Pairnomix

Genetic modeling for patients with a rare condition

Genetic modeling is a process in which a replica of a patient's mutated gene is introduced into human or animal cells growing in a dish or into live animals to model the effects of that gene. This technique is often used to understand the functional consequences of a particular mutation.

What's important to know about genetic modeling is that it usually **does not** require a tissue or blood sample from the patient. As long as the patient has undergone genetic testing to identify their genetic mutation, scientists can custom-build a replica of the gene.

How are these models generated?

- First, a patient's genetic sequencing results are reviewed to understand the specific change(s) in DNA that constitute the mutation.
- Then, a piece of DNA identical to the patient's mutated gene is built in a laboratory.
- Finally, this piece of DNA is inserted into one of two possible "hosts":
    1. Human or animal cells that are grown in a dish *(in vitro)*, or
    2. A laboratory animal *(in vivo)*

Once the piece of DNA is inserted into the cells or into the animal, we'll be able to observe the effect of the mutation on the function of the cells or on the behavior of the animal. This can be seen at the molecular level in the cell lines, and in a change of biological or bodily processes in the animal.

What information can be learned from a genetic model?

After a model of a genetic mutation has been made, its response to a number of different laboratory-based tests is compared against the response of a "wild type" model (a model harboring a normal version of the gene) to those same tests. This comparison allows us to understand what changes are unique to the mutation. Drugs can also be tested against the model harboring the mutation to determine if any compound can improve the dysfunctional response.

What are iPSCs?

Induced pluripotent stem cells, or IPSCs, can be collected directly from a patient or they can be purchased commercially. Within a laboratory setting, these cells can be reprogrammed to have features of other cell types, including brain cells. IPSCs that have been transformed in this way are known as neural stem cells (NSCs), and are capable of serving as a genetic model. For research purposes, scientists can introduce specific genes or genetic mutations into these NSCs, if they are not already present.

Pairnomix empowers patients living with rare disease by performing personalized genetic research that enables physicians to make better-informed healthcare decisions. We're here to answer your questions about genetic research.

Contact us to learn more.

The scientific team at Pairnomix determines which model is best to build for each patient; however, not every gene is able to be modeled with today's technologies.

www.pairnomix.com 844.784.7666

505 Highway 169 North, Suite 850, Plymouth, MN 55441

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