DPY19L2 gene deletion is not a cause for partial globozoospermia

Ebrahimi Nasab Mahya (IR) [1], Sabbaghian Marjan (IR) [2], Totonchi Mehdi (IR) [3], Ghezelayagh Zeinab (IR) [4]

Globozoospermia is a rare but severe teratozoospermia characterized by lacking acrosome. Most of the researches based on describing this morphology deal with patients who produced ejaculates showing 100% round sperm heads, however, it has been shown a number of patients who only had lower percent of round-headed spermatozoa in their ejaculate. Fertile men have 6% round-headed spermatozoa in their semen but with increasing this percent, fertility chance decreases. Studies have shown that in large majority of globozoospermia patients a 200 kb deletion including DPY19L2 gene occurs. Lack of acrosome, decreases the capacity of the sperm for fertilizing the oocyte which consequently results in infertility. The aim of our study was to assess the frequency of DPY19L2 gene deletion in Iranian infertile male with partial or total globozoospermia. In this study, 42 globozoospermic patients (20-100% round headed sperm) and 42 men with normal spermogram referred to Royan Institute in Tehran were included. DPY19L2 gene was assessed in men with globozoospermia and normozoospermic control by analyzing exons 1 and, 19 and a 200 kb encompassing this gene. The results showed a whole DPY19L2 gene deletion in 41.7 % of globozoospermic patients which all had a high percent round head sperms (90%-100%). None of patients with low percent of round head sperms (<90%) and also control group showed this large deletion. Deletion of DPY19L2 gene as the main cause reported for total globozoospermia (100% round head sperm) was not seen in the male with partial globozoospermia. Therefore, it seems that an unknown factor or mutation instead of whole DPY19L2 gene deletion cause similar phenotype in partial globozoospermia.