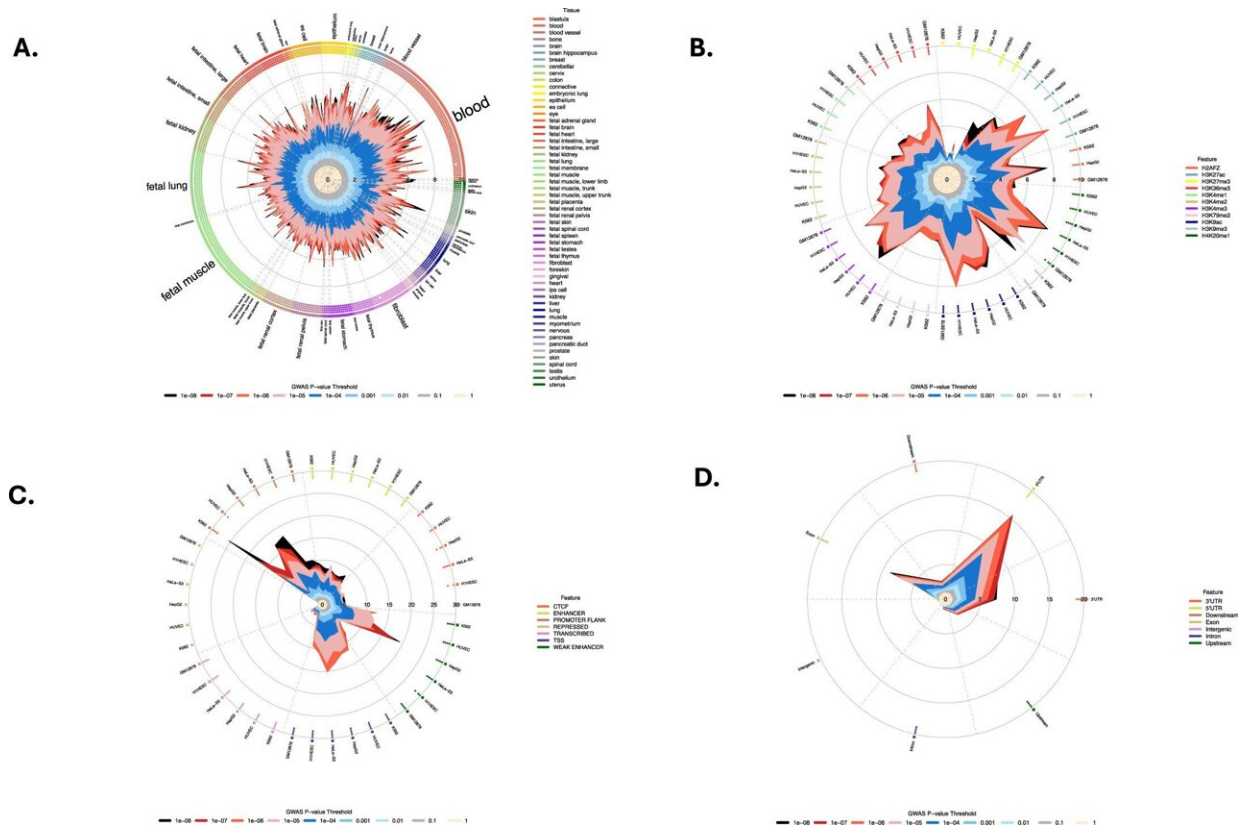# Analysis identifies 50 new genomic regions associated with kidney cancer risk

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In silico analysis of enrichment for putative regulatory annotations among kidney cancer loci. Credit: *Nature Genetics* (2024). DOI: 10.1038/s41588-024-01725-7

In a new analysis of genetic susceptibility to kidney cancer, an international team of researchers has identified 50 new areas across the

genome that are associated with the risk of developing kidney cancer. These insights could one day be used to advance our understanding of the molecular basis of kidney cancer, inform screening efforts for those at highest risk, and identify new drug targets.

The study was led by scientists at the National Cancer Institute (NCI), part of the National Institutes of Health (NIH). The research is published in the journal *Nature Genetics*.

A previous genome-wide association study (GWAS) of people of European ancestry identified 13 regions of the genome that are associated with kidney cancer risk. However, the study population was not diverse.

To identify additional regions, researchers conducted a GWAS in participants of many different genetic ancestries that included 29,020 people with kidney cancer and 835,670 people without kidney cancer. Analysis of the data, which came from published studies, biobanks, and a new study, resulted in the identification of 50 new regions associated with the risk of developing kidney cancer, bringing the total number of such regions to 63.

Among the newly identified genetic variants were several associated with a risk of developing papillary renal cell carcinoma, the second most common subtype of renal cell carcinoma. Another variant, in the VHL gene, was common in individuals of African ancestry and was associated with an estimated three times higher risk of developing clear cell renal cell carcinoma, the most common type of kidney cancer.

Finally, the researchers used the study data to develop a measure of an individual's overall risk of developing kidney cancer, known as a polygenic risk score, that can be combined with established risk factors—such as high blood pressure, smoking, and a high body-mass index

measurement—to possibly enable earlier detection of the disease.

More information: Mark P. Purdue et al, Multi-ancestry genome-wide association study of kidney cancer identifies 63 susceptibility regions, *Nature Genetics* (2024). DOI: 10.1038/s41588-024-01725-7

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