



## Genetic marker for Molar Pregnancy

### Information Summary

Reference Code:	ROI 06009
Technology overview:	Novel genetic test based on NALP7 mutations for poorly characterized reproductive conditions.
Application:	Diagnostic marker to assess reproductive wastage with unexplained clinical origin. Test enables informed decision to proceed with conception, either naturally or through assisted reproductive technology.
Validation:	Different mutations in NALP7 identified in individuals with familial and recurrent molar pregnancy.
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### Technology Description

Research carried out in the laboratory of Dr. Rima Slim at the McGill University Health Center Research Institute has identified a gene, NALP7, which is mutated in patients with a recurrent form of molar pregnancy. Hydatidiform moles (HM), commonly known as molar pregnancy, are the most common gestational tumors.

### Medical Need

Hydatidiform moles are usually benign and result in an aberrant human pregnancy with no embryo and

hydropic degeneration of placental villi. Complications may develop in 15 to 30% of cases where molar tissues invade the wall the uteral wall leading to an invasive mole. In 5% of cases in Canada (up to 30% in Africa) the complications can lead to a gestational choriocarcinoma, a malignant, rapidly growing, and metastatic cancer. In addition to the recurrent molar pregnancies, mutations within NALP7 can also result in other forms of reproductive wastage such as spontaneous abortions at various gestational stages, stillbirths and normal pregnancies with intrauterine growth defects.

### Opportunity

As the first maternal gene linked to reproductive wastage, NALP7 constitutes a prime target for genetic testing. In 2000, the molecular diagnostics market has generated more than one billions US\$ in revenues. HM occur in 1 in every 1500 pregnancies in western countries and up to 1 in every 250 pregnancies in Latin America, the Far East, and some Middle Eastern countries. This test will also be available for a second patient population made up of women suffering from recurrent spontaneous abortion (RSA). For every 3,000,000 US births annually, at least 600,000 embryos or fetuses are spontaneously aborted. Although there exists a multitude of genetic and cytological assays related to reproduction and fertility, these tests focus on prenatal diagnosis for the detection of a disease or condition in a fetus or embryo. NALP7, as the first identified maternal gene causing reproductive wastage, therefore constitutes a very attractive target for diagnostic test development.

### Additional Reference Material

Murdoch, S. et al., Mutations in NALP7 cause recurrent hydatidiform moles and reproductive wastage in humans *Nature Genetics* 38: 300-302.