

## Blindness gene comes to light

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Western Cape scientists, working with their counterparts in Texas, have discovered the gene that causes one of the most common forms of blindness.

Now that they know where the gene is, scientists can begin work on a drug to act specifically on it.

The scientists used Silicon Valley analysis techniques to mine the human genome and identify the gene.

Retinitis pigmentosa affects approximately one in 4 000 people globally. The gene, known as RP1, that contains the mutation responsible for the disorder, has remained elusive for more than 20 years, according to two papers published in the July issue of Nature Genetics.

"By finding a gene on the genome that causes a disease, you know that if you `break' it you get that disease. Now we can start working on finding a cure," says Winston Hide of the South African National Bioinformatics Institute at the University of the Western Cape (UWC), who identified the gene. "It's really cool. In five to 10 years a cure could be emerging, instead of an infinite length of time."

The search for the gene is a tale befitting a Hollywood movie. It involves all the classic thrillers of modern science: information technology, biotechnology, genomics, supercomputers, a human disease, technology transfer and positive growth at UWC.

The human genome is all the genetic material in a human cell. The genome is made up of genes, which determine characteristics ranging from eye colour to the propensity to develop certain diseases.

Many of the world's geneticists are involved in the human genome project aimed at discovering which genes are responsible for which characteristics. This also helps combat illnesses caused by genetic defects or mutations. Only 30 000 of the 100 000 genes in the human genome have been identified.

What Hide's team has done is to generate a virtual human genome, compiled from thousands of human genes in databases around the world created by other geneticists.

Hide was contacted by Lori Sullivan of the University of Texas Health Science Centre, who was working on eye diseases and had spent the last eight years looking for the RP1 gene.

Hide found the electronic version of the gene, or one very like it, in his virtual model and a UWC database helped characterise and define it. Both Hide and Sullivan published their work in Nature Genetics.

While Hide was working at identifying the gene, scientist Eric Pierce of the Children's Hospital in Boston, United States, working independently, revealed that mutations in a gene encoding a photoreceptor protein cause the RP1 form of retinitis pigmentosa.

While computers are now used for all studies of human genetics, they have become increasingly useful for handling the vast amounts of information in human genome research.

The South African National Bioinformatics Institute, which uses a Silicon Graphics Origin 2 000 supercomputer, is dedicated to the use of computational genomics for the study of human diseases. It has developed an electronic library of expressed human genes that is used at institutions throughout the world, including Harvard and Cambridge universities and the Pasteur Institute in France.