Recessive Lethality Study

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https://dmdd.org.uk/

The recessive lethality screen was designed to determine when homozygote mouse embryos were surviving until and to explore potential developmental defects.

The screen was performed on embryos at embryonic day (E) 14.5 from homozygous lethal and sub-viable (at post-natal day 14) mouse lines. Outputs include homozygous viability, gross morphology analysis for abnormalities that could impair the survival of any homozygote animals and High Resolution Episcopic Microscopy (HREM) 3D embryonic imaging.

The RL screen was later extended thanks to the Deciphering Mechanisms of Developmental Disease (DMDD) consortium; a UK based effort to provide insight into novel gene function and new mouse models for human developmental genetic disorders. This programme included additional phenotyping tests such as 3D embryo imaging, placental histopathology, transcriptomics and nervous system functionality assessment. The time points analysed were also expanded to encompass embryonic day E9.5 or E18.5, depending on the findings at E14.5 for each line.

At least 28 genotyped embryos (per time point) from Het x Het matings were obtained in order to make a phenotypic call.