

# Viability Primary Screen IMPC\_VIA\_002

## Purpose

To assess the postnatal viability, sub-viability, and lethality of mice of each zygosity and sex during cohort production.

## Experimental Design

- Monitor genotypes of at least 28 live offspring from a specified breeding strategy (e.g. HetXHet, HetXHom). A single breeding strategy should be used per procedure so that totals of pups can be grouped together. If another breeding strategy is used for some litters, please submit a separate procedure, distinguished by sequence id. Record the genotypes of as many live pups as possible; P-values will be deflated by increased numbers of pups, allowing more specific calls to be made.
- Record age of pups at genotype (P1-P28)
- (Optional) Record the age of the youngest and oldest female parents when cohort breeding starts.
- Record raw numbers of live pups of each sex/zygosity combination.
- Identify viability/subviability/lethality of zygosity/sexes by application of a statistical test and P-value threshold. Centres might also apply hard rules (less than 50% of expected pups) to determine whether to put a particular zygosity through the adult pipeline or embryo pipeline.

## Procedure

1. Determine gene category (see Notes below).
2. Select breeding strategy to generate phenotyping cohort for a colony.
3. Genotype all live pups and record numbers of each sex/zygosity. If possible, record pups grouped by litter, along with the parental IDs and date of birth for each litter. Record all three parents for trio matings. Where litter data is not available, group these pups together as the “noLitter” group.
4. Continue to record genotypes of pups for as long as possible. There is no hard minimum limit on numbers, but P-values are more likely to reach significance with greater numbers, and therefore more granular/specific calls can be made (see Notes below).
5. Submit numbers to seriesParameters. Include zeros for all categories where no pups were recorded, even if they were not expected e.g. WT from a HetXHom mating. Do not submit values to the derived total or outcome parameters (see example submission file).
  - a. Where litter data is available, submit each litter as a single increment of all “Number of...” parameters. Include parental IDs and dates of birth where possible. Use a unique identifier for each litter as the incrementValue. Trios matings are indicated by the presence of a second female parentID; otherwise, omit this ID.
  - b. Where litter data is not available, submit pups grouped together as a single increment of all “Number of...” parameters, using incrementValue=“noLitter”. Omit the litter date of birth for this group. “unknown” can be submitted for parentIDs for this group (or any litter where the parent cannot be identified).

6. Where a particular zygosity is identified as lethal (or severely subviable), an alternative may be put through the adult phenotyping pipeline e.g. in the case of homozygous lethality, heterozygotes can be phenotyped.

## Notes

Anzygous is a term to describe the zygosity of a female animal for a gene on the Y-chromosome i.e. no copies of the gene expected. As with all sex/zygosity categories, please report zeros for all litters/colonies where no animals of this type were found, even if they were not expected from that breeding strategy.

Gene category should be available from production labs at each centre. It can be ascertained for most genes by searching NCBI gene. Any gene on a numbered chromosome is autosomal. Genes at the distal tips of the X or Y chromosomes might be pseudoautosomal, rather than X-chr or Y-chr. Contact production labs or DCC data wranglers for assistance.

## Parameters and Metadata

### Gene category IMPC\_VIA\_034\_001 | v1.0

procedureMetadata

Req. Analysis: false

Req. Upload: true

Is Annotated: false

Description: Type of gene inheritance pattern

Options: Autosomal, X chromosome, Y chromosome, Pseudoautosomal,

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### Male parents genotype IMPC\_VIA\_035\_001 | v1.0

procedureMetadata

Req. Analysis: false

Req. Upload: true

Is Annotated: false

Description: Genotype of male parent(s)

Options: WT, Heterozygous, Homozygous, Hemizygous,

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## Female parents genotype IMPC\_VIA\_036\_001 | v1.0

procedureMetadata

Req. Analysis: false

Req. Upload: true

Is Annotated: false

Description: Genotype of female parent(s)

Options: WT, Heterozygous, Homozygous, Anzygous,

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## Litter male parent specimen id IMPC\_VIA\_037\_001 | v1.0

seriesParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

Description: ID of male parent of litter

Increments: Minimum 1

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## Litter female parent 1 specimen id IMPC\_VIA\_038\_001 | v1.0

seriesParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

Description: ID of female parent

Increments: Minimum 1

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## Litter female parent 2 specimen id IMPC\_VIA\_039\_001 | v1.0

seriesParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

**Description:** ID of optional second female parent for trio matings

**Increments:** Minimum 1

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## Litter date of birth IMPC\_VIA\_040\_001 | v1.0

seriesParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

**Description:** Date of birth of litter

**Increments:** Minimum 1

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## Number of WT males IMPC\_VIA\_041\_001 | v1.0

seriesParameter

Req. Analysis: false

Req. Upload: true

Is Annotated: false

**Unit Measured:** count

**Description:**

Number of wild-type males in litter, or total wild-type males if litter data is not available

**Increments:** Minimum 1

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## Number of WT females IMPC\_VIA\_042\_001 | v1.0

seriesParameter

**Req. Analysis:** false

**Req. Upload:** true

**Is Annotated:** false

**Unit Measured:** count

**Description:**

Number of wild-type females in litter, or total wild-type females if litter data is not available

**Increments:** Minimum 1

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## Number of heterozygous males IMPC\_VIA\_043\_001 | v1.0

seriesParameter

**Req. Analysis:** false

**Req. Upload:** true

**Is Annotated:** false

**Unit Measured:** count

**Description:**

Number of heterozygous males in litter, or total heterozygous males if litter data is not available

**Increments:** Minimum 1

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## Number of heterozygous females IMPC\_VIA\_044\_001 | v1.0

seriesParameter

**Req. Analysis:** false

**Req. Upload:** true

**Is Annotated:** false

**Unit Measured:** count

**Description:**

Number of heterozygous females in litter, or total heterozygous females if litter data is not available

**Increments:** Minimum 1

---

## Number of homozygous males IMPC\_VIA\_045\_001 | v1.0

seriesParameter

**Req. Analysis:** false

**Req. Upload:** true

**Is Annotated:** false

**Unit Measured:** count

### **Description:**

Number of homozygous males in litter, or total homozygous males if litter data is not available

**Increments:** Minimum 1

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## Number of homozygous females IMPC\_VIA\_046\_001 | v1.0

seriesParameter

**Req. Analysis:** false

**Req. Upload:** true

**Is Annotated:** false

**Unit Measured:** count

### **Description:**

Number of homozygous females in litter, or total homozygousfe males if litter data is not available

**Increments:** Minimum 1

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## Number of hemizygous males IMPC\_VIA\_047\_001 | v1.0

seriesParameter

**Req. Analysis:** false

**Req. Upload:** true

**Is Annotated:** false

**Unit Measured:** count

**Description:**

Number of hemizygous males in litter, or total hemizygous males if litter data is not available

**Increments:** Minimum 1

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## Number of anzygous females IMPC\_VIA\_048\_001 | v1.0

seriesParameter

**Req. Analysis:** false

**Req. Upload:** true

**Is Annotated:** false

**Unit Measured:** count

**Description:**

Number of anzygous (i.e. no copies of gene as it is Y-chromosome) females in litter, or total anzygous females if litter data is not available

**Increments:** Minimum 1

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## Total WT males IMPC\_VIA\_049\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Unit Measured:** count

**Description:** Total wild-type males

**Derivation:** sumOfIncrements('IMPC\_VIA\_041\_001', 1)

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## Total WT females IMPC\_VIA\_050\_001 | v1.0

simpleParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

Unit Measured: count

Description: Total wild-type females

Derivation: sumOfIncrements('IMPC\_VIA\_042\_001', 1)

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## Total heterozygous males IMPC\_VIA\_051\_001 | v1.0

simpleParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

Unit Measured: count

Description: Total heterozygous males

Derivation: sumOfIncrements('IMPC\_VIA\_043\_001', 1)

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## Total heterozygous females IMPC\_VIA\_052\_001 | v1.0

simpleParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

Unit Measured: count



**Description:** Total heterozygous females

**Derivation:** sumOfIncrements('IMPC\_VIA\_044\_001', 1)

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## Total homozygous males IMPC\_VIA\_053\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Unit Measured:** count

**Description:** Total homozygous males

**Derivation:** sumOfIncrements('IMPC\_VIA\_045\_001', 1)

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## Total homozygous females IMPC\_VIA\_054\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Unit Measured:** count

**Description:** Total homozygous males

**Derivation:** sumOfIncrements('IMPC\_VIA\_046\_001', 1)

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## Total of hemizygous males IMPC\_VIA\_055\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Unit Measured:** count

**Description:** Total hemizygous males

**Derivation:** sumOfIncrements('IMPC\_VIA\_047\_001', 1)

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## Total anzygous females IMPC\_VIA\_056\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Unit Measured:** count

**Description:** Total anzygous (i.e. no copies of gene as it is Y-chromosome) females

**Derivation:** sumOfIncrements('IMPC\_VIA\_048\_001', 1)

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## Total pups IMPC\_VIA\_057\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Unit Measured:** count

**Description:** Total pups

**Derivation:**

sum(sumOfIncrements('IMPC\_VIA\_041\_001', 1), sumOfIncrements('IMPC\_VIA\_042\_001', 1), sumOfIncrements('IMPC\_VIA\_043\_001', 1), sumOfIncrements('IMPC\_VIA\_044\_001', 1), sumOfIncrements('IMPC\_VIA\_045\_001', 1), sumOfIncrements('IMPC\_VIA\_046\_001', 1), sumOfIncrements('IMPC\_VIA\_047\_001', 1), sumOfIncrements('IMPC\_VIA\_048\_001', 1))

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## Total WTs IMPC\_VIA\_058\_001 | v1.0

simpleParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

Unit Measured: count

Description: Total wild-type pups

**Derivation:**

sum(sumOfIncrements('IMPC\_VIA\_041\_001', 1), sumOfIncrements('IMPC\_VIA\_042\_001', 1))

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## Total heterozygotes IMPC\_VIA\_059\_001 | v1.0

simpleParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

Unit Measured: count

Description: Total heterozygous pups

**Derivation:**

sum(sumOfIncrements('IMPC\_VIA\_043\_001', 1), sumOfIncrements('IMPC\_VIA\_044\_001', 1))

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## Total homozygotes IMPC\_VIA\_060\_001 | v1.0

simpleParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

**Unit Measured:** count

**Description:** Total homozygous pups

**Derivation:**

sum(sumOfIncrements('IMPC\_VIA\_045\_001', 1), sumOfIncrements('IMPC\_VIA\_046\_001', 1))

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## **Total males** IMPC\_VIA\_061\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Unit Measured:** count

**Description:** Total male pups

**Derivation:**

sum(sumOfIncrements('IMPC\_VIA\_041\_001', 1), sumOfIncrements('IMPC\_VIA\_043\_001', 1), sumOfIncrements('IMPC\_VIA\_045\_001', 1), sumOfIncrements('IMPC\_VIA\_047\_001', 1))

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## **Total females** IMPC\_VIA\_062\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Unit Measured:** count

**Description:** Total female pups

**Derivation:**

sum(sumOfIncrements('IMPC\_VIA\_042\_001', 1), sumOfIncrements('IMPC\_VIA\_044\_001', 1), sumOfIncrements('IMPC\_VIA\_046\_001', 1), sumOfIncrements('IMPC\_VIA\_048\_001', 1))

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**Female age earliest start** IMPC\_VIA\_026\_001 | v1.1

procedureMetadata

Req. Analysis: false

Req. Upload: false

Is Annotated: false

Unit Measured: Weeks

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**Female age oldest end** IMPC\_VIA\_027\_001 | v1.1

procedureMetadata

Req. Analysis: false

Req. Upload: false

Is Annotated: false

Unit Measured: Weeks

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**Age of pups at genotype** IMPC\_VIA\_030\_001 | v1.1

procedureMetadata

Req. Analysis: false

Req. Upload: true

Is Annotated: false

Unit Measured: Weeks

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## Homozygous males viability IMPC\_VIA\_063\_001 | v1.0

simpleParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

**Description:** Outcome of viability assessment for homozygous males

**Derivation:**

```
viabilityOutcome('homMale', 'IMPC_VIA_034_001', 'IMPC_VIA_035_001',  
'IMPC_VIA_036_001', sumOfIncrements('IMPC_VIA_045_001', 1), sum(sumOfIncrements  
( 'IMPC_VIA_041_001', 1), sumOfIncrements('IMPC_VIA_043_001', 1), sumOfIncrements  
( 'IMPC_VIA_045_001', 1), sumOfIncrements('IMPC_VIA_047_001', 1)))
```

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## Homozygous females viability IMPC\_VIA\_064\_001 | v1.0

simpleParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

**Description:** Outcome of viability assessment for homozygous females

**Derivation:**

```
viabilityOutcome('homFemale', 'IMPC_VIA_034_001', 'IMPC_VIA_035_001',  
'IMPC_VIA_036_001', sumOfIncrements('IMPC_VIA_046_001', 1), sum(sumOfIncrements  
( 'IMPC_VIA_042_001', 1), sumOfIncrements('IMPC_VIA_044_001', 1), sumOfIncrements  
( 'IMPC_VIA_046_001', 1), sumOfIncrements('IMPC_VIA_048_001', 1)))
```

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## Hemizygous males viability IMPC\_VIA\_065\_001 | v1.0

simpleParameter

Req. Analysis: false

Req. Upload: false

Is Annotated: false

**Description:** Outcome of viability assessment for hemizygous males

**Derivation:**

```
viabilityOutcome('hemiMale', 'IMPC_VIA_034_001', 'IMPC_VIA_035_001',  
'IMPC_VIA_036_001', sumOfIncrements('IMPC_VIA_047_001', 1), sum(sumOfIncrements  
( 'IMPC_VIA_041_001', 1), sumOfIncrements('IMPC_VIA_042_001', 1), sumOfIncrements  
( 'IMPC_VIA_043_001', 1), sumOfIncrements('IMPC_VIA_044_001', 1), sumOfIncrements  
( 'IMPC_VIA_045_001', 1), sumOfIncrements('IMPC_VIA_046_001', 1), sumOfIncrements  
( 'IMPC_VIA_047_001', 1), sumOfIncrements('IMPC_VIA_048_001', 1)))
```

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## Heterozygous animals viability IMPC\_VIA\_066\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Description:** Outcome of viability assessment for heterozygous animals

**Derivation:**

```
viabilityOutcome('hetAll', 'IMPC_VIA_034_001', 'IMPC_VIA_035_001',  
'IMPC_VIA_036_001', sum(sumOfIncrements('IMPC_VIA_043_001', 1), sumOfIncrements  
( 'IMPC_VIA_044_001', 1)), sum(sumOfIncrements('IMPC_VIA_041_001', 1),  
sumOfIncrements('IMPC_VIA_042_001', 1), sumOfIncrements('IMPC_VIA_043_001', 1),  
sumOfIncrements('IMPC_VIA_044_001', 1), sumOfIncrements('IMPC_VIA_045_001', 1),  
sumOfIncrements('IMPC_VIA_046_001', 1), sumOfIncrements('IMPC_VIA_047_001', 1),  
sumOfIncrements('IMPC_VIA_048_001', 1)))
```

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## Homozygous animals viability IMPC\_VIA\_067\_001 | v1.0

simpleParameter

**Req. Analysis:** false

**Req. Upload:** false

**Is Annotated:** false

**Description:** Outcome of viability assessment for homozygous animals

**Derivation:**

```
viabilityOutcome('homAll', 'IMPC_VIA_034_001', 'IMPC_VIA_035_001',  
'IMPC_VIA_036_001', sum(sumOfIncrements('IMPC_VIA_045_001', 1), sumOfIncrements  
( 'IMPC_VIA_046_001', 1)), sum(sumOfIncrements('IMPC_VIA_041_001', 1),  
sumOfIncrements('IMPC_VIA_042_001', 1), sumOfIncrements('IMPC_VIA_043_001', 1),  
sumOfIncrements('IMPC_VIA_044_001', 1), sumOfIncrements('IMPC_VIA_045_001', 1),  
sumOfIncrements('IMPC_VIA_046_001', 1), sumOfIncrements('IMPC_VIA_047_001', 1),  
sumOfIncrements('IMPC_VIA_048_001', 1)))
```

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