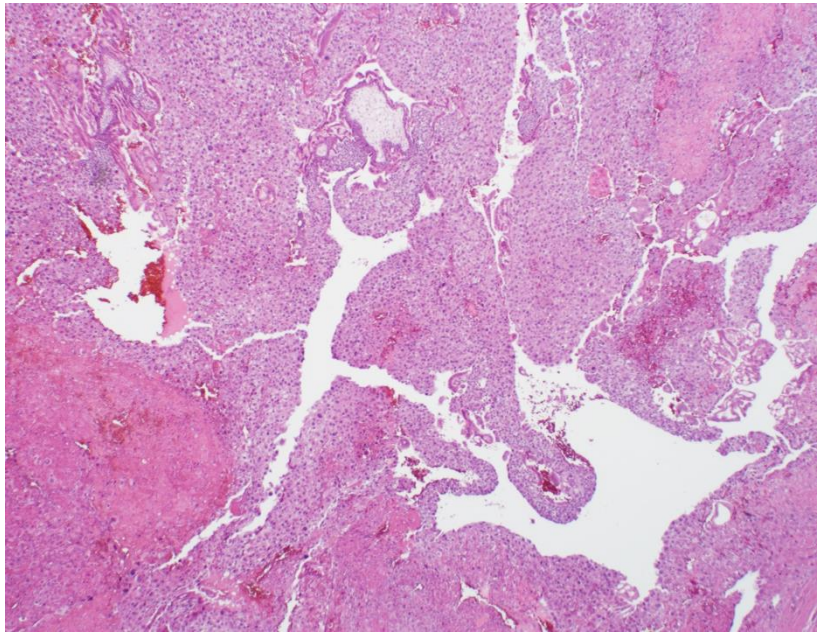
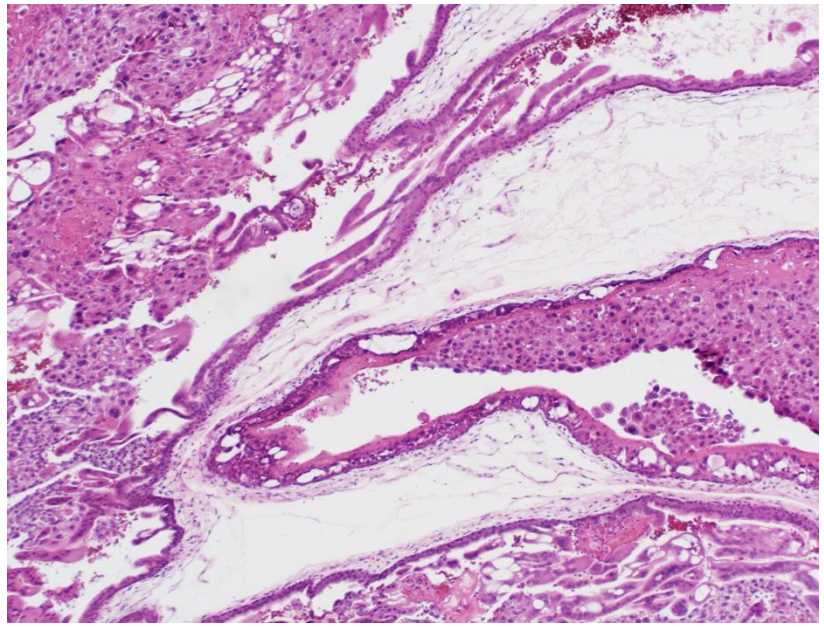
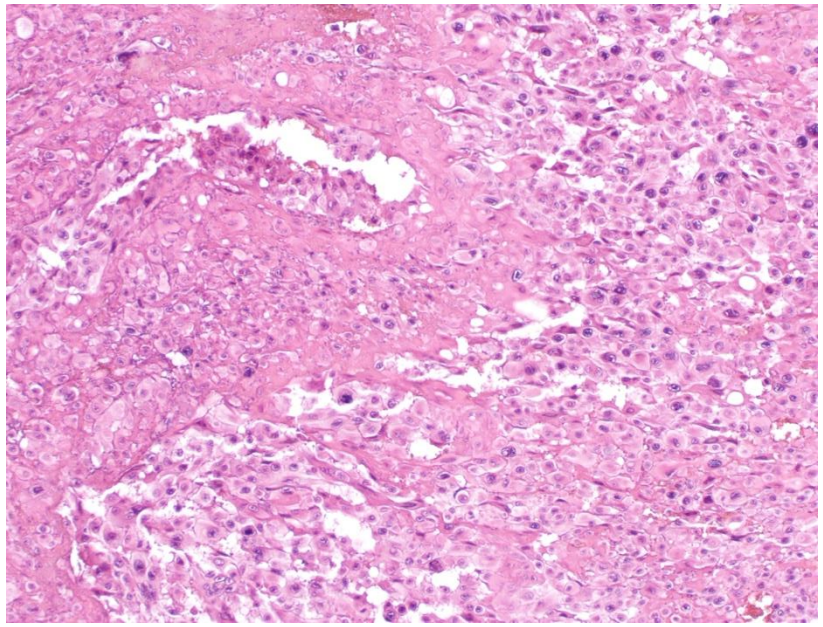
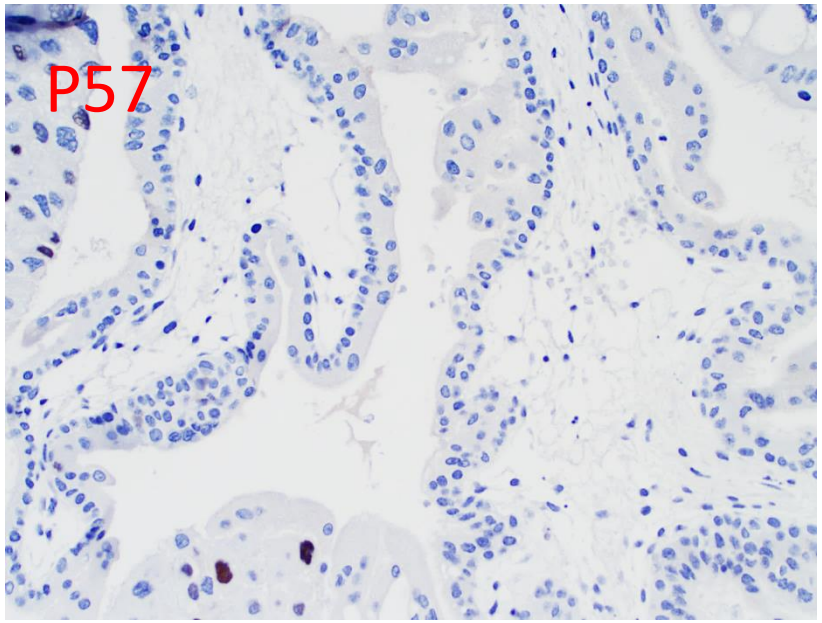
Where Knowledge is Created and Shared
Yale Pathology

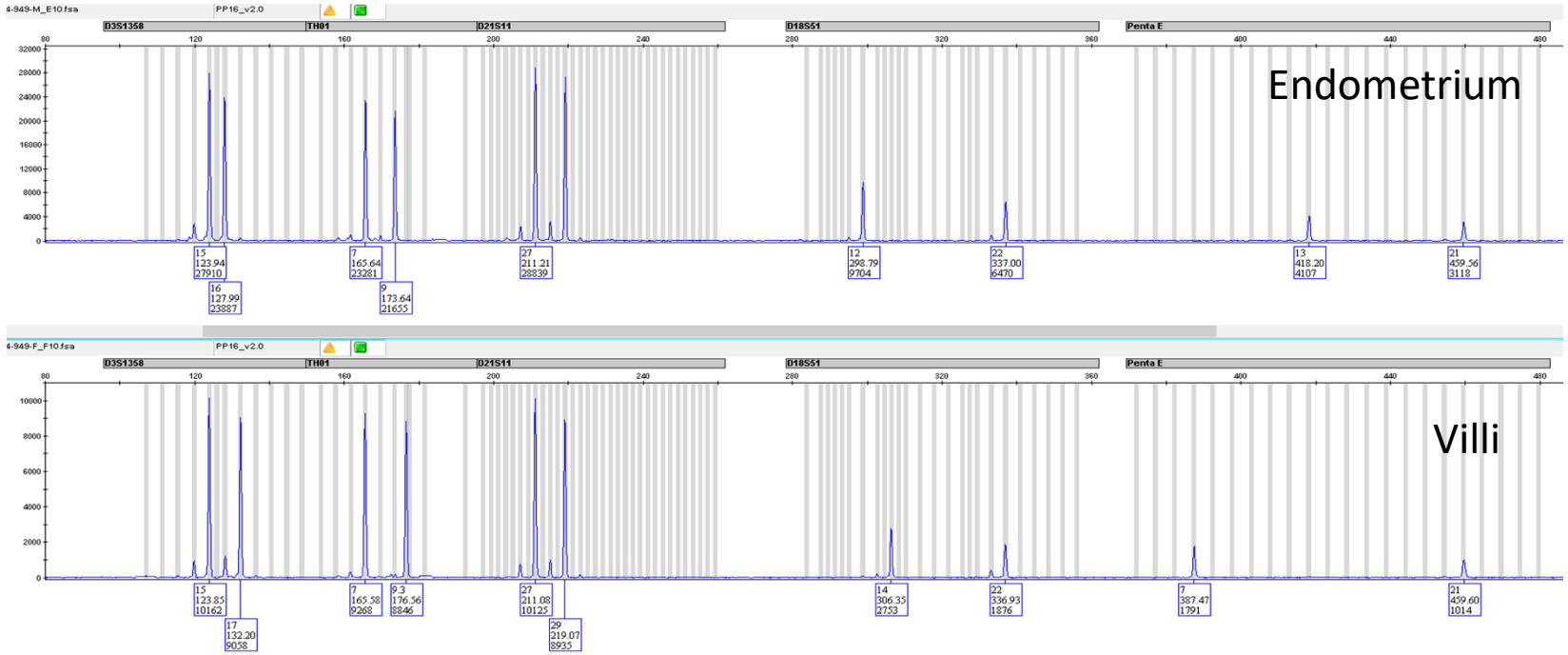
31-year-old patient presenting with abnormal gestation after multiple failed pregnancies, high serum hCG, uterine mass and lung nodules underwent hysterectomy








P57


Endometrium
Villi

Hysterectomy specimen showed an aggressive myoinvasive lesion with embedded hydropic villi, abnormal trophoblastic proliferation, loss of p57 expression in cytotrophoblast and villous stromal cells. However, a balanced/normal biparental STR genotype was observed.

Diagnostic Options

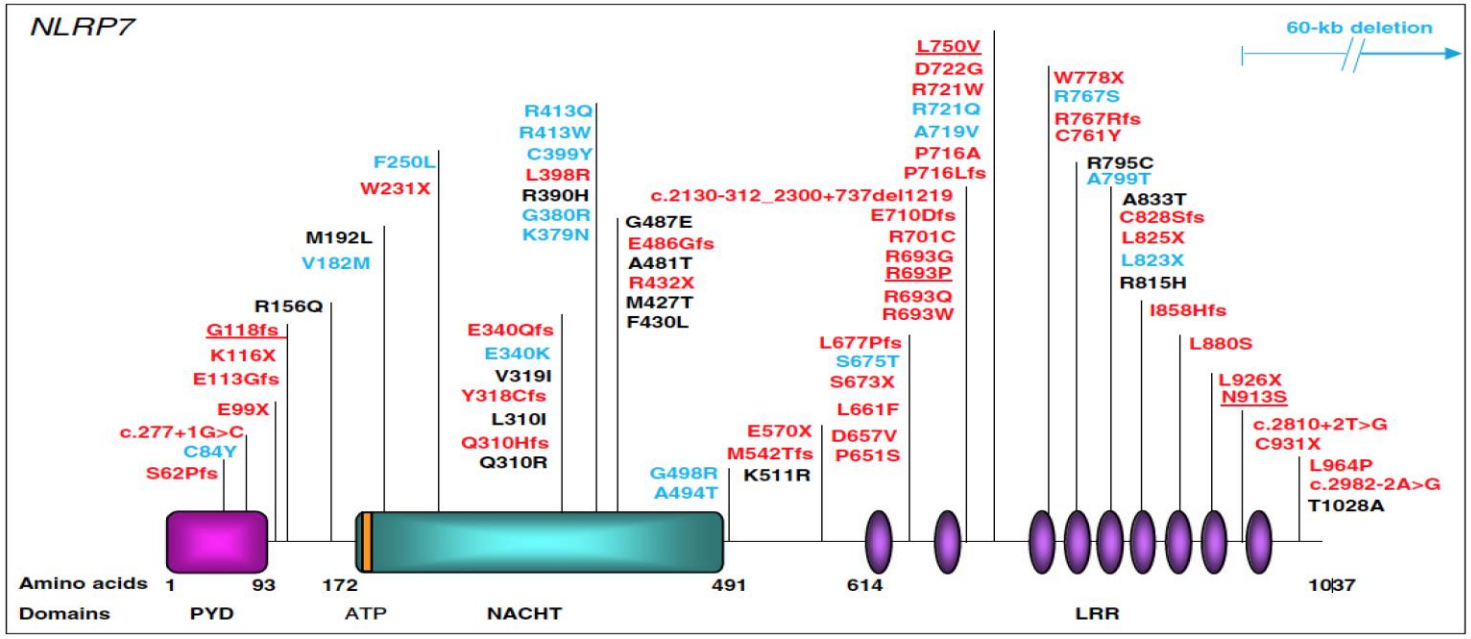
A. Invasive complete mole
B. Invasive partial mole
C. Invasive non-molar hydropic gestation

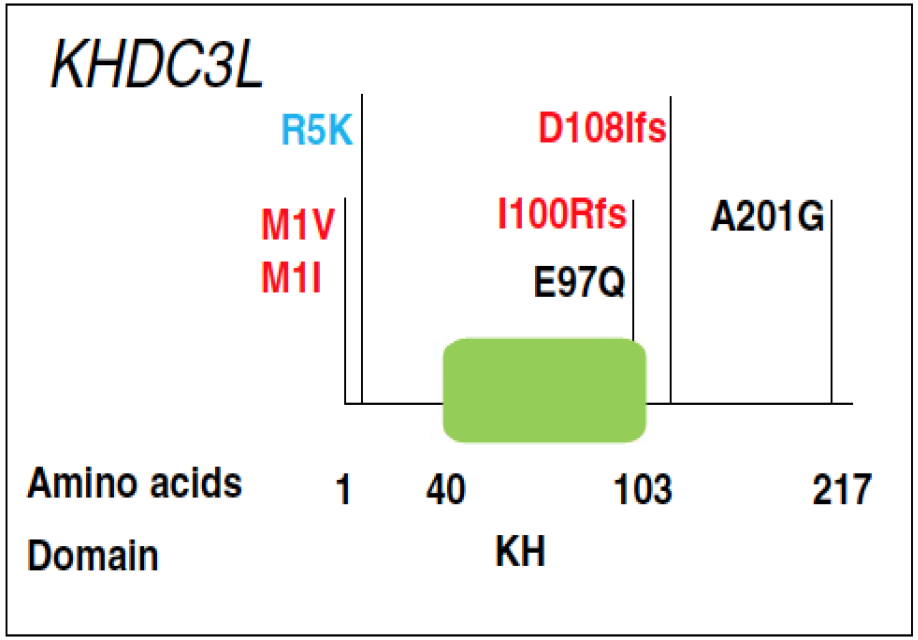
Hydatidiform Moles – Two Subtypes

- Sporadic Hydatidiform Moles (>97%)
- Familial Biparental Hydatidiform Moles

Biparental Recurrent Hydatidiform Moles

Familial recurrent hydatidiform moles represent 0.6 to 2.6% of all hydatidiform moles, among which familial biparental CHM (FBCHM) is an exceptional condition. Initially reported 40 years ago as recurrent moles in multiple pregnancies of sisters in three unrelated families, close to 300 cases of FBCHM have since been documented.





Homozygous or compound heterozygous mutations of *NLRP7* or *KHDC3L* are responsible for the development of FBCHM

Nguyen and Slim:Curr Obsete Gynecol Rep,2014.

Homozygous Leu750Val mutation of NLRP7 is identified by subsequent NGS analysis of the maternal decidua.

Final Diagnosis

Invasive Familial Biparental Complete Mole

The risk of gestational trophoblastic neoplasia or GTN in patients with familial biparental complete mole (FBCHM) is similar to that of sporadic androgenic complete moles. It is important to note that in contrast to sporadic complete moles, patients with inherited biallelic mutations of either *NLRP7* or *KHDC3L* have a rather poor reproductive outcome. Egg donor gestation may be the only option to achieve a successful pregnancy.