Rare mutation could lead to the next blockbuster cholesterol drug

The following is an excerpt.

When Sharlayne Tracy showed up at the clinical suite in the University of Texas (UT) Southwestern Medical Center in Dallas last January, the bandage wrapped around her left wrist was the only sign of anything medically amiss. The bandage covered a minor injury from a cheerleading practice led by Tracy, a 40-year-old African American who is an aerobics instructor, a mother of two and a college student pursuing a degree in business. “I feel like I’m healthy as a horse,” she said.

Indeed, Tracy’s well-being has been inspiring to doctors, geneticists and now pharmaceutical companies precisely because she is so normal. Using every tool in the modern diagnostic arsenal — from brain scans and kidney sonograms to 24-hour blood-pressure monitors and cognitive tests — researchers at the Texas medical centre have diagnostically sliced and diced Tracy to make sure that the two highly unusual genetic mutations she has carried for her entire life have produced nothing more startling than an incredibly low level of cholesterol in her blood. At a time when the target for low-density lipoprotein (LDL) cholesterol, more commonly called ‘bad cholesterol’, in Americans’ blood is less than 100 milligrams per decilitre (a level many people fail to achieve), Tracy’s level is just 14.

Read the full article here: Genetics: A gene of rare effect