**Abstract**

**Purpose:** Endometriosis is a common chronic gynecological disease greatly affecting women health. Prior studies have implicated that dysferlin (DYSF) aberration might be involved in the pathogenesis of ovarian endometriosis. In the present study, we explore the potential presence of DYSF mutations in a total of 152 Han Chinese samples with ovarian endometriosis.

**Methods:** We analyze the potential presence of DYSF mutations by direct DNA sequencing.

**Results:** A total of seven rare variants/mutations in the DYSF gene in 10 out of 152 samples (6.6%) were identified, including 5 rare variants and 2 novel mutations. For the 5 rare variants, p.R334W and p.G941S existed in 2 samples, p.R865W, p.R1173H and p.G1531S existed in single sample, respectively; for the two novel mutations, p.W352\* and p.I1642F, they were identified in three patients. These rare variants/mutations were absent or existed at extremely low frequency either in our 1006 local control women without endometriosis, or in the China Metabolic Analytics Project (ChinaMAP) and Genome Aggregation Database (gnomAD) databases. Evolutionary conservation analysis results suggested that all of these rare variants/mutations were evolutionarily conserved among 11 vertebrate species from Human to Fox. Furthermore, in silico analysis results suggested these rare variants/mutations were disease-causing. Nevertheless, we find no significant association between DYSF rare variants/mutations and the clinical features in our patients. To our knowledge, this is the first report revealing frequent DYSF mutations in ovarian endometriosis.

**Conclusion:** We identified a high frequency of DYSF rare variants/mutations in ovarian endometriosis for the first time. This study suggests a new correlation between DYSF rare variants/mutations and ovarian endometriosis, implicating DYSF rare variants/mutations might be positively involved in the pathogenesis of ovarian endometriosis.