Mutation associated with premature ovarian failure identified

1 December 2014

Premature ovarian failure (POF) is estimated to affect 1-4% of the female population. Women with POF can present with a variety of symptoms and many genes have been linked to this condition.

A new study in the *Journal of Clinical Investigation* identifies a specific mutation in a family that results in POF.

Aleksandar Rajkovic and colleagues at the University of Pittsburgh studied 3 sisters with POF-associated symptoms and identified and mutation in MCM8, a gene involved in chromosome maintenance.

Siblings without the MCM8 mutation did not have any signs of POF. Cells from the affected sisters exhibited an inability to repair DNA damage.

The results of this study indicate that chromosomal instability may be a factor in the development of POF.

More information: Exome sequencing reveals MCM8 mutation underlies ovarian failure and chromosomal instability, *Journal of Clinical Investigation*, 2014.

Provided by Journal of Clinical Investigation