How to search for phenotyping data on a gene of interest?

1. From the Sanger Mouse Portal home page (<u>http://www.sanger.ac.uk/mouseportal/</u>), enter your gene of interest e.g. *Mysm1* into the **Search** field and click search:

| sanger | A- A A+ |
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| institute | Mouse Resources Portal |
| HOME BROWSE HELP ABOUT | Mysm1 Search |

About This Portal

The Wellcome Trust Sanger Institute generates, characterises, and uses a variety of reagents for mouse genetics research. It also aims to facilitate the distribution of these resources to the external scientific community. Here, you will find unified access to the different resources available from the Institute or its collaborators. The resources include: 129S7 and C57BL6/J bacterial artificial chromosomes (BACs), MICER gene targeting vectors, knock-out first conditional-ready gene targeting vectors, embryonic stem (ES) cells with gene targeted mutations or with retroviral gene trap insertions, mutant mouse lines, and phenotypic data generated from the Institute's primary screen.

Phenotyping Summary

The table below displays the **number of alleles** that have completed different stages of the MGP Phenotyping pipeline (funded by WTSI & EUMODIC)

| Comprehensive Phenotyping Substantively Complete | 564 |
|--------------------------------------------------|-----|
| Infection Challenge (Salmonella and Citrobacter) | 495 |
| LacZ Expression (Adult and E14.5) | 339 |

Other Data Summary

The table below displays the **number of unique genes** with each individual product type produced at the Wellcome Trust Sanger Institute.

| Funding: | WTSI EUMODIC | KOMP EUCOMM | KOMP EUCOMM | WT | WT | WТ |
|-----------|-----------------|-------------------|------------------------|-------|---------------|------------|
| Products: | Mice | Targeted ES Cells | Gene Targeting Vectors | MICER | C57BI/6J BACs | 12957 BACs |
| | 904 | 13011 | 15105 | 6719 | 12037 | 11874 |

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Phenotyping Overview

Download a heat map of phenotyping results by assay. This spreadsheet also links to the detailed phenotyping data on this portal.

You can also access (and search) the same data via the MGP Phenotyping BioMart.

NEW - Disease predictions based on semantic similarity between OMIM disorder clinical features and model organism phenotypes.





- 2. This opens a gene specific page containing information split into the following panels that are populated when resources are available:
 - i. Gene Details
 - ii. WTSI Phenotyping
 - iii. Mice
 - iv. IKMC Vectors and ES Cells
 - v. DNA Clone Resources
- 3. The WTSI Phenotyping panel presents the primary phenotyping data as a colour coded heat map style summary of all phenotyping data for that gene. Data are grouped for presentation in two ways:
 - i. As a Mammalian Phenotype (MP) Ontology Based Heatmap

| TP Untology I | Based Heatmap | | | | | | | | | | | | | | | | | | | | | | | | |
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| Allele Name | Colony Prefix | Colony Strain | adipose tissue | behavior/neurological | cardiovascular system | cellular | craniofacial | digestive/alimentary | embryogenesis | endocrine/exocrine gland growth/size | hearing/vestibular/ear | hematopoietic system | homeostasis/metabolism | immune system | integument | limbs/digits/tail | liver/biliary system | mortality/aging | muscle | nervous system | other | pigmentation | renal/urinary system | reproductive system | respiratory system |
| Mysm1 ^{tm1a} (KOMP)Wtsi | MAHN | C57BL/6Dnk;C57BL/6Dnk;C57BL/6N | • | • | 1 | d | | | I | il il | i . | d . | • | i | | i | | d. | | | | 1 | 1 | 1 | |
| Legend: | No Raw Data | No Significant Annotations | Si | gnific | ant A | nnota | tion P | resen | | Lin | k to a | test r | enort | nad | _ | | | | | | | | | | |
| Ab | | | | | | | | reach | | | | | epon | , pag | | | | | | | | | | | |
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| About Thi This heatn perform a annotated Please No Histopatho Study', 'Ta | is Heatmap lap is built up of t given test we can to a top-level MP ote: as mentioned logy', 'Embryo La il Epidermis Whol | op-level terms from the MP (Mammalia say if it should be annotated with the term (or any of its children), the cell w l above we do not annotate all of our p cZ Expression', 'Fertility', 'Heart Histolo emount', 'Tissue Biobank' and 'Viability | n Phe given ill be henot gy', 'l at W | motyp MP te red, i yping Micror eanin | oe) or erm (if the tests nuclei ig'. Yo | ntolog if the result s to th ou car | pheno t was the MP croCT | have have not sig ontolo & Qua our re | e map is sig gnific ogy. 1 antita esults | ped a m nificant) ant, the The tests tive Fax for thes | najorit . This cell w s that itron', se test | y of ti is wh ill be are e: 'Plasi is in th | he pa at yo blue. xclude ma In he 'Ph | rame u see ad ar nmur ienot | eters i e in th re: 'Ac noglob yping | in our iis he dult L bulins I Test | r test atma acZ E s', 'Pr : Base | s to a p - if Expre epuls ed He | appro a sig ssion se Inh atma | priate nifica nibitio ap' be | e MP int re ain/E n', 'R low. | terms sult h ye/Sl tecess | s, so has be kin sive L | when een .ethal | we |



ii. As a Phenotyping Test Based Heatmap, opened by clicking the bar of the same name. Note this action causes the MP Ontology Based Heatmap to roll up.

| - WTSI | Pheno | otyping | | | | | | | | | | | | | | | | | | | | | | | | | | |
|--------------------------------|---------------------------------------------------|-------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------|----------------------|------------------------|--------------------------|----------------------------|-----------------------|------------------------|-----------------|------------|-----------------|---------------|-----------|---------------|----------|-----------------------------|---------------------------|-----------------------|------------------------|-----------------------------|-------------------------|---------------|-----------------------------|----------------|------------------|------------------------|-------------------|
| O MP Onto | ology Bas | sed Heatmap | | | | | | | | | | | | | | | | | | | | | | | | | | |
| O Phenoty | /ping Te | st Based Heat | map | | | | | | | | | | | | | | | | | | | | | | | | | |
| Allele | Name C | Colony Prefix | Colony Strain | Viability at weaning | Recessive Lethal Study | Fertility | Embryo Lac2 Expression | Adult Lac2 Expression | General Observations | Weight Curves | Open Field | Modified SHIRPA | Grip Strength | Hot Plate | Dysmorphology | Rotarod | Non-Invasive Blood Pressure | Prepulse Inhibition | Indirect Calorimetry | Glucose Tolerance (ip) | Auditory Brainstem Response | Body Composition (DEXA) | X-ray Imaging | Stress Induced Hyperthermia | Eye Morphology | Plasma Chemistry | Plasma Immunoglobulins | Haematoloov (FRF) |
| Mysm1 ^t (KOMP)Wt | tmia M | MAHN | C57BL/6Dnk;C57BL/6Dnk;C57BL/6N | 1 | <u>_</u> | <u>i</u> | <u>i</u> | 1 | | 1 | <u>ı</u> | <u>i</u> | <u>i</u> | i | <u>i</u> | <u>i</u> | ii | <u>i</u> | <u>d</u> | <u>i</u> | <u>il</u> . | <u>ı</u> | <u>ı</u> | <u>i</u> | <u>ı</u> | <u>ı</u> | ₫. | 4 |
| Legend Te Te Te | d - hover st comple st is comp st abando | readsheet conta over each headi ite and data/res plete but the dat pned | aining all WTSI phenotyping data ing to get a more complete description ources are available ta are not considered interesting | Te Te Lin | est is d est not | comp t perf a test | llete a forme t repo | and th ed or a | ne dai applio ge | ta are cable | e cons | sidere | ed inte | erest | ing | P | Prel Tes Link | limina t pen < to a | ary ir ding pdf | ndicat | tion c | of an i | intere | esting |) pher | notyp | e | |
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- 4. Both heatmaps list details of each mutant mouse line that has been phenotyped by the Sanger MGP for that gene, one row representing one unique allele:
 - Allele Name (column A): Full allele name carried by the mutant mouse line
 - Colony Prefix (column B): Unique identifier for the mutant mouse line
 - Colony Strain (column C): Genetic background upon which the mutant mouse line was phenotyped



- 5. The remaining columns present a summary of the phenotyping data collected by the Sanger Mouse Genetics Project split either based on top level Mammalian Phenotype ontology terms (for details of the MP Ontology see <u>http://www.informatics.jax.org/searches/MP_form.shtml</u>) or by the phenotyping test. The legend for each presentation is inserted below each heatmap. Note you may need to scroll to the right of the heatmap to see all its contents.
- 6. From the Phenotyping Test Based Heatmap, clicking on any graph symbol describing the standard protocol and parameters collected for that mutant mouse line/phenotypic test combination.







7. Where the test is complete and the data are considered interesting (red cells), the Ink opens the web page on the Sanger Mouse Portal describing the standard protocol, listing the parameters collected and the Mammalian Phenotype (MP) annotations assigned to describe each observed phenotype, and providing a link to view the graph for each parameter considered significant.

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| ray Imaging | Data for | Mysi | m1 (| (MA | HN) | | | |
| Standard Protocol - <i>sig</i> | nificant param | eters | | | | | | |
| X-Ray Morphology (wee using a Faxitron system protocol | k 14) [Mice were MX20 (Faxitron X | fed on Hig (-ray Corp | Ih Fat Di oration) | iet (21.4 . Mice a | 1% crude fat co are anaesthetis | ontent, Western F ed and up to five | RD, 829100, Special Diets Services) fro standard images are taken for each m | im 4 weeks of age]: Digital X-Ray images are acquired nouse. The images are annotated using a standardised |
| ▼ view legend | | | | | | | | |
| Legend - hover over ead | ch heading to get | a more cor | mplete d | descripti | on | | | |
| Test complete and d | data/resources ar | e available | | | Test is c | complete and the | data are considered interesting | Preliminary indication of an interesting phenotype |
| Test is complete but | t the data are not | considere | d interes | sting | Test not | performed or ap | plicable | Test pending |
| / Test shands and | | | | | | | | |
| rest abandoned | | | | | • •• | | | |
| Parameter | Female Het | Female Hom | Male Het | Male Hom | population parameter | Graph | MP Annotation | |
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| Parameter Skull Shape Zygomatic Bone | Female Het | Female Hom | Male Het | Male Hom | population parameter | Graph | MP Annotation MP:0000438 - abnormal cranium mor | rphology |
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| Parameter Skull Shape Zygomatic Bone Maxilla Mandible | Female Het | Female Hom | Male Het | Male Hom | population parameter | Graph | MP Annotation MP:0000438 - abnormal cranium mor | rphology |
| Parameter Skull Shape Zygomatic Bone Maxilla Mandible Teeth | Female Het | Female Hom | Male Het | Male Hom | population parameter | Graph | MP Annotation MP:0000438 - abnormal cranium mor | rphology |
| Parameter Skull Shape Zygomatic Bone Maxilla Mandible Teeth Scapula | Female Het | Female Hom | Male Het | Male Hom | population parameter | Graph | MP Annotation MP:0000438 - abnormal cranium mor | rphology |
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| Parameter Skull Shape Zygomatic Bone Maxilla Mandible Teeth Scapula Clavicle Number of Ribs Right | Female Het | Female Hom | Male Het | Male Hom | population parameter | Graph view graph | MP Annotation MP:0000438 - abnormal cranium mor | rphology |
| Parameter Skull Shape Zygomatic Bone Maxilla Mandible Teeth Scapula Clavicle Number of Ribs Right Number of Ribs Left | Female Het | Female Hom | Male Het | Male Hom | population parameter | Graph Constraints Constraints Constraint | MP Annotation MP:0000438 - abnormal cranium mor | rphology |



8. Click on the kiew graph icon to open a window that allows you to scroll through graphs of data from both sexes for each significant parameter.







- 9. The raw data from mutant mice (Subjects), wildtype mice run on the same week as the mutants (Controls) and, where appropriate, all age, sex, genetic background and pipeline matched wildtype mice (Baseline) are viewable by clicking the view data icon.
- 10. The raw data from mutant mice (Subjects), wildtype mice run on the same week as the mutants (Controls) and, where appropriate, all age, sex, genetic background and pipeline matched wildtype mice (Baseline) can be downloaded as an XML document by clicking the solution download data icon.
- 11. From the MP Ontology Based Heatmap, clicking on a graph symbol in may open the web page on the Sanger Mouse Portal describing one standard protocol or a collection of standard protocols grouped under a single MP term. For example, the MP term "limbs/digits/tail" groups data from Dysmorphology, X-ray imaging, MicroCT & Quantitative Faxitron and, where related MP terms have been assigned, Recessive Lethality.

A- A A+ Mouse Resources Portal Mome BROWSE HELP ABOUT Phenotyping Data for Mysm1 (MAHN) Associated with 'limbs/digits/tail' (MP:0005371) Opysmorphology: Dysmorphology - significant parameters A- A A+ Mouse Resources Portal Search

Recessive Lethal Study: Standard Protocol - significant parameters

MicroCT & Quantitative Faxitron: Standard Protocol

12. Data from each of these phenotyping tests can be accessed as described above (steps 6-10) simply by clicking the standard protocol of interest.



