February 08, 2019

Evolution & Behavior

Marine mammals may suffer dire consequences of ancient gene loss

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This Break was edited by Reinier Prosee, *Scientific Editor* - TheScienceBreaker

ABSTRACT

Genes encode proteins that perform functions in our bodies, so when we lose genes, we lose the ability to perform their associated tasks. For marine mammals, loss of one gene may leave them especially vulnerable to exposure to widely-used chemicals.

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Have you ever lost something? Perhaps you have misplaced your keys or left an umbrella in the bus. Generally speaking, losing things has negative consequences. Without your keys, you might be locked out of your house or car, and without your umbrella, you might get stuck in the rain. Believe it or not, genes in our DNA can also get lost. DNA is like a cookbook that contains thousands of genes that are recipes for who we are! Our genes typically work by coding for proteins that perform some functions in our body, from keeping our hearts beating to helping us digest food to protecting us from toxins in the environment. Genes can change over time and sometimes they change in such a way that they can no longer be translated into proteins. If this happens, a gene can no longer perform its proper function and we say that the gene is lost, or, more precisely, has undergone loss of function. Unlike losing other types of things, gene loss can have positive, negative, or neutral effects on an organism's health and survival. When gene loss has positive or neutral consequences, the loss can spread throughout an entire species over evolutionary time so that the gene no longer functions in that species. The residual, nonfunctional gene remnant is then called a pseudogene.

Although species-wide gene loss often has no negative consequences when the gene is initially lost, it may have negative consequences later. For example, a species that lives in a consistently cool climate may lose genes necessary for lowering body temperature. If the climate in that species' habitat increases after the gene is lost, the species will be at a disadvantage. Additionally, it is easy to lose a gene and difficult to recover it – just imagine how difficult it would be to retrieve an umbrella you left on a public bus. This means that species suffering from negative consequences of ancient gene loss are unlikely to ever recover these lost genes.

A striking example of the negative modern-day consequences of ancient gene loss is the Paraoxonase 1 (PON1) gene in marine mammals. My lab, led by Dr. Wynn Meyer, discovered PON1's loss of function after developing a method to identify pseudogenes in the DNA of mammals. We looked for genes that were lost in ocean-dwelling mammals, but not terrestrial mammals, and PON1 stuck out like a sore thumb. Not only does PON1 appear to be non-functional in all the marine mammals tested, but it also appears to remain functional in all terrestrial species. Even more striking is the fact marine mammals make up three distinct lineages of mammals that independently returned to the ocean – these are the sirenians (manatees and dugongs), cetaceans (whales, dolphins, and porpoises), and pinnipeds (seals and walruses) – and PON1 was lost in each of these lineages. Such consistent loss across a broad range of species suggests that PON1 may have been lost due to the mammalian transition from land back to the water, perhaps in response to a marine diet or the necessity for long, deep diving, although we don't know for sure yet why PON1 was lost.

Regardless of the reason for PON1's initial loss of function, its absence may have serious consequences for modern marine mammals. The PON1 gene codes for a protective protein – it helps prevent plaque formation in the bloodstream by protecting fat molecules from damage, and, perhaps more importantly to our ocean-dwelling friends, it breaks down organophospate products. Organophosphates are found in agricultural pesticide by-products such as chlorpyrifos oxon and diazoxon, and these pesticides can be found in marine environments near farms due to agricultural run-off. Without functional PON1, marine mammals that live near coastlines may have no way to defend themselves against the negative effects of organophosphate exposure.

To test the ability of marine mammals to break down toxic organophospate products, our collaborators obtained blood samples from marine and terrestrial mammal and tested their ability to break down chlorpyrifos oxonase and diazoxonase. Marine mammal blood showed little to no activity, which indicates that marine species' loss of PON1 has left those species unable to break down organophosphates. These findings are particularly alarming in light of the fact that chlorpyrifos has been found in waters off the Florida coastline and organophosphate pesticides have been found along the coast of Australia, waters frequented by at-risk manatees and dugongs, respectively.

Our findings show a striking pattern of ancient PON1 gene loss in marine mammals with potentially severe modern-day consequences – and unlike your misplaced keys, PON1 cannot be regained by just looking in your back pocket. Marine mammals' unique genetics puts them at particular risk of harm from man-made products, and if we want them to continue to survive and thrive, we must consider their specialized needs when protecting their habitats.