Tackling Medical Mysteries with Next-Generation Testing

"Exome sequencing" is faster, more cost-effective than genetic testing

UCLA Health

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NEWS PACKAGE

SUGGESTED TEASE	COMING UP, MEET A MOM WHO WENT ON A MEDICAL MISSION TO FIND ANSWERS FOR HER SON - AND THE DOCTORS WHO WERE DETERMINED TO HELP HER. THEIR STORY NEXT IN HEALTH NEWS.
ANCHOR LEAD	IMAGINE THE FRUSTRATION OF HAVING A RARE CONDITION THAT PUZZLES DOCTORS. EXPERTS ORDER TEST AFTER TEST, TRYING TO FIND ANSWERS - ONLY TO COME UP EMPTY. UNFORTUNATELY, SOME PATIENTS SPEND *YEARS* WAITING FOR THE RIGHT DIAGNOSIS. NOW, IN AN EFFORT TO SOLVE THOSE MEDICAL MYSTERIES, DOCTORS ARE TURNING TO THE NEXT GENERATION OF TESTING. CLARK POWELL HAS DETAILS.,
(PACKAGE START) ---------- **CG: Courtesy: UCLA Health** Shots of Audrey & Calvin at home Still photos of Calvin as a baby Shot of Audrey talking to Dr. Nelson **CG: Audrey Lapidus** **Son had mysterious condition** Shots of Audrey & Calvin with Dr. Nelson	(Nats – Sound mom & son playing) :02 DEEP DOWN, AUDREY LAPIDUS (la-PEED-us) KNEW SOMETHING WAS WRONG. AS A BABY, HER SON CALVIN WASN'T REACHING NORMAL MILESTONES LIKE ROLLING OVER OR CRAWLING. FOUR SPECIALISTS AND BATTERIES OF TESTS LATER, IT WAS STILL A MYSTERY. UNTIL ONE DAY, SHE SAW A DOCTOR AT UCLA HEALTH WHO OFFERED A GLIMMER OF HOPE. :14 *"Just, very fortuitously, as I was pushing for more testing, our geneticist said, 'If you can wait one more month, we're going to be launching this brand new test called the exome.'"* :11 WITH THAT, CALVIN WAS THE FIRST PATIENT TO UNDERGO A TYPE OF GENETIC TEST KNOWN AS EXOME SEQUENCING AT UCLA HEALTH. DISEASES LIKE CALVIN'S ARE SO RARE, DOCTORS

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Shots from lab / microscope	HAVE TO RELY ON GENETIC TESTING TO TRY AND FIND CLUES TO HIS CONDITION. IN THE PAST, THOSE TESTS WERE DONE ONE GENE AT A TIME, WHICH WAS EXPENSIVE.
Shots of exome sequencing	BUT THIS POWERFUL NEXT-GENERATION TECHNOLOGY ALLOWS DOCTORS TO ANALYZE MORE THAN 20-THOUSAND GENES AT ONCE - AT A FRACTION OF THE COST. :21
CG: Stanley Nelson, MD UCLA Health	*"Oftentimes we're now turning to exome sequencing -- sequencing every single protein-coding gene in one fell swoop-- as the go-to diagnostic test"* :10
CG: Julian Martinez, MD UCLA Health	*"Sequencing is like doing a spell check on your genes and making sure that there are no mistakes or spelling mistakes that could be causing problems with your health"* :09
Shots of Dr. Nelson & Calvin	SOON AFTER EXOME SEQUENCING - WHICH ONLY REQUIRES A BLOOD SAMPLE FROM THE PATIENT - CALVIN WAS DIAGNOSED WITH A RARE CONDITION KNOWN AS PITT-HOPKINS SYNDROME. :08
CG: Audrey (CG'd earlier)	*"The diagnosis gave us a point to move forward from rather than that really scary no-man's land of not knowing"* :08
Shots of Audrey giving Calvin medication Shot of Calvin walking	ARMED WITH THAT INFORMATION, CALVIN GOT A HEAD START ON THERAPY, AND IS NOW MAKING STRIDES THAT SEEMED UNLIKELY NOT LONG AGO. AT UCLA HEALTH, THIS IS CLARK POWELL REPORTING. :10
(PACKAGE END) ----------	---
ANCHOR TAG	WHILE EXOME SEQUENCING CAN PINPOINT THE ROOT OF MANY RARE CONDITIONS, DOCTORS NOW WANT TO MOVE TO WHOLE GENOME SEQUENCING IN PATIENTS. THAT WOULD ALLOW THEM TO ANALYZE *THREE BILLION* DIFFERENT COMPOUNDS IN A PATIENT'S GENETIC MAKEUP.

SOCIAL MEDIA

Share it! Suggested tweet:	Experts @UCLAHealth investigate medical mysteries with next-generation testing. http://bit.ly/2rd0DMW
Suggested post:	Experts at UCLA Health are working to uncover clues in medical mysteries by using next-generation testing. Diagnosing rare diseases requires genetic testing, which,

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	in the past, was done one gene at a time and was very expensive. But *exome* sequencing being done at UCLA can test more than 20,000 genes at once, which can lead to faster, more accurate diagnoses. Details: http://bit.ly/2rd0DMW

EXTRA BITES

CG: Julian Martinez, MD **UCLA Health**	Dr. Martinez talks about exome sequencing cost effectiveness: *"What exome sequencing allows us to do is, in a very cost effective way, look at all the thousands of genes that we have in one sample."* Dr. Martinez on future implications of exome sequencing: *"We may find the genetic change that we know predisposes autism and that leads families to get referrals and get a diagnosis of autism for the first time."* Dr. Martinez on benefits of exome sequencing: *"The ability of doing this test is that it can allow us to not only anticipate, but also identify the causes for many symptoms individuals may present with that have gone unexplained for a very long period of time."*
CG: Stanley Nelson, MD **UCLA Health**	Dr. Nelson on exactness of exome sequencing: *"You can sequence all of the protein-coding part of the genome for just a few thousand dollars, interpret it really powerfully and search across all 7,000 possible disease-causing genes in one test."* Dr. Nelson on importance of early diagnosis: *"It's very impactful when you're able to give clarity of diagnosis. If you have a child with a serious, unexplained medical condition, it's incredibly anxiety-provoking for the parents and the whole medical team."* Dr. Nelson on taking out the guesswork: *"The clarity when you give the specific molecular diagnosis has a couple of huge ramifications and one is, instead of kind of being in a big vague cloud of other patients kind of similar to what the physician has seen, instead you're saying, this is the exact disease and there's literature on them."*
CG: Audrey Lapidus **Son had mysterious condition**	Audrey talks about her mission to find answers for Calvin: *"We were really lucky to just be in the right place at the right time and get our information when we did, and also had great doctors at UCLA who knew that we needed to keep pushing and keep trying to get an answer."*

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CG: Audrey Lapidus **Son had mysterious condition**	Audrey talks about finally getting a diagnosis: *"It gives us a roadmap of you know, of where to go and what's realistic as far as therapies and treatments and things like that."*

References

[1]*Clinical exome sequencing in neurologic disease*, **Neurology Clinical Practice, Volume 6 Number 2, April 2016**. Online: http://cp.neurology.org/content/6/2/164.abstract

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