

# GET TO KNOW MOLECULAR DIAGNOSTIC TESTING

Molecular diagnostic tests detect DNA or RNA in fragments in **patient samples** that indicate a genetic condition or infection.

Blood, saliva, urine



## FREQUENTLY USED TESTS



Polymerase Chain Reactions (PCR)



Transcription Mediated Amplification (TMA)



Loop-Mediated Isothermal Amplification (LAMP)



CRISPR-based diagnostics

## WHAT CAN A MOLECULAR TEST TELL US?

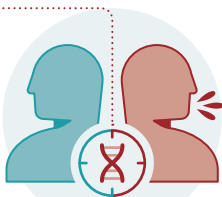
Whether the target DNA/RNA is present in a patient sample



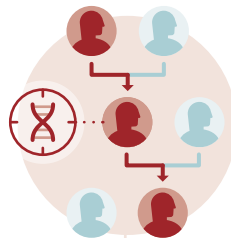
## HOW CAN A MOLECULAR TEST BE USED?

To detect infection in an individual, whether or not they present symptoms

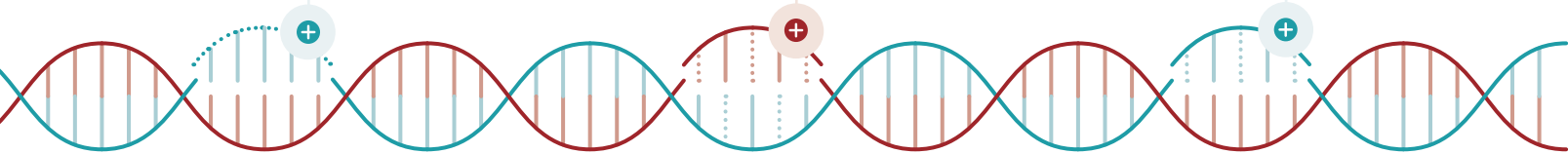
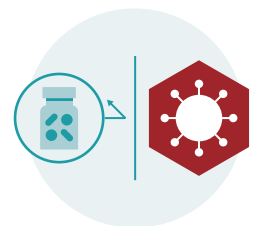
Provides *true positive* results



To identify genetic mutations linked to hereditary or acquired conditions



To detect genetic mutations associated with resistance to therapeutics (antibiotics)



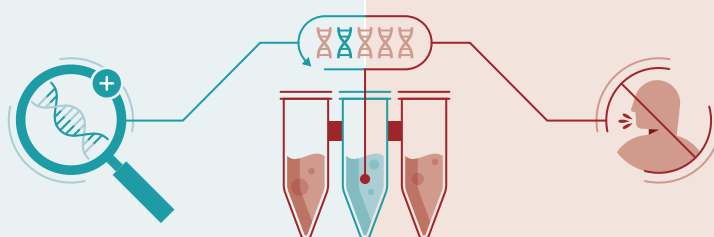
## A MOLECULAR DIAGNOSTIC TEST

Means

Does not mean

That a target pathogen or genetic mutation is present

Even at extremely low levels.



That a patient is necessarily **actively** infectious or ill

In some cases, tests may detect DNA/RNA from inactive or dead pathogens.

# A CLOSER LOOK: HOW PCR TESTS WORK

One of the most commonly used molecular diagnostic methods is **Polymerase Chain Reaction (PCR)** testing.

A lab technique used to “amplify,” or copy, a targeted gene sequence from a test sample, creating millions of copies for analysis.

## KEY ELEMENTS



**Template DNA**  
Purified DNA or RNA from a patient sample (e.g., blood, saliva, urine).



**Primers**  
Short single strands of DNA that tag the target sequence.



**DNA polymerase**  
Enzyme that assembles new strands of DNA from nucleotides.



**Ct value**  
The number of cycles needed to detect a target gene sequence.

## STEPS

1

### DENATURATION

Template DNA splits into two separate strands.

2

### ANNEALING

Primers bind to each end of the target DNA sequence, if present.

3

### EXTENSION

DNA polymerase replicates the target segment of DNA, creating two new double-stranded DNA sequences.

4

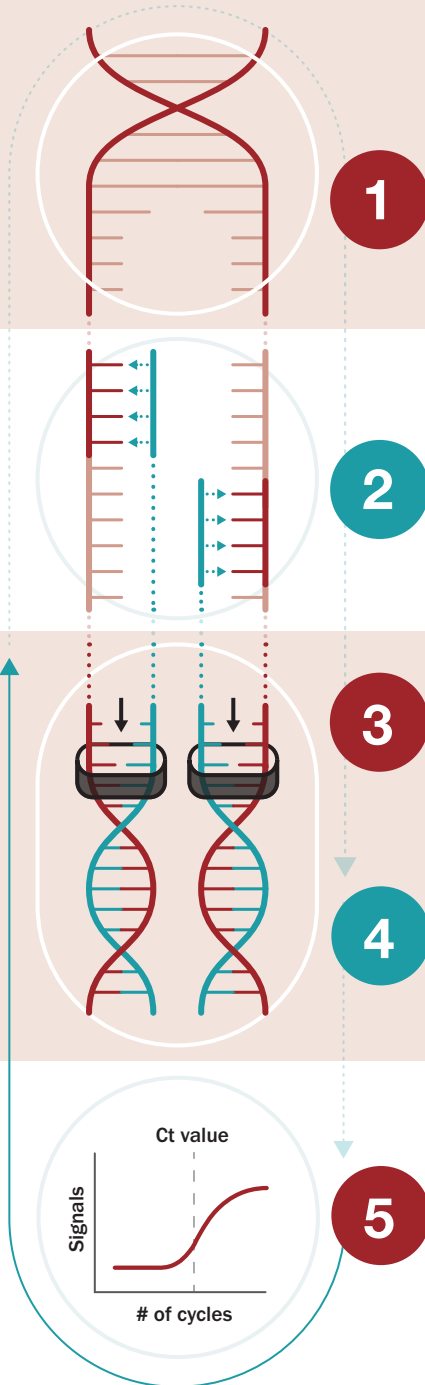
### CYCLING

If the target is present, the process repeats to create many copies each of which emits a signal. Once enough copies are made, a strong enough signal is produced to be detected.

5

### DETECTION

Once enough copies are made, a strong enough signal is produced to be detected.



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Contact us at: 7700 Wisconsin Avenue, Suite 1000 Bethesda, MD 20814 | 240.485.2745 | [www.aphl.org](http://www.aphl.org)

### Sources:

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- **National Human Genome Research Institute:** [www.genome.gov/about-genomics/fact-sheets/Polymerase-Chain-Reaction-Fact-Sheet](http://www.genome.gov/about-genomics/fact-sheets/Polymerase-Chain-Reaction-Fact-Sheet)

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