

HMGU proposals for the CASIMIR Work Package 8 Meeting in Braunschweig:

Questions a user might want to address to a “one-stop-shop” mouse information portal regarding mouse phenotypes (starting point: a mouse with a certain phenotype)

- genes related to a phenotype (in mice or any other species)
- Is the phenotype caused by a mutation in only one gene or in several genes?
- Is the underlying mutation located in a coding gene itself or in a promoter?
- Which databases can I use to find relevant information?
- Does the phenotype consist of one or more altered parameters?
- Is the phenotype strain-dependent? Is the observed phenotype a “typical/frequent finding” in mice of that particular background strain?
- Can the phenotype be observed at certain environmental conditions only (e.g. after challenge tests such as a high-fat diet)?
- Has an adequate assay been used to identify the phenotype? What would be an adequate assay for this phenotype? Are there any SOPs, best practices or publications available describing the assay?
- Are there any other phenotypes that frequently co-exist with the known phenotype?
- Are any diseases in mice associated with the phenotype?
- Related literature (see MGI database for mouse genes)
- Which scientist or which institution is working on that particular field or has expert knowledge?
- What is the best search term to look for in a database that describes the phenotype (e.g. obesity / obeseness / adiposity / adiposeness / overweight); do different search terms always have different meanings? Are there any synonyms or antonyms?
- Are there any mutants available (EMMA, cryo banks) or do I have to generate a mutant myself for a gene of interest?
- Are there any mutants available in cryo banks with the same or a similar phenotype (e.g. we have quite a couple of mutants with kinky tails, white belly spots etc. in our cryo archive which might be caused by mutations in different genes)? If so, which gene is mutated in these mutants?
- Are there mutants in cryo banks having the same phenotype with a higher penetrance?
- Are there mutants with the same kind of phenotype showing a higher score (e.g. having a higher cholesterol level in blood based tests)?
- QTL data available?
- Association of phenotype with a mouse chromosome (for mutagenesis screens with random mutagenesis), see RIKEN database
- What would be the “normal range” for a given phenotyping parameter and mouse strain?
- Can the phenotype be observed in a certain period of the live span only?
- If the phenotype is related to a certain organ or anatomical sub-structure, are there any gene expression data for that particular organ/structure available?
- Is the phenotype related to a piece of non-mouse DNA (e.g. a vector) that hasn't been removed after generating a knock-out mouse?
- Is the phenotype a primary or secondary effect (e.g. reduced grip strength due to malformed digits or peripheral neuropathy)
- Sex-specific distribution of the phenotype?
- Are mutants homozygous for the mutation viable? If so, for how long?
- If a mouse with a known mutation in a gene does not show a phenotype, are there other proteins involved in the same pathway that could take over the job?