

Ancestry, race and ethnicity

A simplified quick guide

Genomics contexts

Genetic ancestry

✓ **Visible in a genome**

Shows where genomic variants have come from, which can be helpful for determining someone's risk of disease.



You need to take a genetic test to know your ancestry. At Genomics England, we follow a standard for grouping ancestry by five larger reference groups, sometimes called "super populations"

European

African

South Asian

East Asian

American

My genome has substantial West African ancestry which links to Sickle Cell Disease.



Social contexts

Race

✗ **Not visible in a genome**
(though inferences can sometimes be made)

A context-specific identity term, now widely agreed to be a socio-political construct.



Commonly used racial categories in the UK include:

White

Brown

Latina/Latino and Arab

Black

South East Asian

I would call myself mixed race (Black and White), but I am often assumed to be White by others because of my lighter skin tone.



Ethnicity

✗ **Not visible in a genome**

Refers to a person's specific cultural, religious and social background.



Commonly used ethnic categories in the UK include:

White British*

Indian*

Syrian

Nigerian Yoruba

Taiwanese

* note that only two of these are options on the current UK census

My ethnicity is Jamaican British, because I feel like this best captures the cultural activities and groups that I align with.



What are the problems with getting this wrong?



Ethnicity and race don't capture the complexity of ancestry

Sometimes, the culture or language you identify as does not correspond to the culture or language of your genetic ancestors. However, the DNA they have contributed is still relevant for analysing disease, which is why a genomic test is needed alongside ethnicity.



In the Individual's genome:
Traces from Southern Africa and West Africa

Their race:
Black African

Their self-reported ethnicity is:
Botswana Bantu (Tswana)

What this means:

Someone with family in Botswana who defines their ethnicity as Tswana may have admixed ancestry from West Africa (Bantu-related) and South Africa (Khoe/San related), which might not be captured in how their ethnicity is defined.



You cannot be 100% certain of someone's race and ethnicity based on their ancestry, and assuming so affects how we group people for research

There are strong links between ancestry and ethnicity and race. Because of these strong correlations, many researchers believe that they can guess race based on ancestry markers, but this ignores the very personal and socio-political context surrounding ethnicity (how you define culturally) and race (what society tends to impose on you).



In the Individual's genome:
Traces from areas in South Asia, Pakistan

Their race:
South Asian

Their self-reported ethnicity is:
Indian

What this means:

Population labels inferred using your DNA may conflict with your personal identity. This may be particularly true for ethnic groups from neighbouring regions, across whom individuals can share many recent genetic ancestors. Using an individual's inferred genetic ancestry as a ground-truth risks reducing the importance of the impact that shared environment or culture linked to their ethnicity can have on disease outcomes.



The social context surrounding how a patient is perceived affects the quality of the genomic medicine services they receive

Sometimes, a patient or participant's ethnicity is assumed by their clinician or healthcare professional. Researchers risk a reductive approach to research engagement - they might contact an ethnic group that they believe is best related to an ancestry-related disease, but are actually engaging with the wrong group.



In the Individual's genome:
West African traits linking to sickle cell

Their race:
Mixed Black and White

Their self-reported ethnicity is:
Mixed - Jamaican and English

What this means:

This person has lots of West-African ancestry traits linking to sickle cell. This can be common for people with Jamaican ethnicity. In healthcare settings, some healthcare professionals assume this person's race is white, because of their lighter skin tone, and this can affect the type of care they receive. This would not be captured in our data but is important to bear in mind as it shows how our science is impacted by social contexts.