



Memorial Sloan Kettering  
Cancer Center.

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### **Aggressive Ovarian Cancer Mutation Identified**

New York, NY, March 23, 2014 – Memorial Sloan Kettering researchers have identified a genetic mutation that appears to cause small cell carcinoma of the ovary, hypercalcemic type (SCCOHT), a rare but very aggressive type of ovarian cancer in young women. The findings will be published on March 23 in *Nature Genetics*.

Around 300 women are diagnosed with this cancer each year, and there “are many examples of women in their late teens or early 20s, dying quickly from this disease,” says gynecologic oncologist Douglas Levine, who led the research. “The median age is 23, and they frequently die within two years – it’s devastating. Identifying this potential genetic target could be a major advance because current therapies have very little effect.”

The finding was made through genetic analysis of tumor samples from 12 patients with SCCOHT. MSK genomics researcher Michael Berger sequenced 279 genes that have been implicated in the development or behavior of tumors and found that all 12 samples had a mutation in a gene called *SMARCA4*. According to Dr. Levine, it is quite rare to find a single gene universally mutated in a given cancer.

Using human cancer cells, Dr. Levine and colleagues demonstrated that *SMARCA4* plays a functional role in promoting or blocking cancer growth. Inactivating *SMARCA4* increased cancer growth, while boosting its expression slowed it down. This buttressed the theory that the *SMARCA4* mutation was key to the rare disease.

Dr. Levine adds that next steps include investigating whether this one mutation is actually sufficient to transform ovarian cells into cancer, or whether other factors are involved. The researchers also plan to develop potential therapies and test them on cell lines that carry the *SMARCA4* mutation.

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#### **About Memorial Sloan Kettering Cancer Center**

We are the world’s oldest and largest private cancer center, home to more than 12,000 physicians, scientists, nurses, and staff united by a relentless dedication to conquering cancer. As an independent institution, we combine 130 years of research and clinical leadership with the freedom to provide highly individualized, exceptional care to each patient. And our always-evolving educational programs continue to train new leaders in the field, here and around the world. For more information, go to [www.mskcc.org](http://www.mskcc.org).