

The Study of Pregnancy Loss in Uzbekistan: Unveiling the genetic landscape of miscarriage through GWAS Analysis

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The Advanced Pregnancy Loss Study in Uzbekistan (ALSU) is a new research project aimed at investigating the genetic factors contributing to miscarriage in women from the Uzbek population in Central Asia. This collaborative effort involves the Center for Advanced Technologies and the Institute of Biophysics and Biochemistry at National University of Uzbekistan, Tashkent, Uzbekistan. This study aims to collect information and biological samples from ~5,000 Uzbek women with/without idiopathic (recurrent) miscarriage to perform genome-wide association study (GWAS) and search for genetic factors underlying susceptibility to miscarriage in the Uzbek population.

The participants are enrolled at the Republic Centre of Gynaecology and Obstetrics, as well as at five medical centres across Tashkent and the Jizzakh region of Uzbekistan. To date, the ALSU study enrolled 1,338 women, including 214 women with one miscarriage, 184 with two or more miscarriages, 61 with stillbirth and 879 with no miscarriage and one life-birth as control group. All participants complete a questionnaire comprising 144 questions, covering family provenance information, anthropometry, environmental factors, reproductive history, clinical data, previous infectious and non-infectious diseases, as well as information about partners, parents, and close relatives, where applicable.

The age of participants ranged between 20 and 53 y.o. (mean±standard deviation, SD=29.8±6.45y.o.); while in cases, at the time of miscarriage age range was 17 to 42 y.o. (mean±SD=25.8±5.27 y.o.). The number of pregnancies was between one and 16.

The findings from ALSU will provide valuable insights into the aetiology of miscarriage, aiming at improvement of preventive precision medicine approaches to pave the way for interventions to prevent or mitigate pregnancy loss. Ultimately, this research will contribute to advancing reproductive health and maternal well-being.

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