Student correctly guesses mystery disease on Netflix series 'Diagnosis'

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Lilian Fung, a third-year medical student at VCU, is featured in "Diagnosis," a new documentary series on Netflix. Credit: Virginia Commonwealth University

Lilian Fung was taking a break from studying when she came across a column in The *New York Times Magazine* called "Diagnosis" that

described hard-to-solve medical mysteries.

The column, written by Lisa Sanders, M.D., tells a patient's story and crowdsources diagnoses from readers. Fung, a third-year medical student in the Virginia Commonwealth University School of Medicine, submitted the correct diagnosis for the column she read. Sanders' column is the basis for a new Netflix documentary series, and Fung is featured in the first episode.

Fung spoke with VCU News about the show, her studies, and correctly identifying the mystery in Sanders' column.

How did you learn about the show?

It was another routine night of studying, with randomly scheduled mental breaks where I would either pause to browse my social media feed or watch another 20 minutes of "The Great British Baking Show."

I happened to scroll past a New York Times article where the caption caught my eye. I opened the article and it told the story of Angel and her undiagnosed condition. It was a thorough retelling of her medical history that, to my surprise, even included her medical records. The end of the article encouraged readers of all backgrounds and experiences to give their input on what Angel's diagnosis could possibly be.

So, I guess you can say it was luck and chance that that title happened to catch my eye!

To be quite honest, I think luck played a big part in me guessing correctly. This New York Times article happened to be published when I was still in my preclinical years so rarer diseases were not long forgotten.

What makes this different from my experiences in the hospital is that it

can often become overwhelming to learn from bits and pieces of information, from different people and places, and then come up with the important questions to ask a patient as well as a game plan for their hospital stay. The fact that the details of Angel's symptoms were compiled into one organized story so that I could mull over it for a long period of time (instead of studying for class) undeniably played a big part as well.

Season 1 of "Diagnosis" aired on Netflix in August. Fung is featured in the first episode.

Can you give a general overview of Angel's condition?

In Angel's case, notable symptoms included muscle pain, aches and fatigue that developed after exercise. On multiple emergency department

visits, she had elevated creatine kinase levels in her blood, which is a marker of muscle breakdown. She also had dark-colored urine, a finding that suggests either increased red blood cell breakdown (due to pigmentation from a red blood cell oxygen-binding protein called hemoglobin) or increased muscle breakdown (due to a protein related to hemoglobin found only in muscles called myoglobin).

However, her other lab tests were not consistent with characteristics commonly observed with increased red blood cell breakdown, therefore making myoglobin the more likely candidate for darkened urine.

## What is carnitine palmitoyltransferase 2 and how did you come to guess Angel had a CPT2 deficiency?

Once the body begins to run out of sugar, it turns to fats as an energy source. CPT2 is one of the molecules involved in the pathway of transporting fatty acids (a type of fat molecule) into energy-generating structures inside our cells called mitochondria. If CPT2 is not functional, it can result in muscle breakdown as our muscles run out of an energy source.

Thinking back to when I was debating between McArdle disease or CPT2 deficiency, I remember scrolling through various websites looking for symptoms that could help make a distinction. One thing I considered was the timing of symptom onset after beginning exercise. In a description of one of her "attacks," she had completed a hike up to a waterfall before she began experiencing muscle pain and weakness. My thought process was that if it were McArdle's, Angel would have experienced symptoms much earlier in the timeline considering how strenuous that activity was.

In addition, one of the features of McArdle's was the "second wind

phenomenon" where after first experiencing the muscle pains and fatigue an individual would experience improvement of symptoms as increased blood flow provides fats to the muscles. Just based on the article alone, there was no such description of a "second wind phenomenon" occurring with Angel's attacks. Hence, I decided to go with CPT2 deficiency for my submission.

How did your experience at VCU prepare you to answer the question correctly?
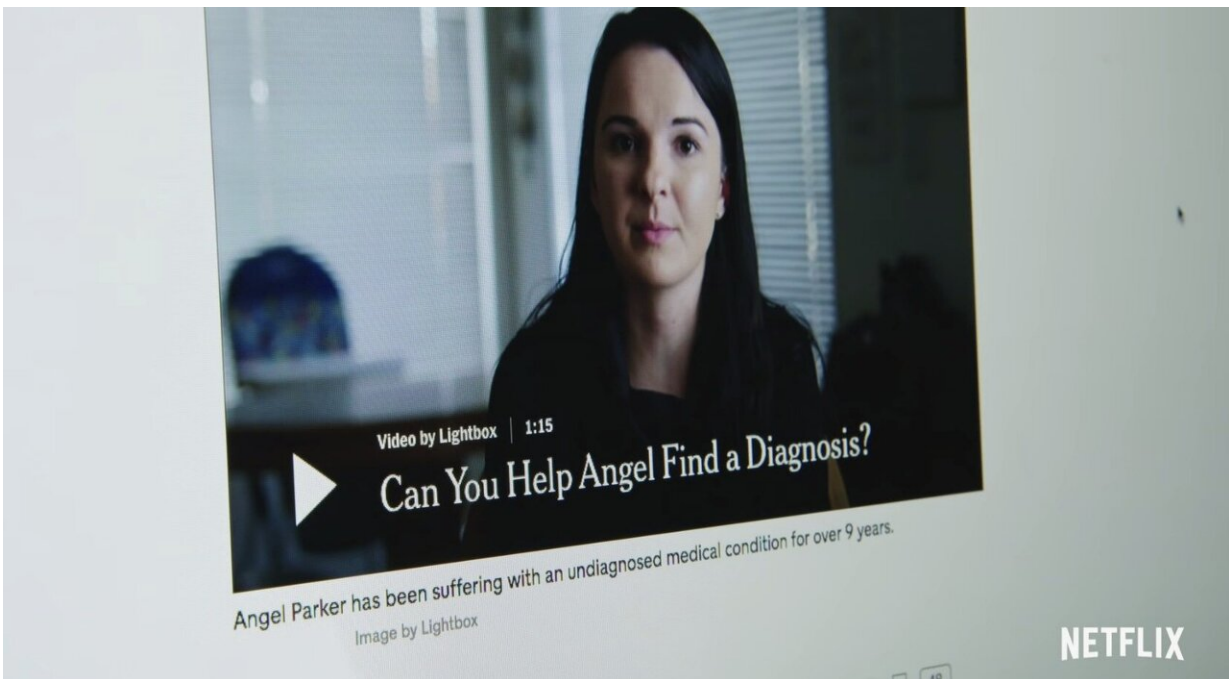
Fung was taking a break from studying when she came across a column in The New York Times Magazine called "Diagnosis" that described hard-to-solve medical mysteries. Credit: Netflix

The way I formed my initial thoughts was based on these questions:

What disease would first present itself in an otherwise healthy child and continue to young adulthood? When does she experience her symptoms? What are the symptoms? What do the labs suggest and do they match up with the pathophysiology of potential diseases?

This is essentially the same thought process that VCU medical students are taught in our first year and something that we continue to refine as we progress further in our medical career.

During our preclinical years, we have the chance to practice our clinical skills with standardized patients who are actors that play the role of a patient, as well as with small groups under the guidance of an attending doctor and a fourth-year medical student.

It was during these sessions where we learned to ask and think about important aspects of the patient history that could guide us to form a "differential diagnosis," where we formulated a list of possible conditions that an individual might have based of the information we gathered. Key things to consider include type of symptoms, timing of symptoms, potential inciting events, etc.

What excites you the most about learning medicine?

One thing that excites me the most about medicine is how dynamic it is, and as a result, I am learning every day from my peers, residents, attendings and my patients. I remember when I began my third year on my neurology rotation, and I saw how the (long and detailed) neurologic exam, which I had previously practiced on standardized patients or my friends, actually told me a lot about a patient's condition. It was a new and exciting feeling to see how material that we spent hours studying actually became important and applicable to someone's life. I find it fascinating to think about how all the body systems interact in a healthy individual as well as how various diseases disrupt these processes.

What I have truly come to treasure is the impact we as health care workers can have on a stranger's life, no matter how big or small. It may be as simple as taking a few minutes of your day to say hello and chat to make their hospital stay a little less mundane.

Provided by Virginia Commonwealth University