

Genetic risk factors in women with endometriosis: A comparative analysis between Indian and European datasets

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Abstract

Endometriosis is a chronic gynaecological condition in which endometrium-like tissue grows outside the uterus, affecting about 10% of women of reproductive age, including approximately 50 million women in India. Endometriosis impacts overall well-being by affecting mental and physical health with chronic pelvic pain, menstrual disturbances and fertility issues. With 50% of the risk attributed to genetic factors, GWAS have identified 42 risk loci; however, the populations represented are mainly European, with limited studies in Asian populations. Overall, Indian ancestry remains significantly underrepresented in genetic studies, limiting the global understanding of genetic diversity.

To address this gap, the Endometriosis Clinical and Genetic Research in India (ECGRI) was established at ICMR-NIRWoH, integrating clinical and genomic data to better characterise disease susceptibility in the Indian context. As part of this initiative, we conducted a genome-wide association study (GWAS) using high-density genotyping data, following standard quality control, population stratification, and association analysis pipelines, with an aim to generate insights into the genetic basis of endometriosis that are more representative of Indian populations, thereby contributing to improved understanding of disease biology and future risk stratification approaches. This work was carried out as part of the DHR-ICMR International Fellowship at the University of Queensland, Australia, and will be presented as a fellowship report presentation.