Agenda

- An introduction to IMPC and organisation of the data
  - Speaker: Sharon Cheng
- An introduction to querying Solr and use of the IMPC Solr APIs
  - Speaker: Marina Kan
  - Exercises: to build Solr query skills
- IMPC disease associations and use of the Phenodigm Solr core
  - Speaker: Diego Pava
  - Exercises: to practice using the Phenodigm core

Housekeeping

- Keep cameras and microphones off

- If you have any questions
  - First Talk:
    - Q & A at the end of the talk
  - Second & Third Talks and Exercises:
    - Feel Free to post your questions on the chat anytime
    - Reacting with ✅ when writing our response

IMPC

International Mouse Phenotyping Consortium

@impc

IMPC Introduction

2024-05-20

Sharon Cheng Data Wrangler

IMPC Data Coordination Centre

mousephenotype.org

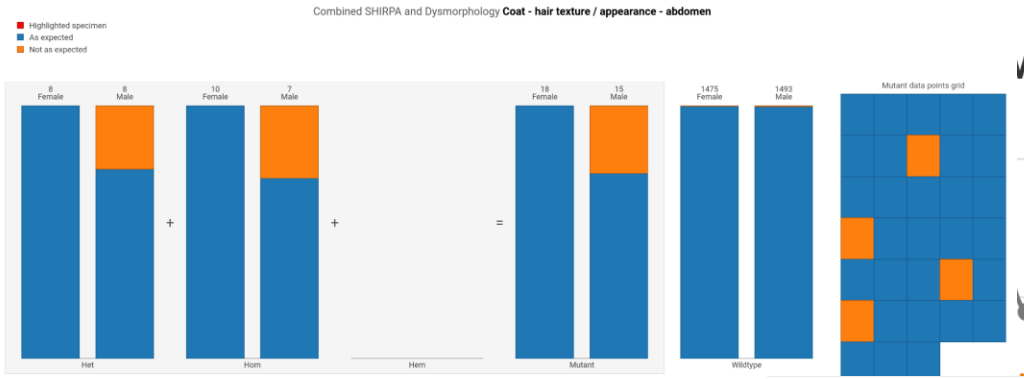
International Mouse Phenotyping Consortium

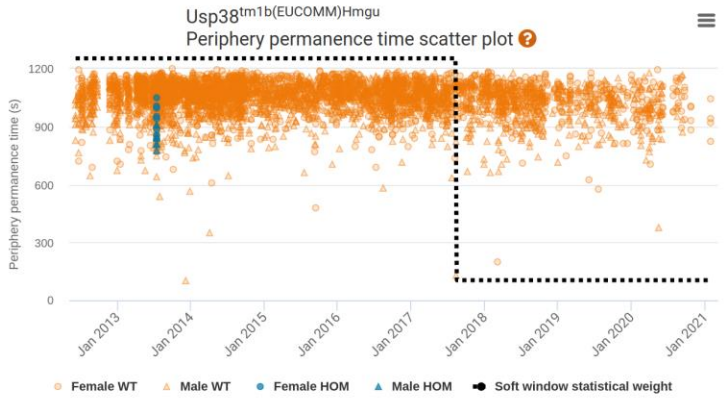
- IMPC aims to systematically knock out the protein coding genes in the mouse genome and carry out comprehensive characterisation of the mutant lines

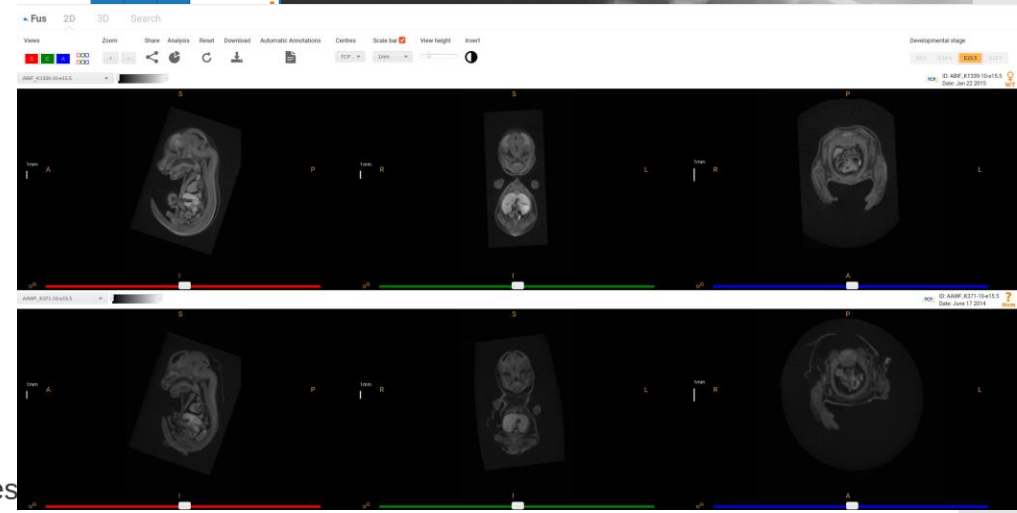
Creating a comprehensive catalogue of mammalian gene function.

IMPC in numbers (Data Release 21)

- 12 phenotyping centres
- 8,901 genes
- 9,594 mutant lines
- 95M data points
- 794K images
- 106K phenotype hits



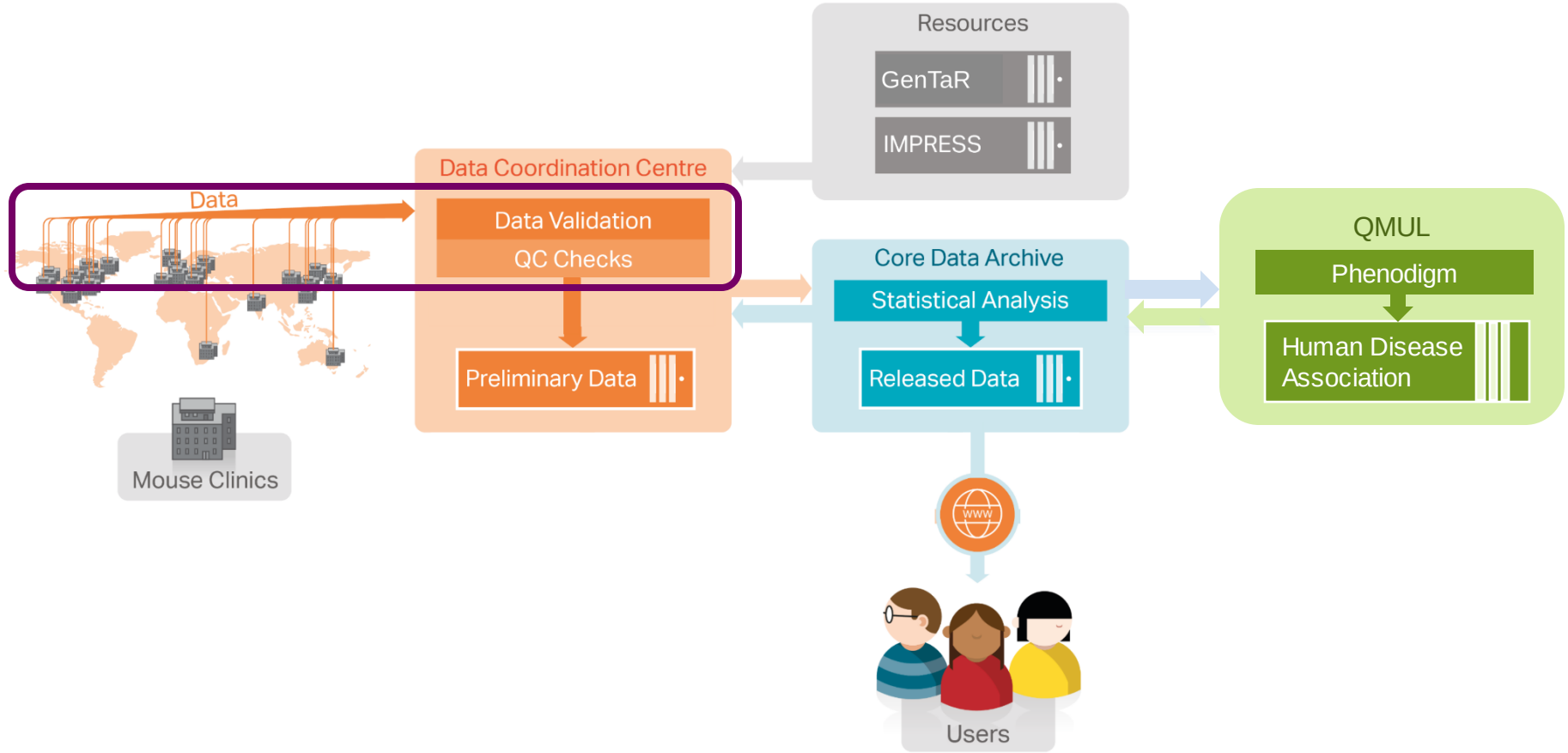




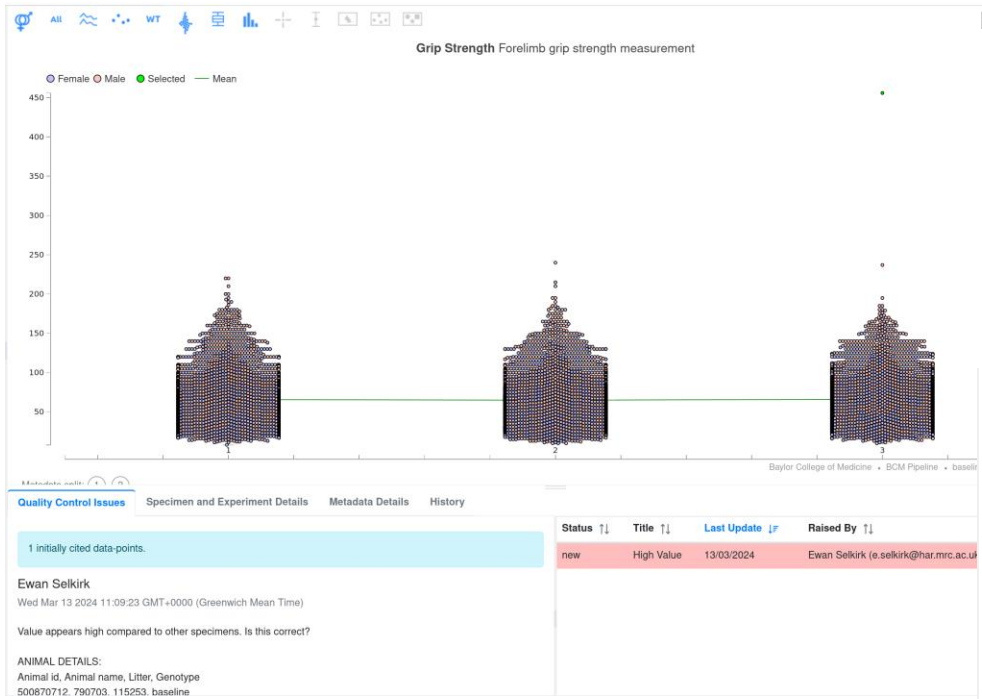
Phenotyped genes

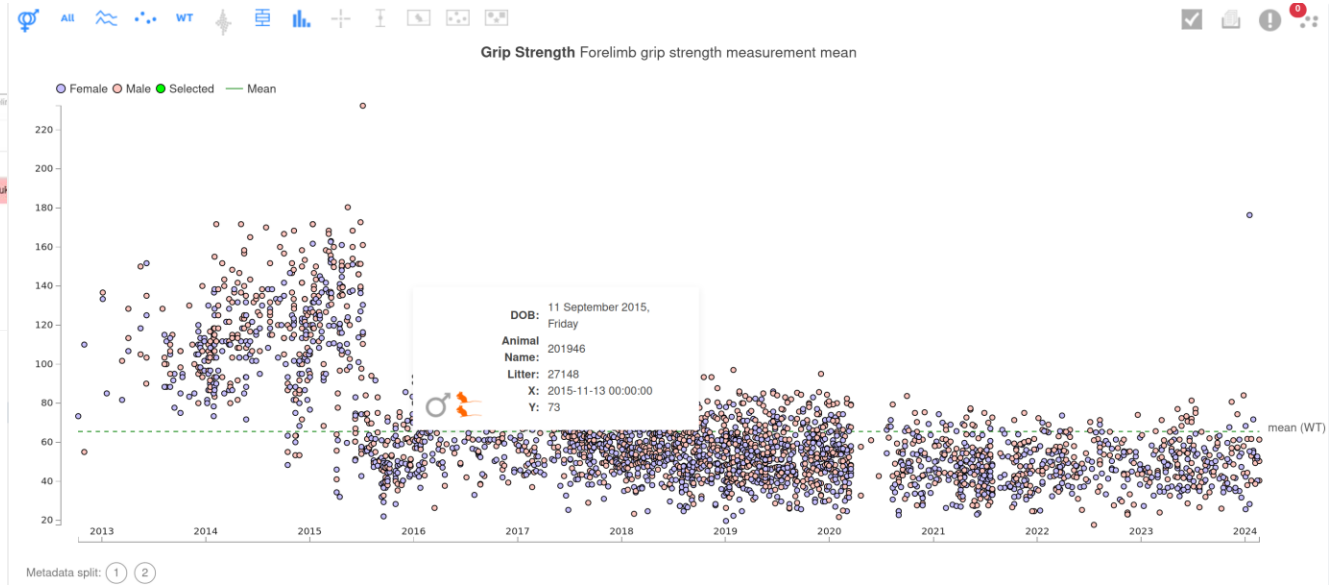
Creating a comprehensive catalogue of mammalian gene function.

Data flow from collection to available results

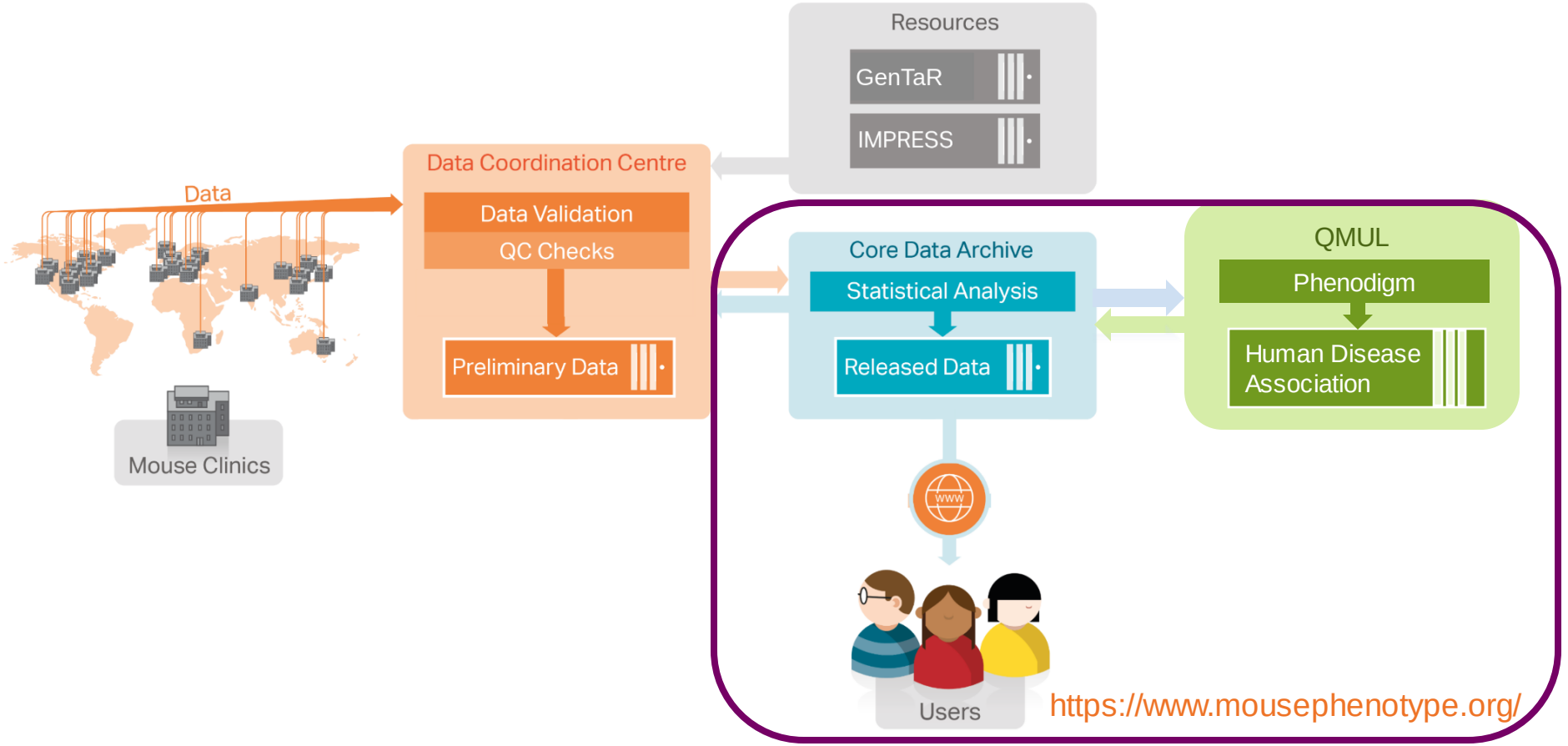


Data Quality Control

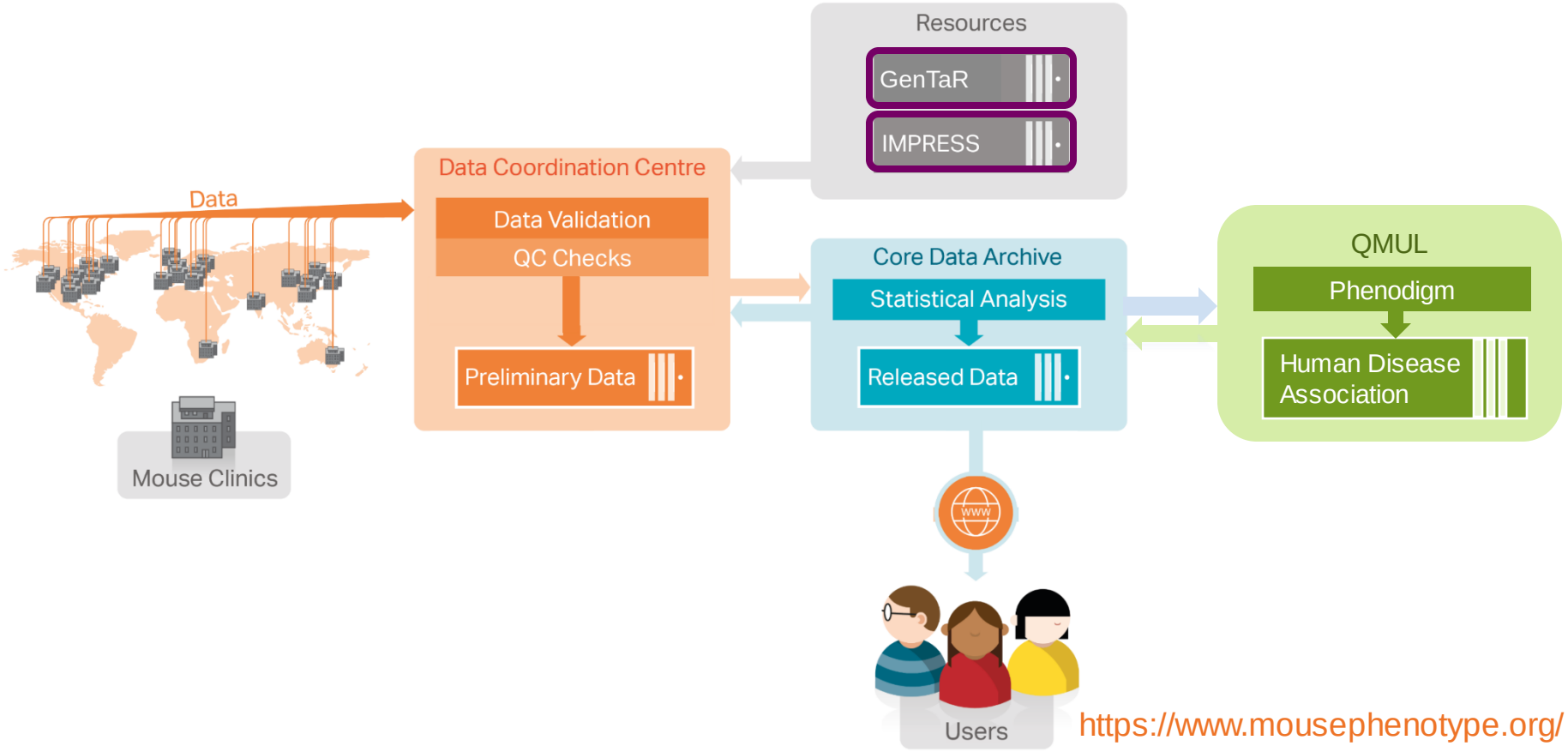




Data flow from collection to available results



Data flow from collection to available results



IMPReSS: International Mouse Phenotyping Resource for Standardised Screens

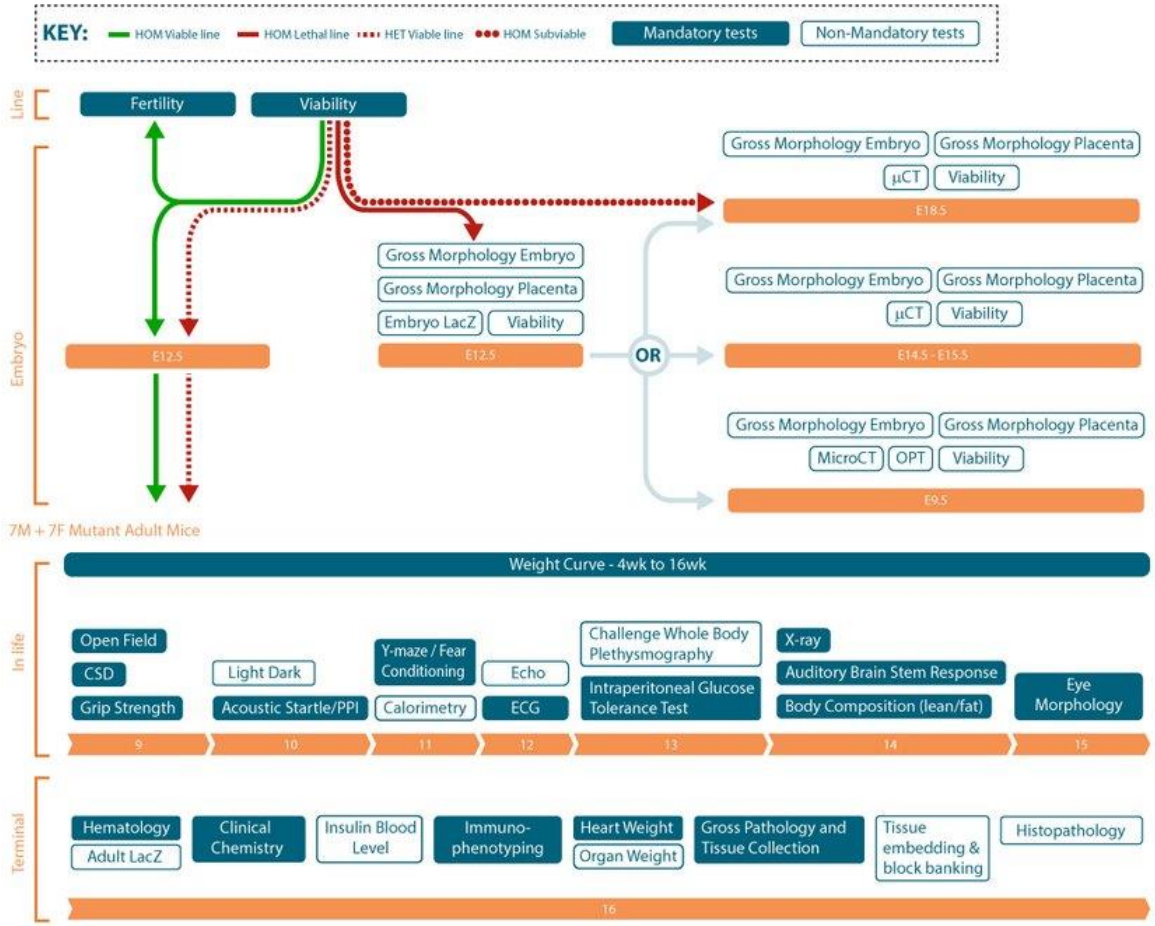
https://www.mousephenotype.org/impress/

Pipelines
(IMPC_001)

Procedures
(IMPC_GRS_001)

Parameters
(IMPC_GRS_001_001)

Ontologies
(increased grip strength)



Creating a comprehensive catalogue of mammalian gene function.

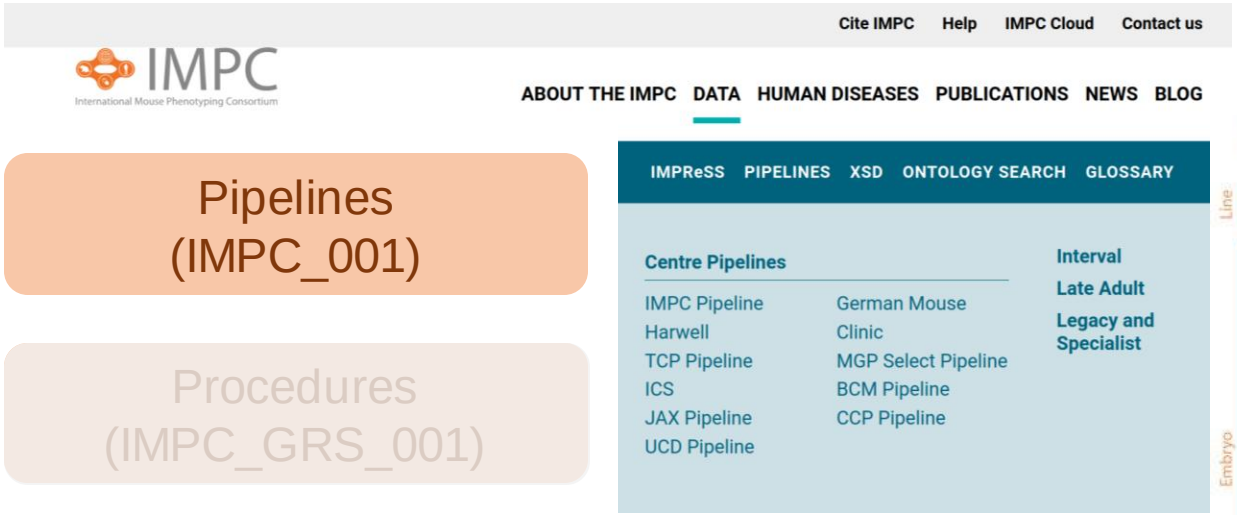
IMPReSS Pipelines

Pipelines (IMPC_001)

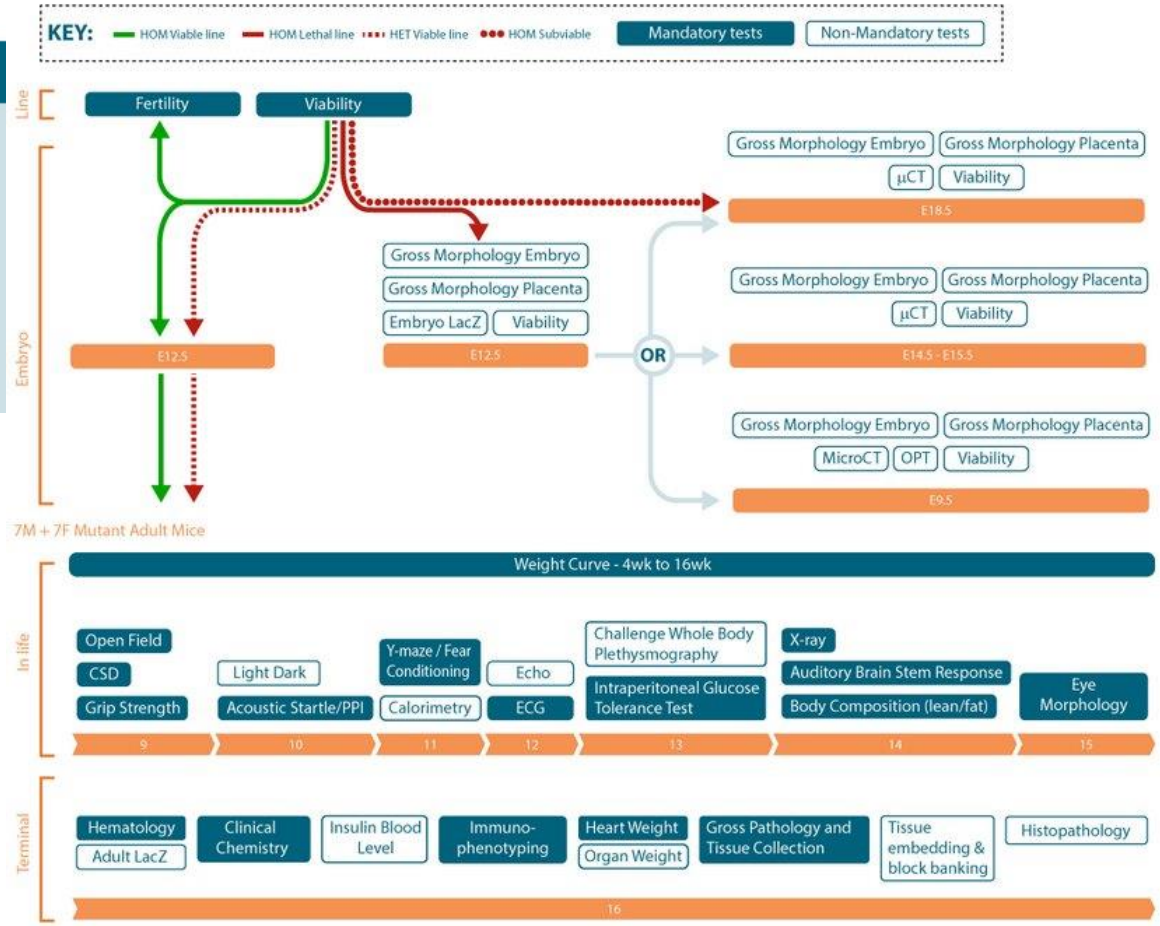
Procedures (IMPC_GRS_001)

Parameters (IMPC_GRS_001_001)

Ontologies (increased grip strength)



https://www.mousephenotype.org/impress/



Creating a comprehensive catalogue of mammalian gene function.

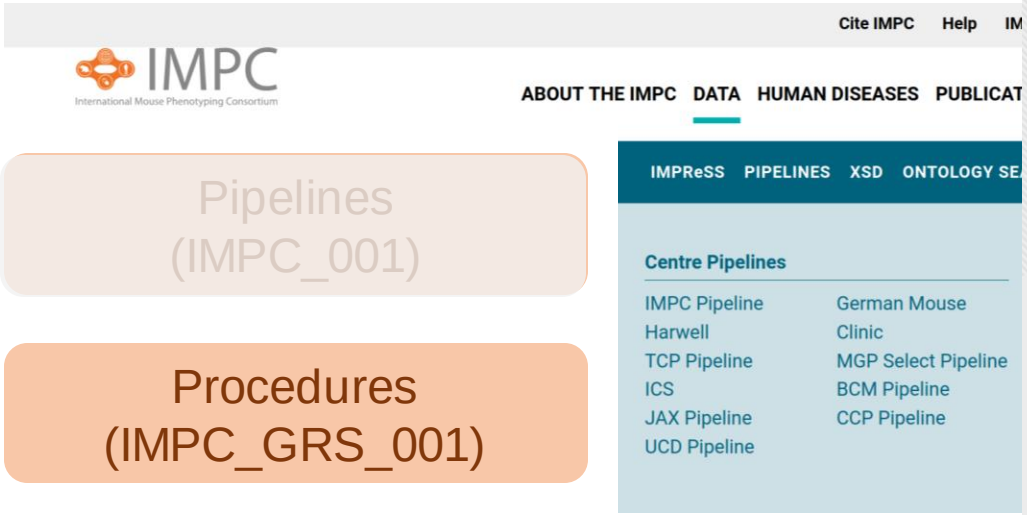
IMPReSS Standard Operating Procedures

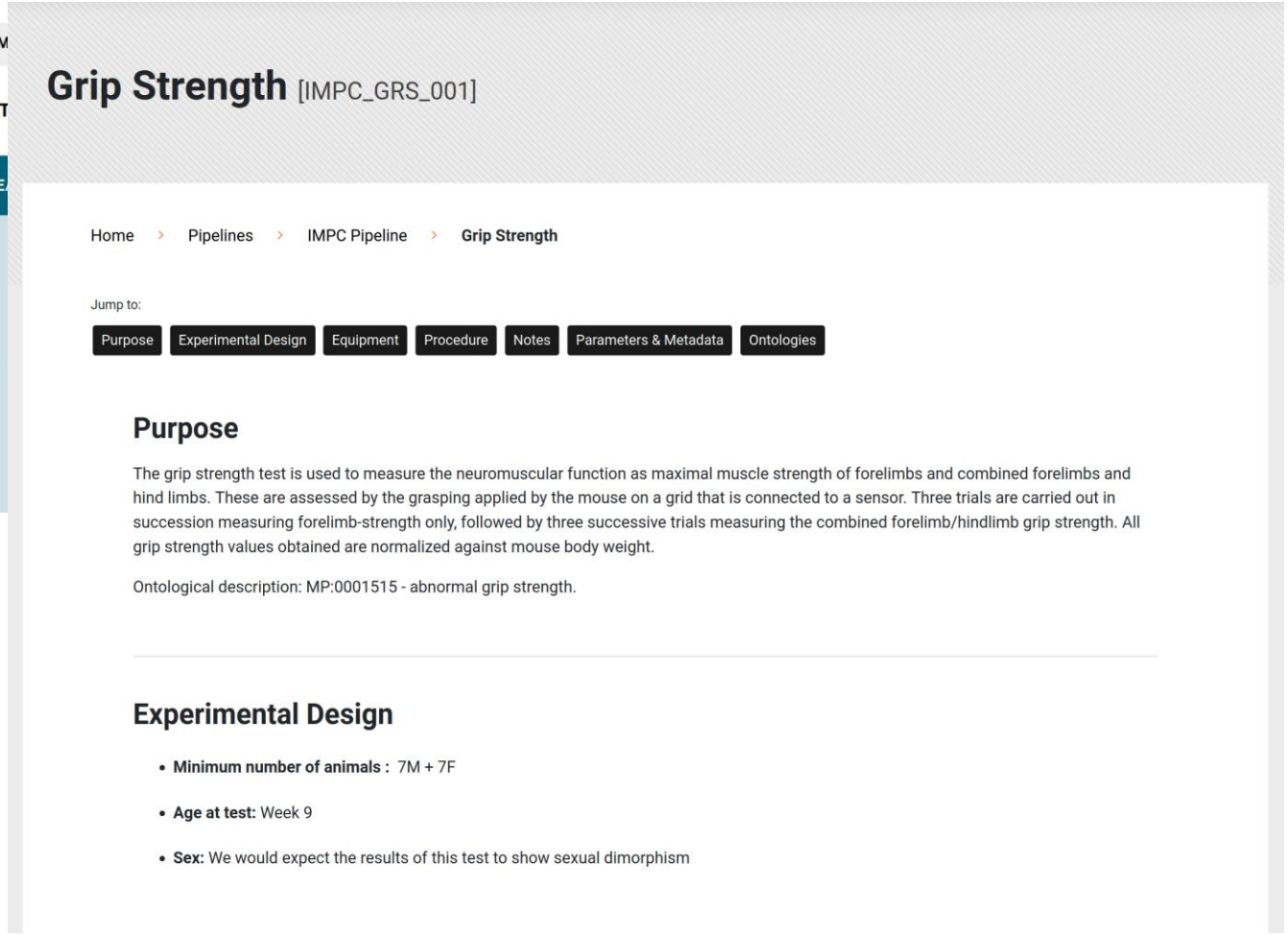
Pipelines (IMPC_001)

Procedures (IMPC_GRS_001)

Parameters (IMPC_GRS_001_001)

Ontologies (increased grip strength)





Creating a comprehensive catalogue of mammalian gene function.

IMPReSS Parameters

Pipelines (IMPC_001)

Procedures (IMPC_GRS_001)

Parameters (IMPC_GRS_001_001)

Ontologies (increased grip strength)

Name	Req. Upload	Req. Analysis	Annotation	Increment	Options	Unit	Data Type
+ Forelimb grip strength measurement [IMPC_GRS_001_001] seriesParameter				1 2 3		g	FLOAT
+ Forelimb grip strength measurement mean [IMPC_GRS_008_001] simpleParameter						g	FLOAT
Equipment manufacturer [IMPC_GRS_006_001] procedureMetadata					Chatillon Bioseb Columbus Instruments		TEXT
Grid model [IMPC_GRS_007_001] procedureMetadata					HMGU plate Not HMGU plate 45 Degree cross wired		TEXT

IMPReSS Ontologies

Pipelines (IMPC_001)

Procedures (IMPC_GRS_001)

Parameters (IMPC_GRS_001_001)

Ontologies (increased grip strength)

simpleParameter:

Forelimb grip strength measurement mean [IMPC_GRS_008_001]

Option	Increment	Ontology Term	Ontology ID
INCREASED		increased grip strength	MP:0010052
DECREASED		decreased grip strength	MP:0010053
ABNORMAL		abnormal grip strength	MP:0001515

Ways to access data https://www.mousephenotype.org/

- Data available in different ways
  - **Non-programmatic: gene pages, FTP site**
  - Programmatic: SOLR API

Human diseases caused by Plod1 mutations

The analysis uses data from IMPC, along with published data on other mouse mutants, in comparison to human disease reports in OMIM, Orphanet, and DECIPHER.

Phenotype comparisons summarize the similarity of mouse phenotypes with human disease phenotypes.

Human diseases associated with Plod1 (2 diseases) | Human diseases predicted to be associated with Plod1 (236 diseases)

The table below shows human diseases predicted to be associated to Plod1 by **phenotypic similarity**.

Disease	Similarity of phenotypes	Matching phenotypes	Source
Pleoconial Myopathy With Salt Craving		Neonatal hypotonia, Paralysis, Myopathy, Proximal amyotrophy	OMIM:262900
Aortic Aneurysm, Familial Thoracic 9		Atrial fibrillation, Aortic tortuosity, Ascending aortic dissection, Thoracic aortic aneurysm	OMIM:616166
Neuronopathy, Distal Hereditary Motor, Type VIIa		Tremor, Vocal cord paralysis, Vocal cord paresis, Distal amyotrophy	OMIM:158580

Phenotypes

The IMPC applies a panel of phenotyping screens to characterise single-gene knockout mice by comparison to wild types. Click on the different tabs to visualise significant phenotypes identified by the IMPC, as well as all data that was measured.

Significant phenotypes (2/2) | Measurements chart (218/218) | All data table (656/656)

Search

Phenotype	System	Allele	Zyg	Sex	Life Stage	P Value
decreased grip strength		Plod1$^{tm1b(EUCOMM)Hmgu}$	HOM	♀♂	Early adult	4.13×10^{-33}

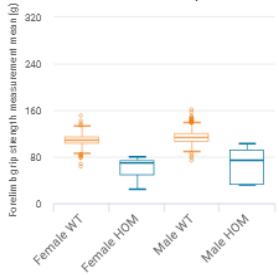
A **Grip Strength** phenotypic assay was performed on **1393 mice**. The charts show the results of measuring **Forelimb grip strength measurement mean** in **8 female, 8 male** mutants compared to **709 female, 668 male** controls. The mutants are for the **Plod1$^{tm1b(EUCOMM)Hmgu}$** allele.

* The high throughput nature of the IMPC means that large control sample sizes may accumulate over a long period of time. See the animal welfare guidelines for more information.

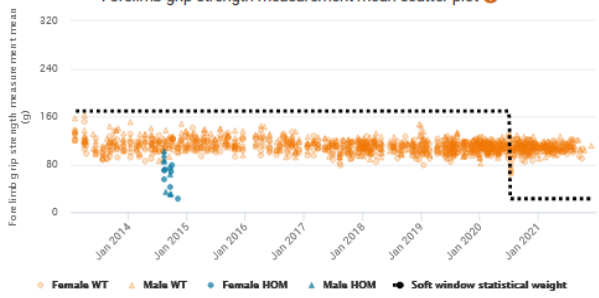
Testing protocol	Grip Strength
Testing environment	Lab conditions and equipment
Measured value	Forelimb grip strength measurement mean
Life stage	Early adult
Background Strain	involves: C57BL/6NCrl
Phenotyping center	TCP
Associated Phenotype	decreased grip strength

Mouseover the charts for more information. Click and drag to zoom the chart. Click on the legends to disable/enable data.

Plod1$^{tm1b(EUCOMM)Hmgu}$
Forelimb grip strength measurement mean box plot



Plod1$^{tm1b(EUCOMM)Hmgu}$
Forelimb grip strength measurement mean scatter plot



Ways to access data https://www.mousephenotype.org/help/programmatic-data-access/

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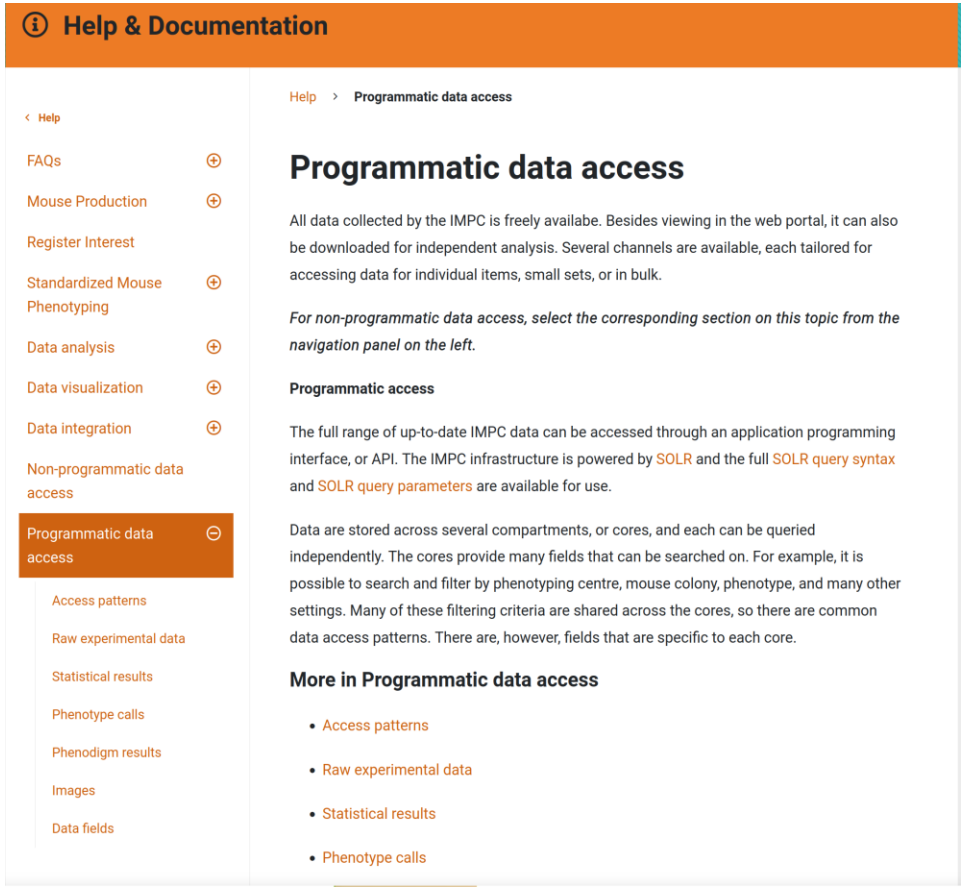
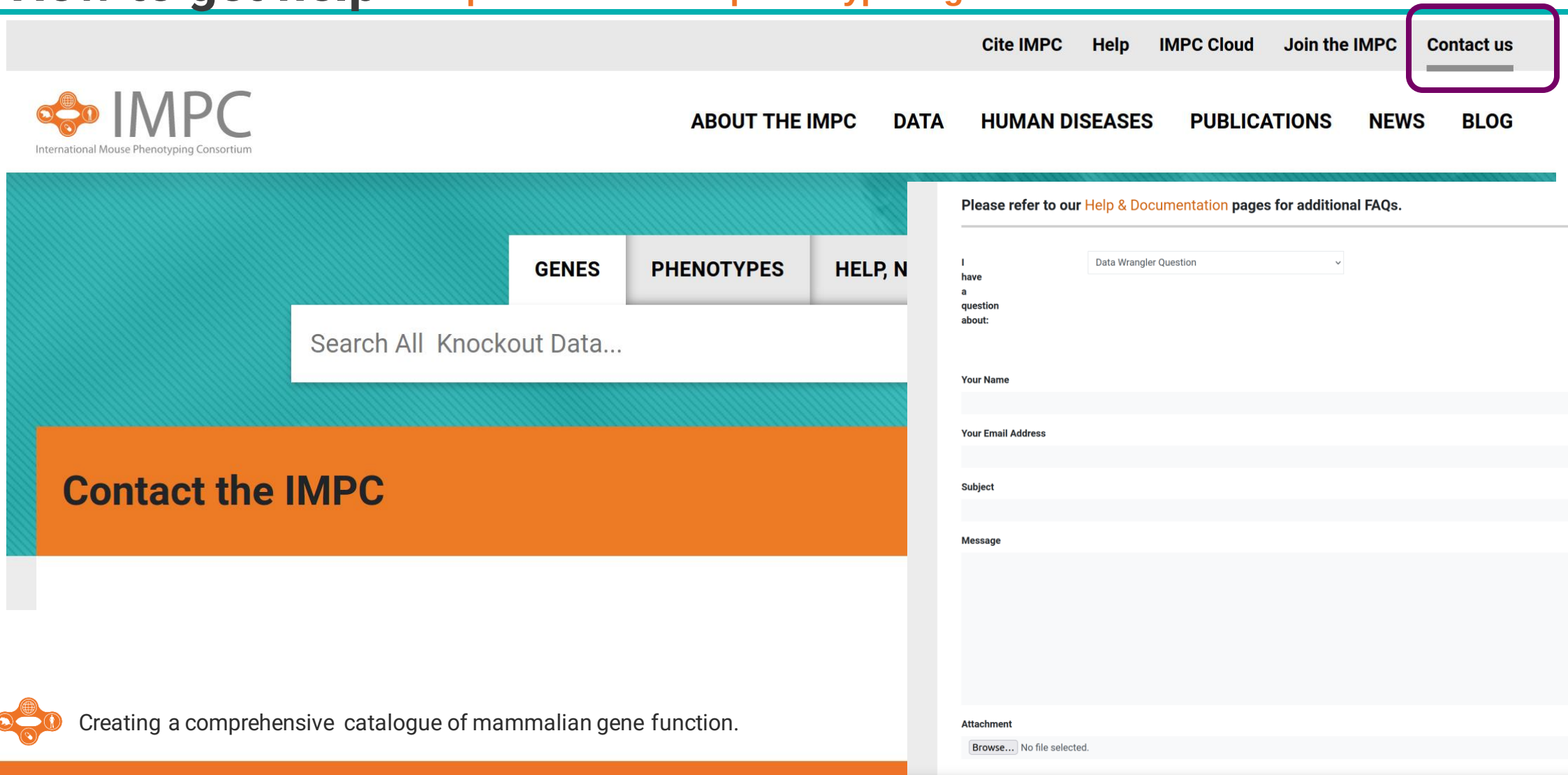


How to get help https://www.mousephenotype.org/contact-us/



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