

Questions on Protas et al.
3-Mar-08

Joo Hyun

What does LOD score imply in this experiment and how do you come up with the number?

Kevin M.

In the first paragraph on page 109, the authors describe the process by which they located the mutations which probably cause albinism. I am a little confused about how they did this. Did they just delete the exons which differed between the two species and look to see what would happen? How did they specifically perform this test?

Emily

On p. 108, the authors say that in order to see if mutations for albinism occur in the same gene or closely linked genes, they did a complementation test between a Molino and a Pachon and found that all offspring were albino. How does this prove that the mutations occur in the same gene? What sort of genetic model would we use to describe something like this?

Jillian

I may have just missed something important, but on page 109, the authors describe how human Oca2 genes are often mutated because they are very large and repetitively coded. Then they admit that "... it is not yet known whether the Astyanax Oca2 locus is similarly characterized by a large size and a large number of associated repeat sequences,..." I wondered how these separate sequences can be named the same thing if they don't know the structure of the Astyanax sequence. Is it just because they cause the same phenotype when mutated?

Max

Is there a particular type of mutation that occurs to produce these phenotypes in cavefish? I know they talk about genomic deletions at the bottom of page 108 in the right-hand column, but is it solely deletions? Why or why wouldn't other types of mutation work and would there be different effects?

Hannah

What is the advantage of having two different areas of deletion in a genotype that cause the same phenotype to be displayed in the same species? (This comes from page 109 where the article describes how exon 21 and 24 are deleted in the two different cave populations, respectively). I guess another way of phrasing this is why does it need to be more complicated to get the same phenotype by two different methods?

Kevin M.

If a trait can mutate independently in several locations along the same gene, what does this say about traits that are polygenetically inherited, such as skin pigmentation? Can several individuals of one phenotype actually have one or more mutations in different locations along different genes? Can individuals with very similar genotypes have vastly different phenotypes? How do Cladists deal with these issues?

Matt

The article by Protas, et al, concludes that the convergence of albinism in cavefish is a result of deletion mutations in the same gene, but at different locations within it. Because these deletions occur partially because of a lack of detrimental effects on the other aspects of the fish's anatomy, does this mean that locations where mutations occurred in the Molino and Pachon populations are mostly random, or are there particular ecological reasons for why each mutation occurred where it did?

Ben

On page 109 the authors state "It is possible in the cave environment, loss of Oca2 function is actually advantageous, for some yet unknown reason". What is a hypothesis that would potentially explain why the loss of Oca2 is advantageous in cave environments.

Kevin C

I'm curious if there are other genes that influence pigmentation or loss of eyes and would also yield viable cave fish albinos if mutated, and is Oca2 favored only because mutations produce few deleterious effects? Also, are there any estimates on how long these cave fish populations have been separated from the surface dwelling ones?

Stephanie

Could you further discuss the difference between coding and regulatory mutations (pg 110)? What kind of range is there in the number of regulatory genes affecting each coding gene? I also don't understand why coding mutations are easier to find, if regulatory mutations "span large areas of sequence."

JP

At the end of the article, the authors describe regulatory mutations. Given the brief description, I understand why regulatory mutations are advantageous because they only alter a gene in a subset of regions. This alteration of specific areas is interesting but I am confused as to how regulatory mutations arise compared to normal mutations which seem to come about randomly?

Kerry

On page 109, Protas et al discuss their examination of another cave population Japonese that has albino individuals. What did the sequencing of Oca2 from one of the albino Japonese show since the albinism phenotype can not be attributed to deletion like the Molino and Pachon populations

Leah

The structure of the paper is not really divided up into the same abstract/introduction/materials and methods/results/discussion sections we are used to. Why did they do that?