetic variants prioritization by PEDIA scoring approach.

PEDIA

- Prioritization of Exome Data with Image Analysis



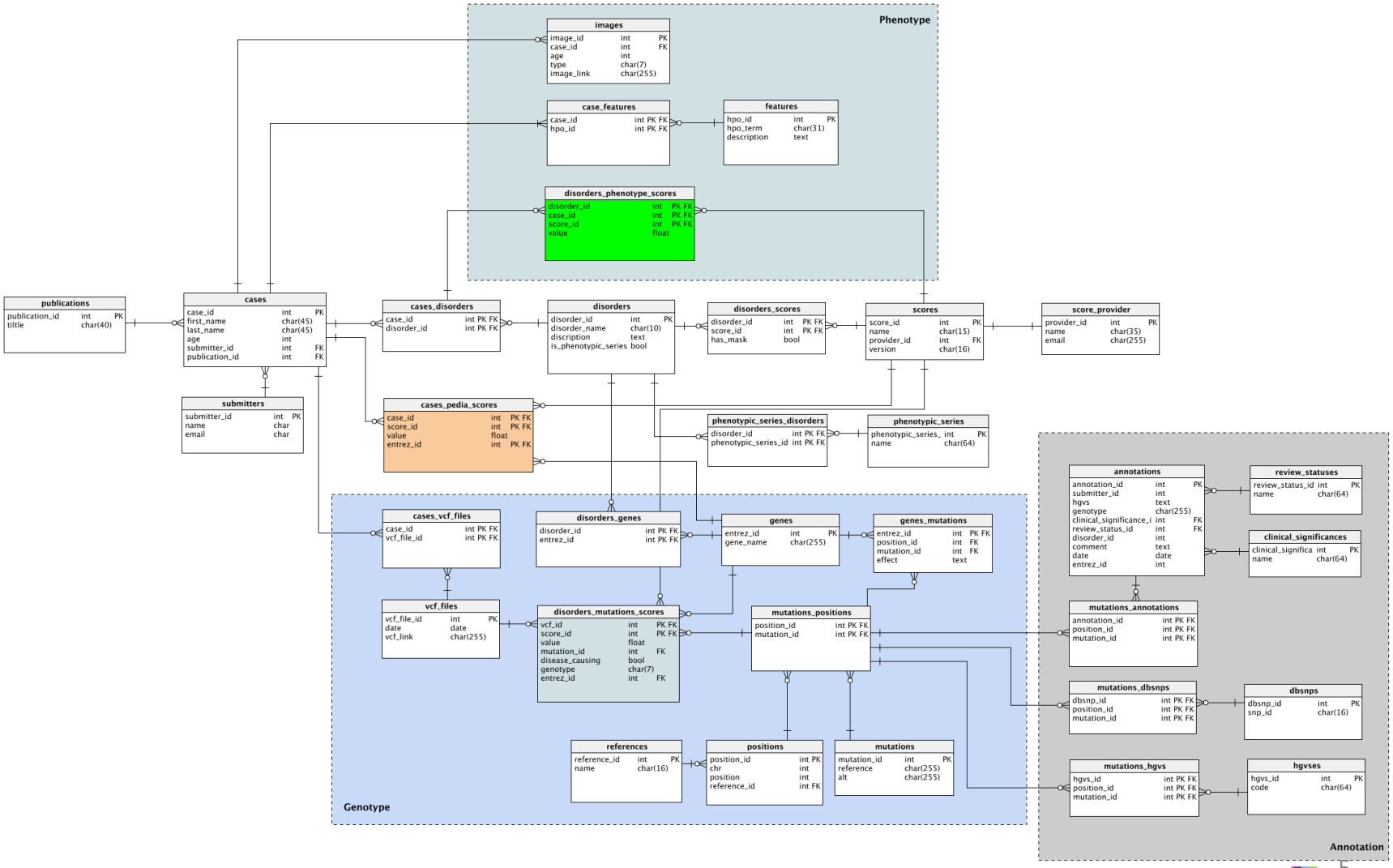
PEDIA Results

- 10 times 10-fold cross validation

Tab. 3. Result of 10-fold cross validation result on all cases

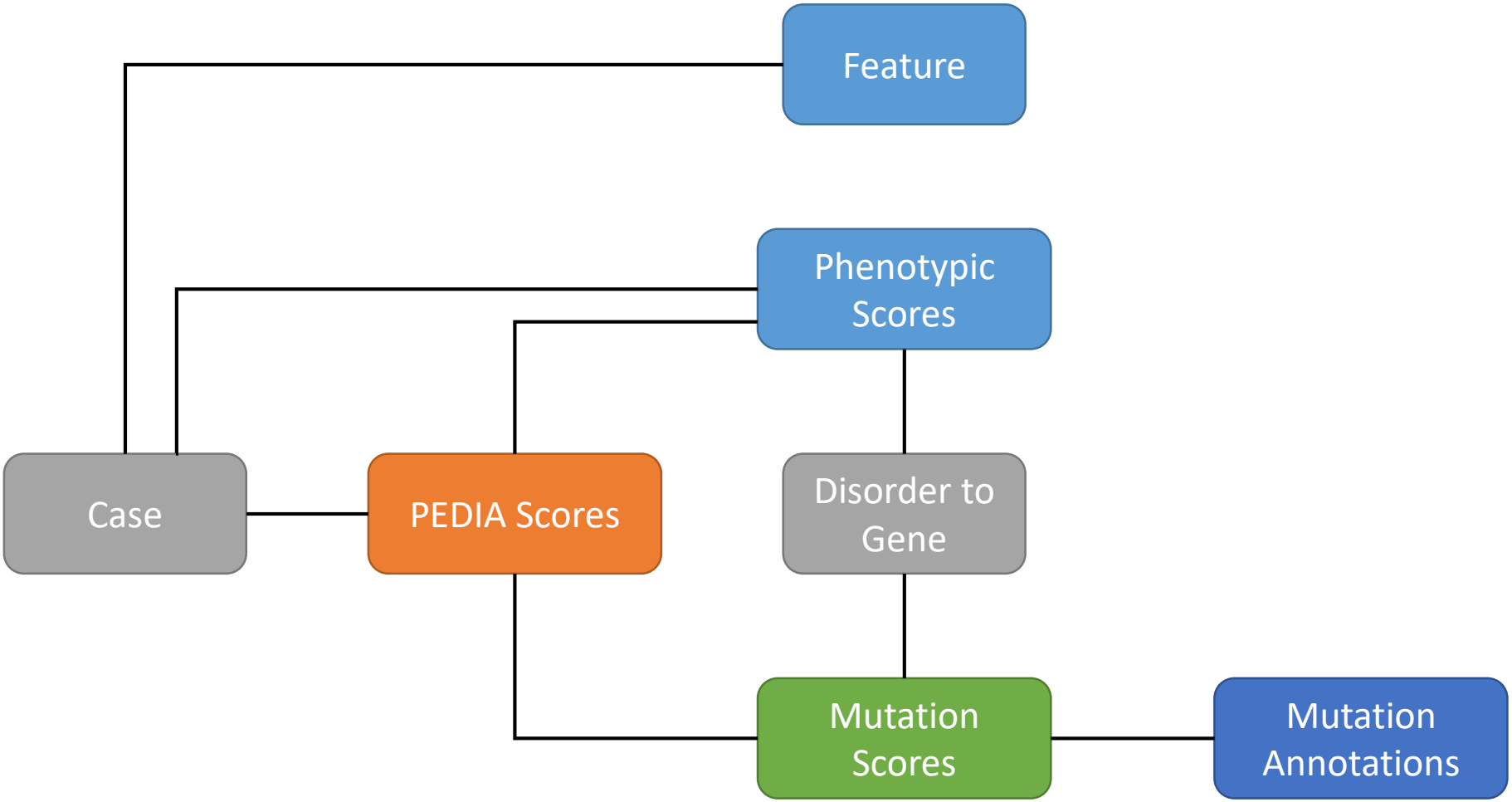
	1KG			ExAC			IRAN		
Rank	Count	Cumu(%)	pedia	Count	Cumu(%)	pedia	Count	Cumu(%)	pedia
1	260.4	76.81	4.95	264.5	78.02	4.75	252.9	74.6	5.05
2-10	62.1	95.13	1.53	61.8	96.25	1.42	64.8	93.72	1.83
11-99	13.6	99.14	-0.3	11.0	99.5	-0.52	18.3	99.12	-0.26
100+	2.9	100.0	-2.96	1.7	100.0	-2.9	3.0	100.0	-2.97
Total	339			339			339		

Structure

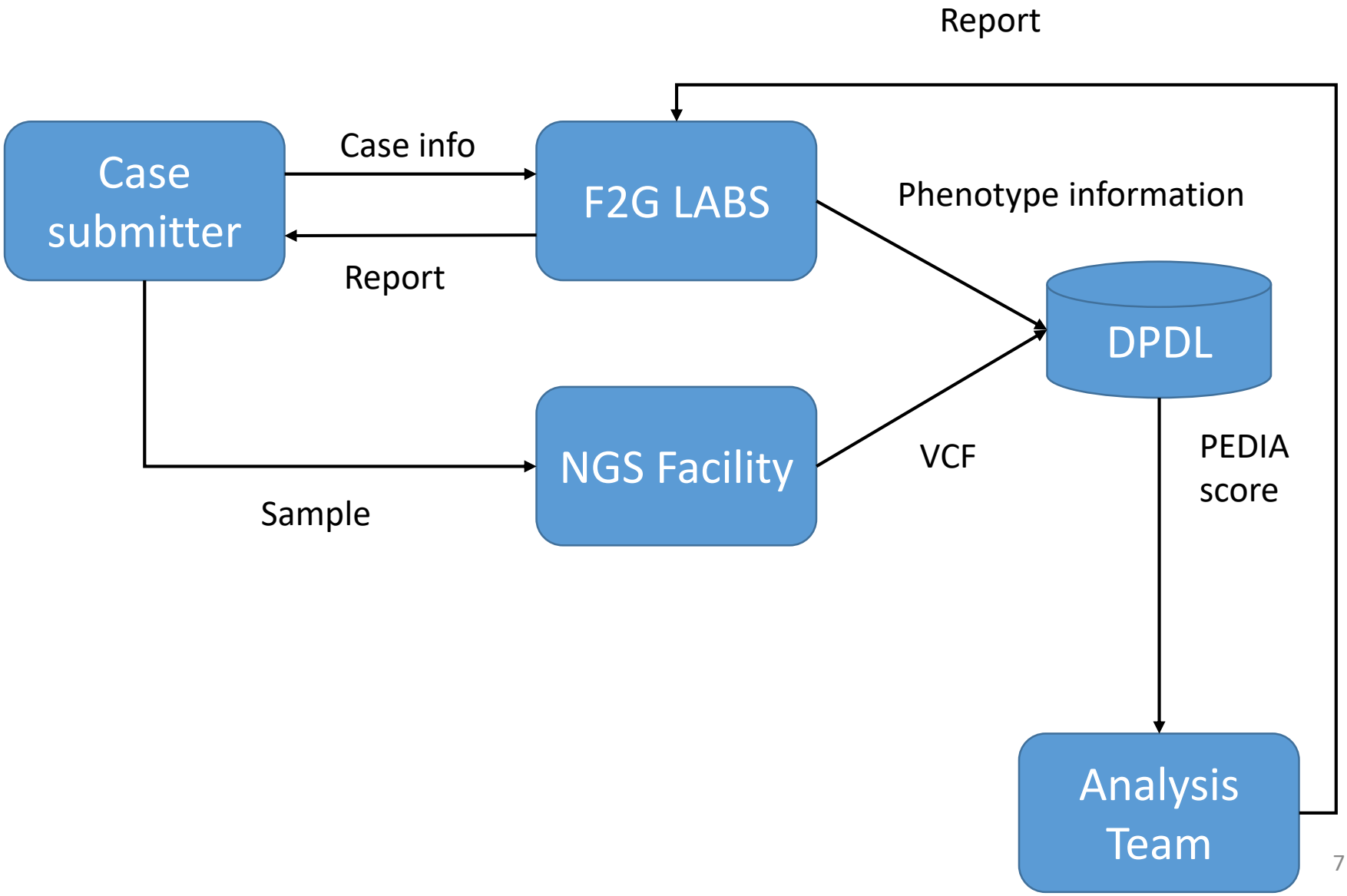


Vertabelo

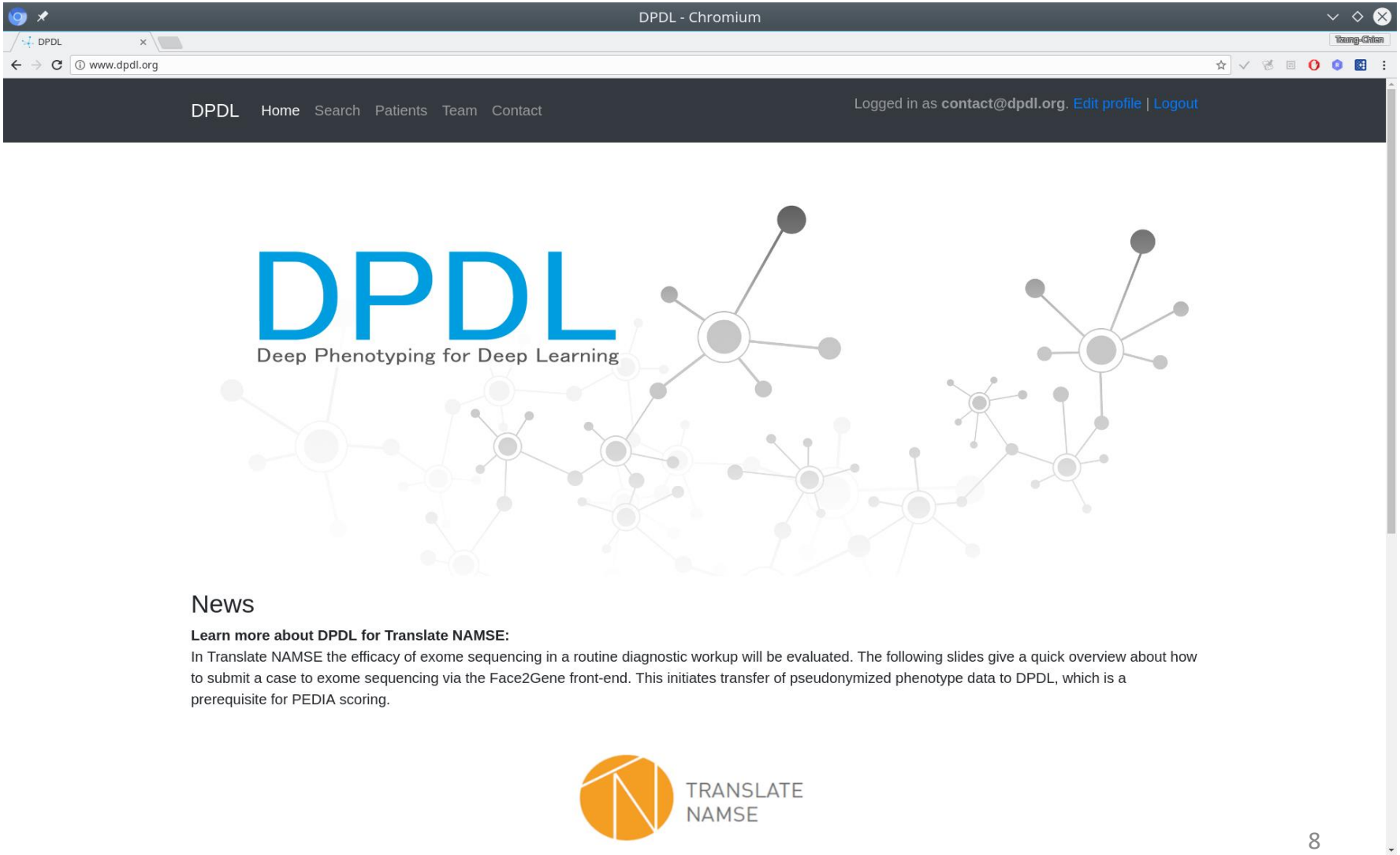
Structure


Feature
Phenotypic Scores
Case
PEDIA Scores
Disorder to Gene
Mutation Scores
Mutation Annotations

Workflow


Report
Case submitter
Case info
F2G LABS
Report
Phenotype information
DPDL
NGS Facility
Sample
VCF
PEDIA score
Analysis Team

Demo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News
Learn more about DPDL for Translate NAMSE:
In Translate NAMSE the efficacy of exome sequencing in a routine diagnostic workup will be evaluated. The following slides give a quick overview about how to submit a case to exome sequencing via the Face2Gene front-end. This initiates transfer of pseudonymized phenotype data to DPDL, which is a prerequisite for PEDIA scoring.
TRANSLATE
NAMSE

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