Project Details	
Project Code	PHS27Br Paternoster
Title	Using data from diverse populations to explore the genetic causes of skin disease
Research Theme	Population Health Sciences
Project Type	Dry lab
Summary	The Skin Genetics Consortium (www.skingeneticsconsortium.org) is conducting genome-wide association studies (GWAS) in >4 million individuals to identify genomic regions associated with ~100 skin conditions. The inclusion of diverse genetic ancestries presents both challenges and opportunities for discovery. This PhD will use large-scale data analysis tools to extract biological insight from these results. Multi-ancestry fine-mapping will refine causal variants at each locus, while integration of multi-omic data, including gene expression and proteomic, will help identify the effector genes and underlying biological pathways. These findings are expected to support equitable development of new drug targets for a wide range of skin conditions.
Description	Background In 2025, the World Health Assembly designated skin diseases as a Global Public Health Priority [1]. Despite previous large-scale genetic studies identifying variants associated with common conditions, such as eczema, psoriasis and acne - including work from our group in Bristol [2], the majority of skin conditions remain genetically underexplored. In addition, most existing studies are heavily biased towards individuals of European ancestry, limiting the generalisability of findings to global populations. The Skin Genetics Consortium (SGC, co-led by the University of Bristol, King's College London and NYU Langone) is addressing this gap by conducting genome-wide association studies (GWAS) across ~100 skin conditions using large globally representative biobanks (>4million individuals). This effort substantially expands ancestral diversity and will generate GWAS summary statistics by the end of 2026. Recent advances in statistical genetics, combined with rapidly growing multi-omic resources, now enable more powerful interpretation of GWAS signals. However, many existing post-GWAS methods are designed primarily for single-ancestry datasets and may not fully capture cross-ancestry heterogeneity or shared biology. This PhD project will develop, evaluate and apply state-of-the-art multi-ancestry data integration methods to elucidate the biological mechanisms underlying skin disease. The work will leverage SGC GWAS resources alongside diverse functional genomic datasets, with established collaborations with pharmaceutical partners facilitating translation of findings into drug discovery pipelines. Aims The primary aim is to functionally characterise GWAS signals from the SGC in order to identify causal genes and biological pathways involved in skin disease, and to determine whether these are shared or ancestry-specific across populations. The project may focus on selected skin conditions of interest or apply methods across the full disease spectrum. Objectives

	- Develop and evaluate methods for multi-ancestry fine-mapping and multi-omic data integration - Refine causal variant resolution at GWAS loci using multi-ancestry fine-mapping approaches - Identify putative effector genes and biological pathways through integrative multi-omic analyses - Prioritise therapeutic targets, distinguishing shared versus ancestry-specific signals relevant to drug discovery Methods This project will focus on large-scale statistical genetics and data integration within high-performance computing environments and trusted research platforms. Analyses will be conducted using R, Python and Bash with secure computational infrastructures. 1. Multi-ancestry fine-mapping – A range of emerging methods will be implemented and evaluated to identify causal variants at GWAS loci under different modelling assumptions, including MESuSiE [3], mJAM [4], and Env-MR-MEGA [5]. These approaches will be applied to SGC multi-ancestry GWAS summary statistics to assess improvements in resolution and robustness across diverse populations. 2. Multi-omic integration – Publicly available expression (eQTL) and protein quantitative trait loci (pQTL) datasets, including those with diverse ancestral representation, will be systematically identified and integrated. Colocalisation and Mendelian Randomization analyses will be used to link GWAS signals to effector genes and downstream pathways [6]. The project will further assess heterogeneity in gene and pathway effects across ancestries. 3. Translation and target prioritisation - In collaboration with pharmaceutical industry partners, the most promising genetic signals and biological targets will be prioritised for further evaluation in drug discovery pipelines, supporting the translation of genetic discoveries into therapeutic development. References 1. World Health Organisation (2025), https://apps.who.int/gb/ebwha/pdf_files/EB156/B156_(24)-en.pdf 2. Budu-Aggrey, A et al (2023). Nat Comms 14(1), 6172 3. Gao, B et al (2024). Nat Genet 56, 170–179 4. Shen, J et al (2024). Genetic epidemiology, 48(6), 241–257 5. Wang, S et al (2024). Commun Biol 7, 1512 6. Shen, S et al (2024). J Invest Dermatol 44(6),1189-1199.e8
Can be completed part time?	Yes
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