

## Common Terminology:

**Colony** – A group of mice which have arisen from the chimera(s) with a certain modified allele or alleles. The same gene or genes altered in a different way would be a different colony.

**Colony Prefix** – a unique short code for a colony of typically 3 or 4 characters.

**Mouse ID/name** – in the format XXXX99.9x

- XXXX is the upper-case colony prefix the mouse is born into.
- 99 is the mating number the mice were born from.
- 9 is the litter number for that mating.
- x is the individual lower-case letter (a-z) assigned to the mouse.
- e.g. CALB102.4b indicates CALB colony, 102nd mating, 4th litter for mating 102 and the second mouse in the litter.

**Mouse Barcode** – In the format M00164837; M for Mouse and a unique number.

**Cohort** - A group of mice from a particular colony that was grouped together based on pipeline and date of birth. A cohort would enter the pipeline and go through each of the tests together. There will be multiple cohorts from the same colony, issued at different time points to give a total of 7 mutant male and 7 mutant female mice (earlier pipelines may have had more mutants).

**Cohort Name** – Consists of colony prefix, pipeline, average DOB in format YYYYMMDD e.g. MEAD MGP Select 20121129.

**Pipeline** – A series of experimental assays grouped together and always performed in a specific order at designated time points. Mice on a pipeline would have undergone all of those assays unless deemed unfit to do so.

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## Populations:

A population was an internal WSI concept that consisted of 3 types of mice:

*Subjects* - The mutant mice with the desired allele, usually from the same colony. The subjects were made up of multiple cohorts of mice to give the complete set of experimental animals for a colony.

*Controls* – Typically wild-type mice of the same age and genetic background as the mutant mice, processed through the assay pipeline at the same time as each of the cohorts of mutant mice.

*Baseline* - All of the wild-type mice of the same age and genetic background which underwent the same assay pipeline of the mutant mice. Used to provide information on how a normal group behaves.

By grouping these 3 types into this 'population', it was possible to graph the assay data on a per parameter level allowing comparison between the local and baseline controls against the subject data.

Additionally, a population was specific to a phenotyping pipeline and background strain, both chosen on creation.

Once created, the baseline group was automatically updated within the database as part of the overnight regeneration/update cycle. The only 2 groups amended by the user would be the controls and subjects.

A population would also be 'locked' to prevent any further change once all the mice had been processed through the assay pipeline. This allowed for all nightly graphing activity to stop for that population and for the end-users to start making significance calls and adding annotation terms.

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### **Heat Map:**

The heatmap is a visualisation of the phenotypic calls of significance made for each assay an allele was tested for. Mutant data were compared to the relevant baseline, composed of all animals of the same sex, age and genetic background that had been screened using that particular experimental protocol.

See the README and Data Structure & Analysis files for more details as well as the documentation in the Heat\_Map subdirectory.

*MP Terms* – Mammalian Phenotype Ontology Term

([http://www.informatics.jax.org/vocab/mp\\_ontology/](http://www.informatics.jax.org/vocab/mp_ontology/)); a method for organizing and classifying phenotyping information from mammalian species. This allows for easy comparison of data from difference centres and sources to facilitate discoveries of genes involved in different phenotypes.

*MA Terms* – Adult Mouse Anatomy Term ([http://www.informatics.jax.org/vocab/gxd/ma\\_ontology/](http://www.informatics.jax.org/vocab/gxd/ma_ontology/)); a method for easily classifying the area of the mouse affected by an observation.

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### **Consortia:**

*IMPC* – International Mouse Phenotyping Consortium (<https://www.mousephenotype.org/>)

An international collaboration of several research institutes and universities with the goal to discover functional insight for every gene by generating and systematically phenotyping 20,000 knockout mouse strains.

*DMDD* – Deciphering the Mechanisms of Developmental Disorders (<https://dmdd.org.uk/>)

A Strategic Award granted by the Wellcome Trust to identify abnormalities in embryo and placental structure for embryonic lethal mouse lines. A combination of detailed imaging and morphological phenotyping as well as transcriptional profiling was used.

*OBCD* – Origins of Bone and Cartilage Disease (<http://boneandcartilage.com/>)

A Strategic Award granted by the Wellcome Trust to the Sanger Institute in collaboration with Imperial College London to investigate the causes and developments of joint disorders in mice (e.g. osteoarthritis and osteoporosis). Mice were produced at the Sanger and samples shipped post-necropsy to Imperial for analysis.

*3i* – Infection, Immunity, Immunophenotyping (<https://www.immunophenotype.org/threei/home>)  
A Strategic Award granted by the Wellcome Trust to the Sanger Institute in collaboration with King's College London, Imperial College, Albert Einstein College of Medicine, and the Universities of Manchester, Cambridge, and Oxford. *3i* conducted a broad and deep immunological phenotyping of mouse lines generated by the Wellcome Sanger Institute.

*EUMODIC* – European Mouse Disease Clinic (<https://cordis.europa.eu/project/id/37188>)  
A forerunner to the IMPC project, *EUMODIC* was a consortium of centres that worked to scale up phenotyping of mutant mouse lines. Over 400 lines were produced and over 300 of those were phenotyped, laying the foundations for the IMPC protocols and procedures that came later.