

Protein that limits the severity of genetic kidney disease found

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Researchers from Kumamoto University, Japan have identified a protein that limits the severity of Alport syndrome, a type of genetic kidney disease. The finding can provide clues that point toward new therapeutic approaches for Alport syndrome.

Genetic disease, a generic term covering many diseases, is caused by mutations in genes. Several forms of <u>genetic diseases</u> produce serious symptoms, and the development of new therapies is highly desired.

The current fundamental treatment method for genetic diseases has been gene therapy, where specific genes are directly targeted to treat the disease. This, however, has not shown sufficient clinical usefulness.

Methods for creating iPS cells have recently been established, and with them new genetic therapies using a patient's own cells have been receiving attention. Mutant genes within cells taken directly from a patient could be corrected, and the repaired cells are proliferated before being placed back into the patient.

These breakthrough therapies, however, are not without their issues. The amount of time to establish the required technology may be significant, or the treatment might require enormous cost to the patient.

Some teams of researchers began focusing their attention on the progression of a disease state, rather than attempting to find a cure.



Most genetic diseases are progressive. Although symptoms may not appear soon after birth, they usually emerge at some point during a patient's lifetime. The research team from Kumamoto University, Japan focused on the point at which symptoms started to progress to inhibit the development of a specific genetic disease.

Alport syndrome is a progressive genetic kidney disease with high risk for kidney failure, which requires dialysis treatment or kidney transplant

"We put our finger on a key protein of Alport syndrome, p53, which has an important function related to decelerating the progression of the symptoms in the kidney," said Prof. Hirofumi Kai, who led the research.

p53 is well known as a gene that inhibits cancer, and drugs are clinically available to activate this protein. The team found that the p53 protein maintains the integrity and formation of cells that filter urine from the blood. They also found that the activity of p53 <u>protein</u> is significantly reduced when the symptoms of Alport syndrome are progressing.

"Our research shows that <u>p53 protein</u> is an important molecule to decelerate the progression of Alport syndrome," said Prof Kai. "Recovering the function of the <u>p53 gene</u> will help to inhibit the progression of <u>symptoms</u>. We also expect this breakthrough finding to develop new treatment strategies for genetic diseases similar to Alport <u>syndrome</u>."

The findings were published in the January 2016 issue of the *Journal of the American Society of Nephrology*, one of the top journals for <u>kidney</u> studies, and featured photos from this study on the cover.

More information: R. Fukuda et al. Podocyte p53 Limits the Severity of Experimental Alport Syndrome, *Journal of the American Society of*



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Provided by Kumamoto University

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