

Geneticists question convention of 'essential' genes

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion and analysis.

Most genes are in our genomes for a reason. If they aren't able to code proteins properly, then we simply won't survive — or will be saddled with the symptoms of any number of serious diseases.

That's been the conventional thought. But geneticists are beginning to question that idea. They're finding more and more people who are perfectly healthy despite having defective copies of a supposedly "essential" gene — and those anomalies aren't just making scientists rethink the workings of the human genome. They may well lead to new million-dollar, life-saving drugs.

"The amazing thing about these variants is that they provide us with a model — in fact, the only model — where we can see in a living human being what happens when a particular gene is inactivated," said Daniel MacArthur, a geneticist at the Broad Institute and Massachusetts General Hospital.

[A study](#), published in the journal Science, reported the newest crop of these rare genetic mutations. By sequencing genetic material from the Pakistani community of East London, in which marriage between cousins is common, the researchers were able to find a higher concentration of these disabled protein-coding genes than in other, more genetically diverse populations.

They even zeroed in on one woman who was lacking a gene that was thought to be essential for human reproduction — and found that she had had three kids.

Read full, original post: ['Essential' genes may not be so essential after all](#)