GENETICS OF RECURRENT HYDATIDIFORM MOLE: DETECTION OF TWO NOVEL MUTATIONS IN THE NLRP7 GENE IN TWO EGYPTIAN FAMILIES

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Objectives: Hydatidiform mole is an aberrant pregnancy with hyperproliferative vesicular trophoblast and defective fetal development. In 2006, mutations in NLRP7 were found to be responsible for recurrent hydatidiform moles (RHM), but genetic heterogeneity has been demonstrated and mutations of C6orf221 were later reported in several families. Here we report a new Egyptian family in which two sisters had eleven and four molar pregnancies, respectively. The objective was to present the results of the mutation analysis of NLRP7 and C6orf221 genes in Egyptian women with RHM.

Study design: Three women from two unrelated Egyptian families; two sisters and a previously described sporadic case, all presenting with RHM, were enrolled. The cases were subjected to detailed history taking, karyotyping and screening for mutations in NLRP7 and C6orf221.

Results: Two NLRP7 mutations have been detected, one in each family. In the first family, sequencing identified a homozygous 2 bp deletion in the seventh coding exon of NLRP7, while a homozygous G-to-A substitution in the third coding exon of NLRP7 was detected in the second family. Both of them result in a truncated protein. The two mutations have not been previously described in the literature. No mutations in C6orf221 were found in any of the samples.

Conclusion: The detection of an NLRP7 mutation in both the familial and the apparently isolated case of RHM provides further evidence for the previously established role of NLRP7 mutations in the pathophysiology of RHM and increases the diversity of mutations described in the Egyptian population. Our results also expand further the spectrum of reproductive wastage associated with NLRP7 mutations to patients with recurrent spontaneous abortion.