Short telomeres a hallmark of genetic cardiac disease, Stanford researchers find

By now many people are familiar with the concept that telomeres — the protective caps on the ends of chromosomes — serve as a kind of aging clock that counts down the lifespans of each of our cells. But research indicates that telomere length also plays a role in diseases like Duchenne muscular dystrophy — an inherited muscle-wasting disease that often ends in heart failure.

Now microbiologist and immunologist Helen Blau, PhD, and Alex Chang, PhD, an instructor of cardiovascular medicine and of microbiology and immunology, have found that the telomeres in certain heart cells of people with genetic heart diseases called cardiomyopathies are 25 to 40 percent shorter than those of healthy controls or unaffected relatives.

They published their research in Proceedings of the National Academy of Sciences.

From our release:

Although it’s not yet known whether the stunted telomeres directly affect the function of the cardiomyocytes [a heart cell responsible for contraction] or arise as a result of heart failure, the finding opens the door to an intriguing line of research and drug discovery. It also may one day allow researchers and clinicians to identify people at risk for heart failure due to cardiomyopathy.

The findings dovetail with those of a previous study from Blau’s laboratory showing that people with Duchenne muscular dystrophy also have abnormally short telomeres, and that
telomere shortening activates a DNA damage response that harms the cells’ mitochondria and interferes with the cells’ ability to pump blood effectively.

Blau, who is also the Donald E. and Delia B. Baxter Foundation Professor and director of the Baxter Laboratory for Stem Cell Biology, explained:

The shortening of telomeres in cardiomyocytes appears to be a reliable hallmark of cardiac failures that arise due to genetic defects. [...] Now we can study this phenomenon in the lab in real time and start to ask questions about cause and effect.

We’d love to know, for example, how this shortening might impact the DNA damage response, mitochondrial dysfunction and cell-death pathways. It opens up a whole new line of investigation.