Traveller Genes Newsletter

TRAVELLER GENES

Scottish Travellers: New Insights into Your Genetic Heritage and Health

A Special Newsletter for the Scottish Traveller Community and Supporters

In this issue, we share the findings from our ground-breaking study of the Scottish Traveller community and spotlight Traveller Genes researcher, Ashwini Shanmugam.

The research is dedicated to the Scottish Traveller Community. We thank everyone who contributed to the study.

***"When yellow's on the broom, when yellow's on the broom, I'll tak' ye on the road again when yellow's on the broom."* — Adam McNaughton**

[Traveller Genes Homepage]

"I enjoyed working with the Irish Traveller community and have always been interested in the Scottish Travellers. So, when I was approached by the community requesting a genetic study, it was easy to agree, and we mapped out the aims together. I'm delighted to finally have the results, which show that the Scottish Travellers have a distinct gene pool of their own, with deep indigenous origins in Scotland. We have also uncovered important information on genetic health risks faced by the population, which clinicians should be made aware of."

- Prof Jim Flett Wilson, University of Edinburgh

“For far too long, the history and culture of Scottish Travellers have been defined by outsiders and their many theories. With myself and other Travellers collaborating with Jim and his team, we embarked on this DNA journey which shows us what we have always argued: that our roots are in Scotland and always have been.”

- Samantha Whyte Donaldson, Scottish Traveller

"When I look in a mirror, I see my reflection. Now with the Traveller Genes study, I gain a strong sense of history and I feel that those who shared my bloodlines are looking back at me. I feel our sense of place and therein lies our story.”

- Jess Smith, Scottish Traveller

Scottish Travellers are a unique community with a rich oral history, culture, and traditions. Recognised as a distinct ethnic group, Travellers have long contributed to Scotland’s story, but have also faced social isolation and discrimination. Nowadays, the majority of Travellers live in houses, with some living permanently on sites or continuing to shift (travel) when the better weather arrives.

There has been next to no scientific research into their genetic origins, population structure, or health risks related to inherited diseases. The Traveller Genes study was initiated in partnership with the Scottish Traveller community to address these gaps and improve representation in genetic research.

Community and Cultural Context

Scottish Travellers have a strong culture, including unique languages and traditions. They were recognised as a distinct ethnic group in 2008. The community has faced centuries of discrimination, including harsh laws, forced settlement, and social exclusion. Socioeconomic disadvantage is widespread, with high rates of poverty, low educational attainment, and poor health outcomes compared to the general Scottish population. Life expectancy and general health is significantly lower, and the community was disproportionately affected by COVID-19.

Sparse coverage in historical records means there are conflicting narratives regarding Traveller origins, with proposals ranging from European Romani to indigenous Scots, or even Picts.

Why was this study done?

For the first time, Scottish Travellers partnered with our researchers to explore their genetic ancestry and health risks. The aims were to:

- Work out if the Scottish Travellers are genetically distinct
- Discover where Scottish Travellers come from

- Understand how closely related they are to other Traveller groups and settled Scots
- Describe any population subgroupings
- Identify any inherited health risks that might be more common in the community

Until now, nothing was known about the genetic background of Scottish Travellers. The community has often been cautious about taking part in scientific studies because of past discrimination. The community was involved in designing the research to ensure respect and trust.

Public Involvement Panel

What did our researchers do?

Nearly 150 people with Traveller backgrounds volunteered for our study and donated a saliva sample. Our researchers extracted and analysed the DNA and compared it with people from Scotland, Ireland, and England, as well as European Roma. They looked for patterns in the DNA to see how closely related these groups are, and to find any rare genetic changes that might cause disease.

Study Design and Participant

- The study recruited 148 participants with Traveller grandparents, mainly Scottish Travellers, between 2021 and 2022.
- DNA samples were collected and analysed using state-of-the-art methods, including genome-wide array genotyping, whole-exome and mtDNA sequencing.
- Participants were categorised based on their grandparental ancestry (Scottish Traveller, Highland Traveller, Lowland Traveller, Irish Traveller, English Gypsy, or Mixed Traveller).
- Scottish Travellers (including Highland and Lowland Travellers) were compared with settled Scots, settled Irish, settled English, settled Welsh, Irish Travellers, English Gypsies, and European Roma.

What Did Our Study Find?

Family and Community Ties

Study of family trees revealed a small number of common surnames, high levels of surname sharing, and inter-marriages between Traveller families. The seven most common surnames (Stewart, McPhee, Williamson, Townsley, Reid, MacDonald and Whyte) together account for 58% of the reported Scottish Traveller grandparents.

Traveller families are spread across Scotland from Shetland to Roxburghshire, but with concentrations in Aberdeenshire, Perthshire, Caithness, Ross-shire, the Western Isles and Argyll.

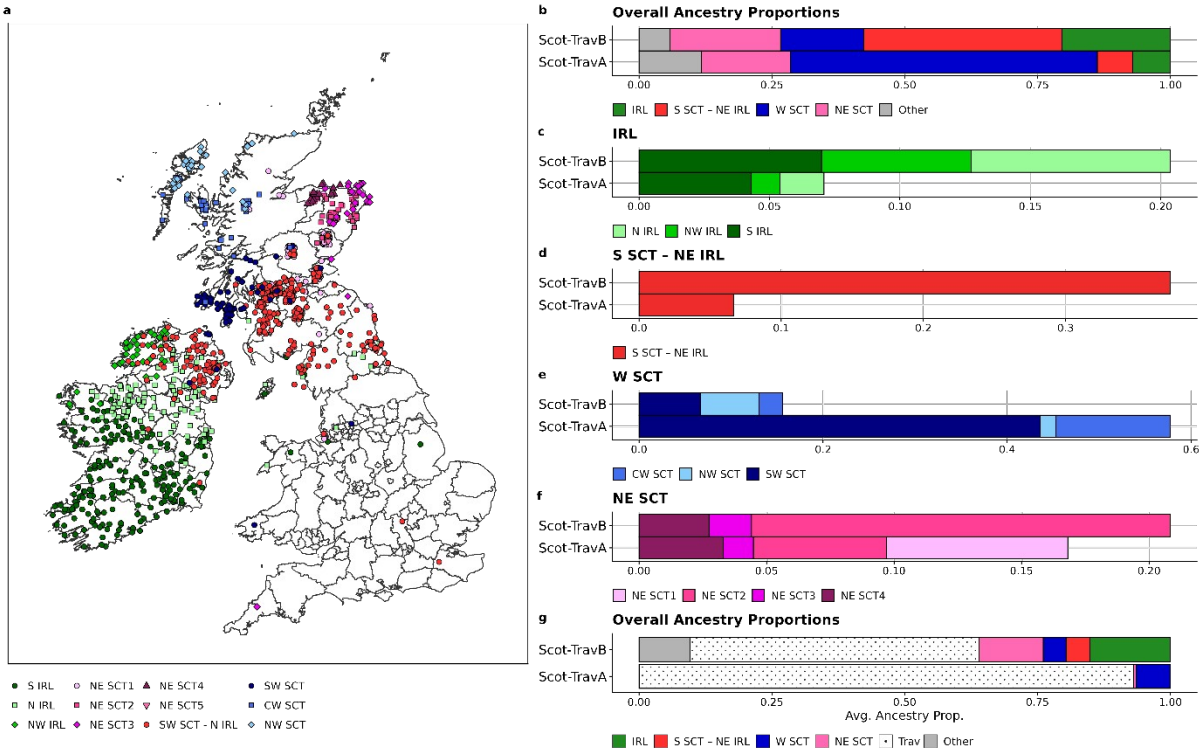
A few surnames are associated with specific regions, e.g., Stewart with Aberdeenshire and Williamson with Caithness. There is some fluidity in how individuals were identified as Highland, Lowland, or Scottish Travellers.

A Unique Genetic Identity

We used genome-wide data to make comparisons with settled Scottish, Irish, English and Welsh, as well as European Roma. Scottish Travellers were shown to be genetically distinct from the settled populations, as well as from Irish Travellers, English Gypsies, and European Roma. They share deep ancestry with the settled Scots, while the Irish Travellers were related to the settled Irish, and the English Gypsies, while related to the settled English, also had some links to the Roma people.

Two main genetic subgroups were identified within the Scottish Traveller population, called ***Scot-TravA*** and ***Scot-TravB***. Group A is more genetically distinct than group B, which while also clearly a Scottish Traveller group, shares more genetics with the settled Scots. ***Scot-TravA*** is in fact as distinct from the settled Scots as Italians are!

Looking in more detail at the ancestry of the two subgroups, we can see that ***Scot-TravA*** is closest genetically to western Scotland and northeast Scotland, while ***Scot-TravB*** has connections to a group prominent in southern Scotland and Northern Ireland, as well. The distribution of ancestry among these two groups suggests that ***Scot-TravA*** probably reflects Highland Traveller ancestry, while ***Scot-TravB*** likely represents Lowland Traveller ancestry.



Demographic History

The cousin marriage seen in the study of family trees was also reflected in the patterns of genetic sharing, similar to that seen for Punjabis from Pakistan and Tamils from Sri Lanka. This was stronger for the ***Scot-TravA*** subgroup than for ***Scot-TravB***. Patterns in the DNA also preserved evidence of a *population bottlenecks*, that is severe reductions in the size of a population at some point in history. The bottlenecks looked to be stronger than those in the history of the Orkney and Shetland islands.

Maternal Lineages

We studied mother-line lineages using a piece of DNA that is inherited from mother to daughter to granddaughter down the generations (called mtDNA). We can identify ancient genetic lineages – which are given names using letters and numbers – and work out where in the world they come from. Complete mtDNA sequences were studied from 91 individuals who identified their mother's mother as a Traveller. Notably, none of the mtDNA lineages typically associated with the South Asian origin of the European Romani people were present in the Scottish Traveller samples. Instead, almost all of the lineages were of European origin.

Notable among these was an mtDNA lineage known as T2f1a1 found among Scottish, Highland and Lowland Travellers, and which accounted for fully 24% of the maternal lines! This was shared between ***Scot-TravA*** and ***Scot-TravB*** subgroups and was particularly prevalent among Travellers with Central or Northern Scottish grandmothers. There is a Traveller-specific branch of this lineage, and it appears to originate in Scotland.

Seven other lineages (J1c2e, H16, K1c1b, U4b1b1, V2, J1b1a1a and U1a1a3) were also seen multiple times in the Scottish Traveller sample, and together with T2f1a1, they sum to 64% of the Scottish Traveller mtDNA pool. Genetically isolated populations often show this effect, where a small number of ancestral founder lineages account for a large proportion of the population, for example, the Iberian Roma (65%) and Ashkenazi Jews (75%).

The majority of mtDNA lineages have identical or near-identical matches in Scotland and thus likely a local Scottish origin, consistent with the genome-wide analysis. However, a different pattern is seen for the U1a1a3 and V2 lineages, which both have southern or eastern European matches (including Near Eastern for U1a1a3: Assyrian and Mazanderani) and no matches in the British Isles, perhaps preserving evidence of an ancient Mediterranean contribution to the Scottish Traveller community.

Two different lineages in the small sample of English Gypsies showed evidence of Roma origins, while the Irish Traveller mtDNAs were of European heritage.

Genetic Health Risks

Inherited disorders are caused by small changes in our DNA, for instance when one letter in the genetic code is accidentally changed to another one when it is being copied for the new generation. These changes are called variants and they happen all the time. The great majority of them have no effect, but if the variant occurs in an important gene, it can increase the risk of disease. These variants are then inherited down the generations, along with the rest of our DNA. That is why some diseases run in families. Most disease-causing genetic variants are very rare, but sometimes they can become more common. One example of this is in genetically isolated communities, where many people in the population descend from shared ancestors hundreds of years ago. If one or more of those ancestors had a genetic variant, then it is possible for many of their descendants today to also carry it.

The strong signals of genetic isolation seen in the genetic analyses predict that variants of medical importance may be found at different frequencies in the Scottish Traveller community. We therefore carried out a survey of variants which can cause serious disorders. We found no Travellers with variants in the *BRCA1* gene (causing breast and ovarian cancer), or *BRCA2* (causing breast, ovarian and prostate cancer), or *LDLR* (causing inherited high cholesterol), or *TTN* (causing the heart condition cardiomyopathy). However, variants were seen in the *HFE* gene, which can cause the iron-overload disorder haemochromatosis and also in *BTD*, which causes a metabolic disease leading to a deficiency of the vitamin biotin. Both of these are also common among settled Scots.

We then went on to investigate genetic variants which don't cause disease unless you inherit two copies of them, one from both your mother and your father – these are called *recessive* variants. If you carry only one copy, it does not affect you, but you are a carrier and if you have children with another carrier, we would expect one quarter of them to be affected by the disease. We observed the common northwest European *CFTR* variant which causes the lung disease cystic fibrosis, but also several others which are extremely rare in the settled population. Five of them in particular were much more common in Scottish Travellers than in the general population, including:

- A *PIEZO1* variant causing a disorder of the lymphatic system (the lymph nodes and vessels), which is important for fluid drainage and immune function
- A *CRB2* variant causing the kidney disease glomerulosclerosis
- A *CANT1* variant causing dysplasia or abnormal development of the cartilage
- A *HOGA1* variant causing a metabolic disease with high oxalate in the urine, which can lead to kidney stones and other kidney problems

The four variants above are over 1000 times more common in the Scottish Travellers. This enrichment of disease-causing variants in the Scottish Traveller community increases the risk of babies being born with these otherwise very rare disorders. Similar findings, but involving different variants, have been made by others for Irish Travellers. At the same time, it is important to realise that the majority of Scottish Travellers are not carriers for these conditions.

Doctors and other health workers who care for Scottish Travellers should be made aware of the increased risk of these conditions in the community. As genomic medicine enters healthcare, in order to prevent further disparities from arising, the opportunity should be taken to engage regarding a bespoke community-based reproductive carrier screening programme for the Scottish Travellers, similar to J-netics. J-netics is a charity organisation in the UK, whose purpose is to prevent and diagnose 47 predominantly Jewish disorders, and raise awareness about them. Jewish individuals and couples can be screened to check they are not both carriers of the same variant, to allow them to make informed reproductive decisions.

Why does this research matter?

This is the first genetic study of the Scottish Traveller community.

The results provide valuable insights into their unique genetic history and health risks. Scottish Travellers have an ancient Scottish heritage and so can take their place along with other indigenous non-Romany Traveller groups in Europe. The study emphasises the importance of respectful, community-led research and the potential benefits of tailored genetic screening. The findings could inform public health strategies and policies to address health inequalities among Scottish Travellers. It also shows the importance of including all communities in health research, so that no group is left out or disadvantaged.

What's next?

The findings highlight the need for discussions about community-driven genetic screening, which could help identify health risks early and improve health outcomes for Scottish Travellers. Continued partnership with the community is essential to ensure research is respectful and beneficial. Clinicians who care for Scottish Travellers should be made aware of the scope for otherwise rare genetic diseases to be more common than is usually the case in Scotland.

In summary

The Scottish Traveller community is genetically distinct from the Irish Travellers, English Gypsies and European Roma, as well as from the settled populations of the British Isles. Despite strong signals of genetic isolation, an indigenous Scottish origin can be inferred. Two subgroups were identified, probably reflecting Highland Traveller and Lowland Traveller ancestry. The isolation and within-community marriage have led to an increased risk of certain inherited diseases. Our study, conducted in partnership with the community, provides a foundation for improving health outcomes and reducing disparities through targeted genetic research and healthcare.

For more information or to get involved in future research, please contact the study team.

Click on the link below to read the original research paper published in Nature Communications:

https://www.nature.com/articles/s41467-026-74969-y

Researcher Spotlight: Dr Ashwini Shanmugam

Ashwini is a Postdoctoral Researcher at the Royal College of Surgeons in Ireland (RCSI) and part of the Traveller Genes research team.

What's your research focus?

My research focussed on understanding how the history of a community has shaped their current genetic variation. I specifically study populations with Irish and/or British ancestry. Through characterising the genetic relationships between groups of people, we can better understand changes in demographic history — such as population size, evidence of migrations, and how isolated populations have been. This both reveals insights into our history, and where our genetic health factors come from.

What made you interested in pursuing a career in health research?

I worked at as a clinical research scientist for a few years at a precision diagnostics company. While I was there, I saw the genetic tests we developed for cancer helped guide treatment decisions for patients and how this changed their prognosis. This crystallised my resolve to work in health research. Population genetics gives me an opportunity to study to take a step back and look at disease at a population-level perspective.

How will Traveller Genes volunteer data support your research?

This study represents a unique opportunity to help understand the history of an under-represented group, and how that has shaped their genetic diversity. This is a great opportunity to use genetic insights to complement oral and community histories in a way that wouldn't be possible in other studies.

Tell us about the most interesting research you've worked on.

It is a project that I am currently working on that looks at polycystic kidney disease (PKD) in the Irish population. We are trying to find if PKD patients with the same disease-causing mutation have a common ancestor, and using the genetic data from them, if we can predict other carriers of the same mutation in the dataset. It is interesting because we are leveraging population genetics methods to study how and when a disease-causing mutation came into being within a population.

Meet the Team

Contact Us

Our research team is based at the University of Edinburgh, in the Usher Institute.

If you ever have any questions, you can email us at TravellerGenes@ed.ac.uk.

Your Data Privacy

We want to make sure you're aware of how we protect your data when conducting our research. For more information about how we use your data and keep it safe, please see our Privacy Policy at https://traveller-genes.ed.ac.uk/privacy-notice, or let us know if you'd like to have a copy posted to you.