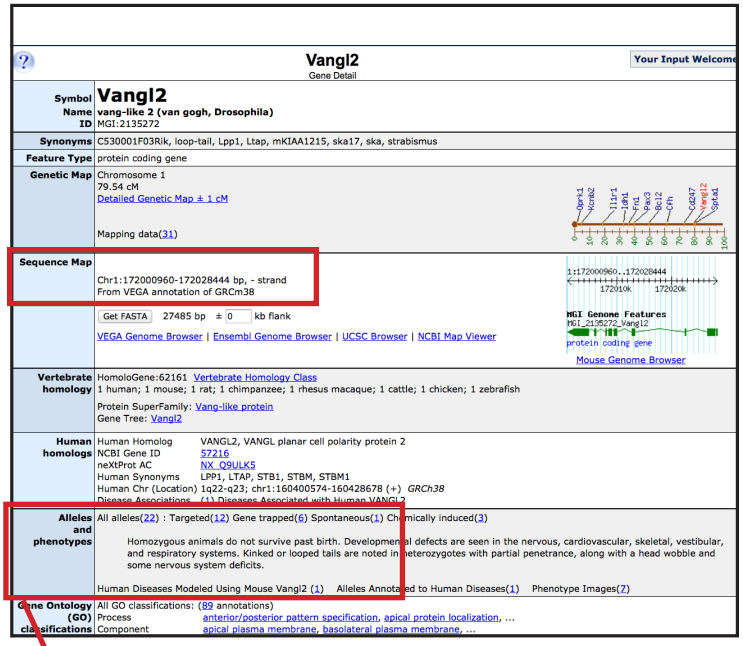
Introduction Module

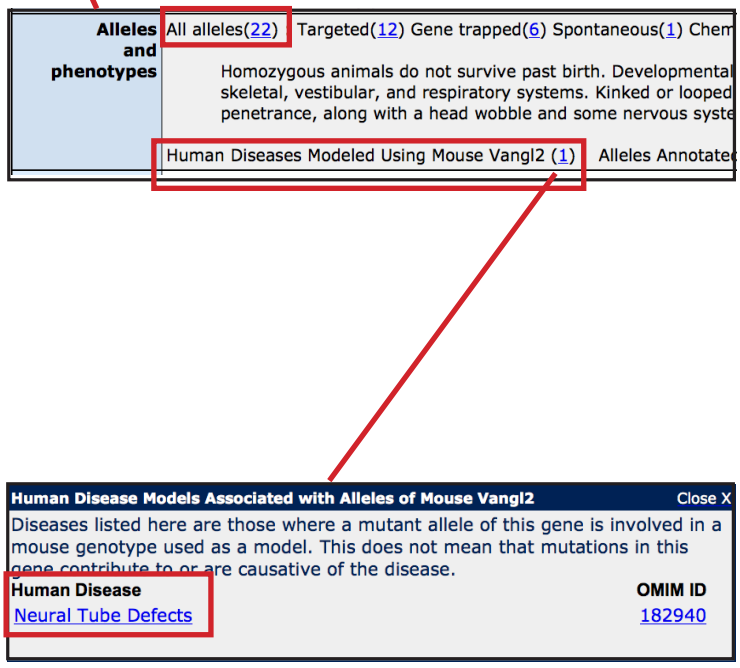
1. On which mouse chromosome would you find the vang-like 2 gene (*Vangl2*)
   - a) Chr 1
   - b) Chr 4
   - c) Chr 10
   - d) Chr 19
   - e) Chr X

Gene positional coordinates can be found in several places in MGI, but notably within the "**Sequence Map**" row on Gene Detail pages. Gene detail pages can be accessed via the Quick Search.



2. How many phenotypic alleles have been described for *Vang2?*
   - a) 1
   - b) 3
   - c) 7
   - d) 14
   - e) 22

Phenotypic alleles are described on Gene Detail pages in the "**Alleles and Phenotypes**" row. For *Vangl2*, the hyperlinked number next to "**All alleles**" is 22. The hyperlinks reading 12, 6, 1 and 3 correspond to specific allele category subsets.

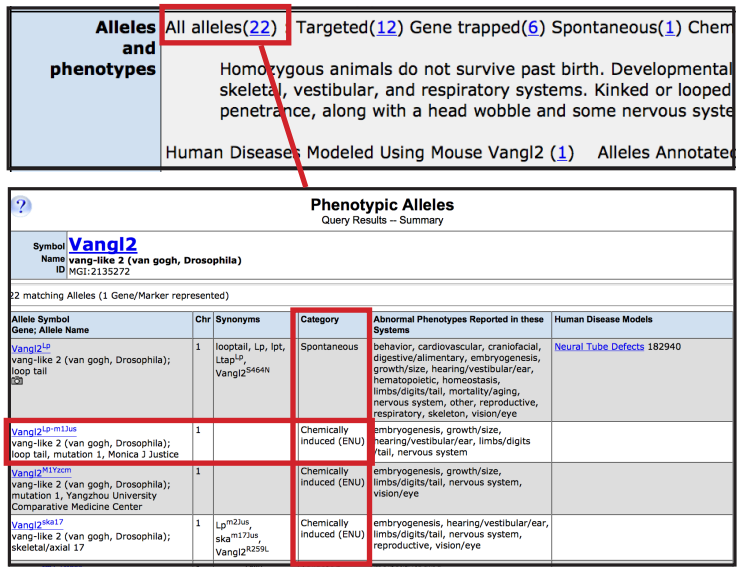


3. Has a mouse mutant of this gene been used to model a human disease? If yes, which one?
   - a) No
   - b) Yes, Neural Tube Defects
   - c) Yes, Cancer
   - d) Yes, Alzheimer Disease
   - e) Yes, polydactyly

At the bottom ot the Alleles and phenotypes row is an annotation for "**Human Diseases Modeled Using Mouse Vangl2 (1)**". Clicking the hyperlinked number in brackets will generate a pop-up for **Human Disease Models Associates with Alleles of Mutant Vangl2** which indicates the specific disease associations(s) - in this case, Neural Tube Defects - along with hyperlinks to MGI's corresponding Human Disease and Mouse Model page(s) and OMIM page(s).

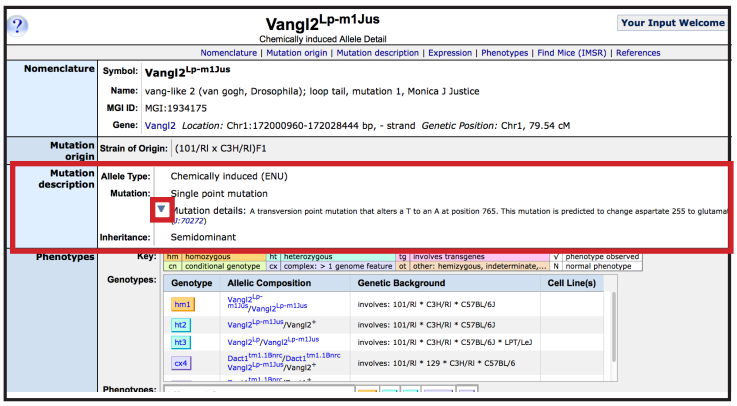
4. What category of allele is *Vangl2*$^{Lp\text{-}m1Jus}$?
   a) Reporter
   b) Gene trap
   c) Spontaneous
   d) Chemically induced (ENU)
   e) Targeted (null/knockout)

Go to the Phenotypic alleles summary by clicking the hyperlinked (22) next to **All Alleles**. Locate the allele symbol *Vangl2*$^{Lp\text{-}m1Jus}$ in the first column, then look to the corresponding information found in the **Category** column. Allele generation type and attributes are also located in the **Mutation details** section of Allele Detail pages.



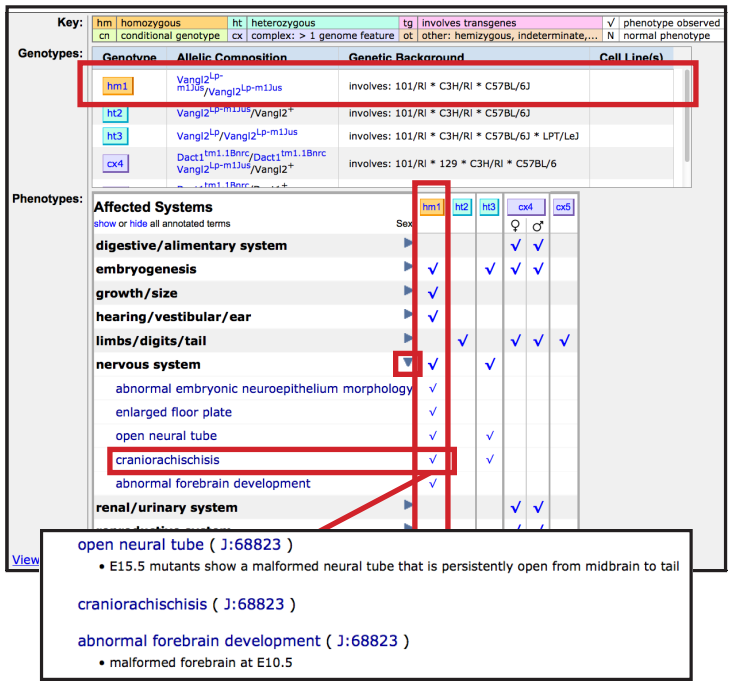
5. What are the mutation details for *Vangl2*$^{Lp\text{-}m1Jus}$?
   a) Exon 1 is floxed
   b) Exon 4 was targeted and replaced by a neomycin cassette
   c) A transversion mutation changing Asp255 to Glu
   d) A stop codon mutation truncating the protien at amino acid 410
   e) The entire gene was deleted

Specific information about an allele's construction can be found in the **Mutation details** row of Allele Detail pages. Click on the triangle toggle ( ) next to **Mutation description** to expand this section and reveal specifics, in this case, an Asp255Glu substitution. A J# will aslo appear indicating the original publication which describes the generation or characterization of the allele. Click on an allele symbol to go to the Allele Detail page.

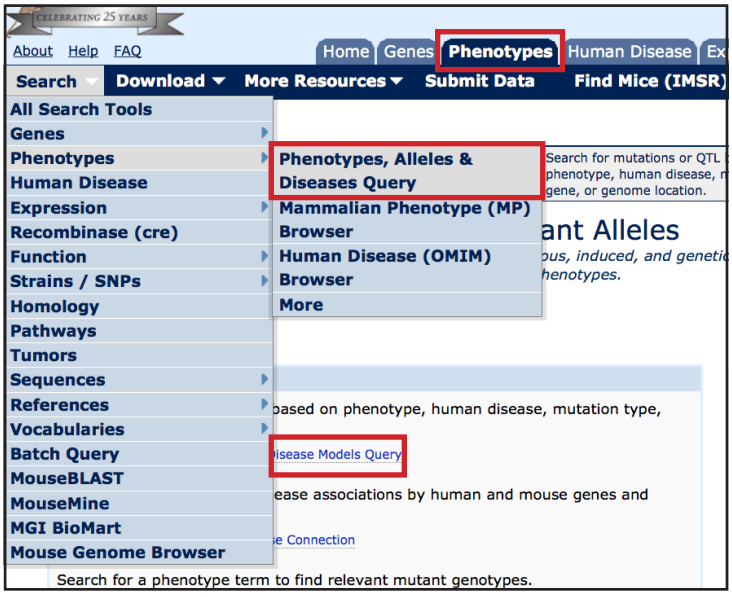


6. Which of the following **nervous system** phenotypes have been observed in mice **homozygous** for *Vangl2*$^{Lp\text{-}m1Jus}$?
   a) craniorachischisis
   b) abnormal synapse morphology
   c) abnormal kidney morphology
   d) decreased Purkinje cell number
   e) no observed nervous system phenotypes

In MGI, phenotypes are annotated to genotypes, rather than alleles (which can have background dependence), or genes (where different alleles may have varied impacts). Find the *hm1* genotype which indicates homozygosity in the **Allele composition** column, click the triangle ( ) next to **nervous system** in the **Affected Systems** table to expand sub-terms and locate check marks in the column which corresponds to genotype hm1. Click on checks or column headers for a free text popup and references for each observation.

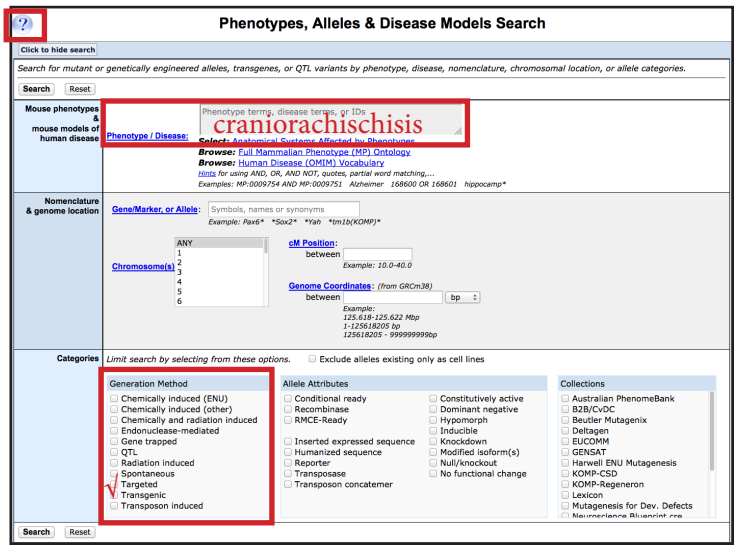


12. Using the **Phenotypes, Alleles & Diseases Query** form, how many targeted alleles have been annotated to *craniorachischisis*?
Hint: prior to May 22nd "Categories: targeted (all)", after May 22nd, "Categories: Generation Method: Targeted"
    a) 1
    b) 3
    c) 13
    d) 15
    e) 35



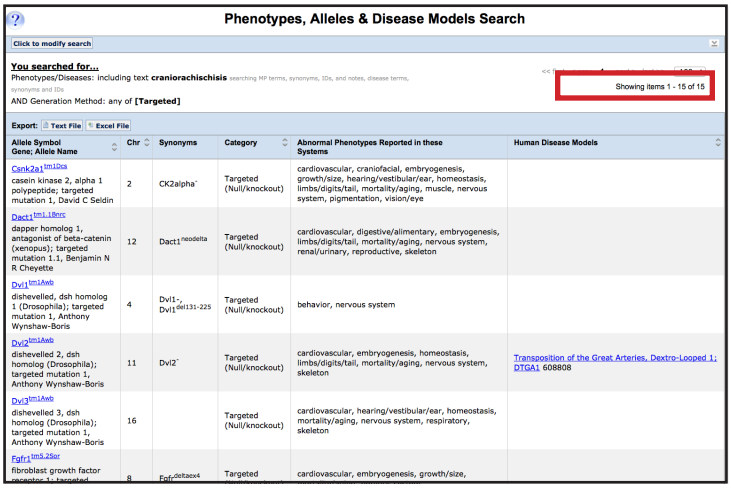
The **Phenotypes, Alleles and Diseases Query** form can be accessed through the options on the **Search** drop down menu, or from the **Phenoypes** (see tab) home page (partially hidden).

On this query form, type "craniorachischisis" into the **Phenotype/Disease** box. Leave the second section ("**Nomenclature & genome location**") blank in this case, but selecting options here can be used to restrict your list, either by genes/alleles which share text similarity (e.g. certain gene families, or lab codes for alleles), or to markers within a defined genomic region.



In the **Categories** section, you can apply filters to select **Generation Method**(s), **Allele attribute**(s) and mutant **Collections**. A link in the help documentation at the top of the page (see question mark icon) provides definitions for each of these terms. Multiple selections can be made if desired, but to answer question 6 above, simply select "*Targeted*" in "**Generation Method**". Click Search to execute the query.

The **Phenotypes, Alleles and Disease Models Search** returns a table listing the alleles that match your selected parameters. Look to the count in the top-right corner of the page, or tally the number of rows, which should come to 15.



For any questions, please contact:

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