# Article information:

A founder COL4A4 pathogenic variant resulting in autosomal recessive Alport syndrome accounts for most genetic kidney failure in Romani people
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9948603/>

# Article summary:

1. Romani people have a high prevalence of kidney failure, and this study examined a Romani cohort for pathogenic variants in the COL4A3, COL4A4, and COL4A5 genes affected in Alport syndrome (AS).

2. The study found that a founder COL4A4 pathogenic variant resulting in autosomal recessive AS accounts for most genetic kidney failure in Czech Romani people.

3. The two identified founder variants contribute to the high prevalence of kidney failure in Czech Romani, with an estimated population frequency of autosomal recessive AS from these variants and consanguinity by descent at least 1:11,000 in Czech Romani.

# Article rating:

May be slightly imbalanced: The article presents the information in a generally reliable way, but there are minor points of consideration that could be explored further or claims that are not fully backed by appropriate evidence. Some perspectives may also be omitted, and you are encouraged to use the research topics section to explore the topic further.

# Article analysis:

The article titled "A founder COL4A4 pathogenic variant resulting in autosomal recessive Alport syndrome accounts for most genetic kidney failure in Romani people" provides important insights into the high prevalence of kidney failure among the Romani population in the Czech Republic. The study examines a cohort of 57 Romani individuals with clinical features suggestive of Alport syndrome and their family members to identify pathogenic variants in the COL4A3, COL4A4, and COL4A5 genes that are associated with AS.

The study finds that 19% of the Romani cohort had autosomal recessive AS caused by a homozygous pathogenic c.1598G>A, p.Gly533Asp variant in COL4A4 or a homozygous c.415G>C, p.Gly139Arg variant in COL4A3. The p.Gly533Asp variant was associated with a more severe phenotype than p.Gly139Arg, with an earlier age at end-stage kidney failure and more macroscopic hematuria. Microscopic hematuria was very common in heterozygotes with both variants.

The article provides valuable information on the genetic basis of AS among the Romani population and highlights the need for genetic testing for individuals with persistent hematuria. However, there are some potential biases and limitations to consider.

Firstly, the study only examines a small cohort of Romani individuals from the Czech Republic, which may not be representative of all Romani populations worldwide. Additionally, there is no comparison group of non-Romani individuals with AS to determine if these variants are specific to the Romani population or if they occur more widely.

Secondly, while the study identifies two pathogenic variants associated with AR AS among the Romani population, it does not explore other potential genetic or environmental factors that may contribute to kidney failure among this population. For example, socioeconomic factors, access to healthcare, and environmental toxins may also play a role in the high prevalence of kidney failure among the Romani population.

Finally, the article does not provide information on potential risks or limitations associated with genetic testing for AS. For example, false positives or negatives may occur, and there may be psychological or social implications for individuals who receive a positive diagnosis.

In conclusion, while the article provides important insights into the genetic basis of AS among the Romani population in the Czech Republic, it is important to consider potential biases and limitations in interpreting these findings. Further research is needed to explore other factors that may contribute to kidney failure among this population and to evaluate the risks and benefits of genetic testing for AS.

# Topics for further research:

* Socioeconomic factors and kidney failure in the Romani population
* Environmental toxins and kidney disease in the Romani community
* Genetic testing for Alport syndrome: risks and benefits
* Prevalence of Alport syndrome in non-Romani populations
* Psychological and social implications of genetic testing for kidney disease
* Treatment options for Alport syndrome and end-stage kidney failure

# Report location:

<https://www.fullpicture.app/item/29567a9ddeb886e0e75bcd83f98c086a>