BAP1 mutation passed down over centuries and is associated with high incidence of several cancers

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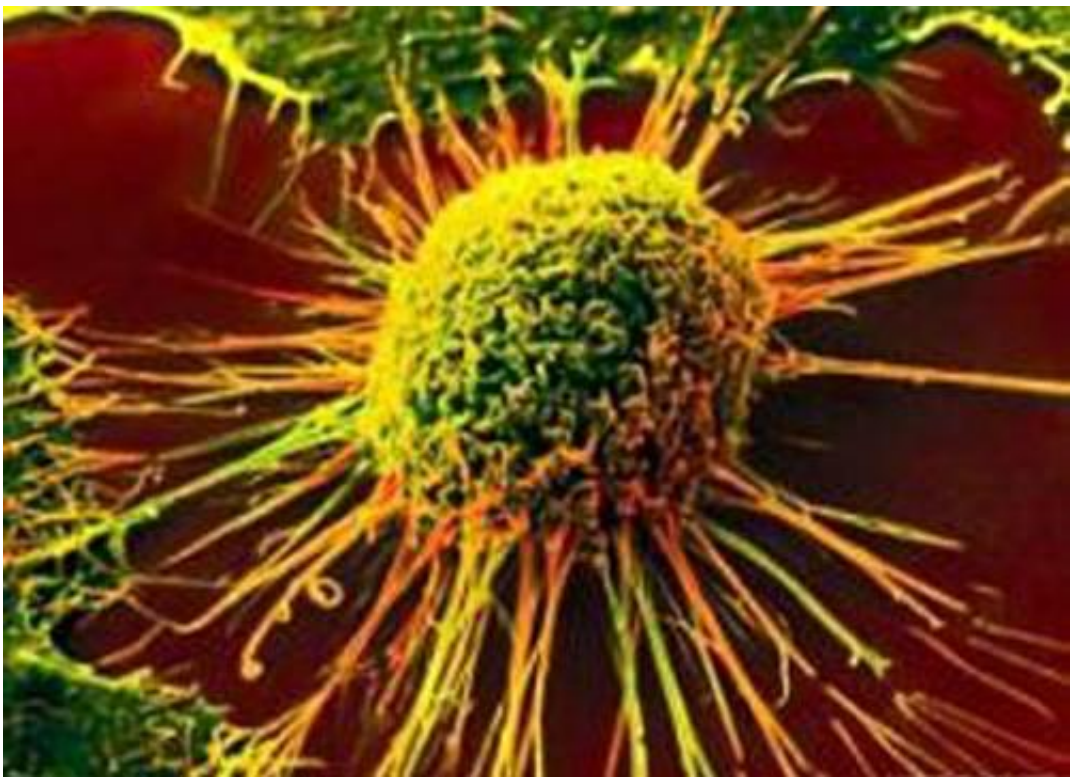

Michele Carbone and colleagues, from the University of Hawaii, discovered that members of 4 families, apparently unrelated and living in different US States, shared the identical mutation of a gene called BAP1 that is associated with a higher incidence of mesothelioma, melanoma,

renal carcinoma and other cancers.

This raised two possibilities:

1) These 4 families were related although they did not know it, or

2) The researchers had found a hot spot for BAP1 mutations.

Through genetic and genealogical studies, it was demonstrated that the families were related, and that they descended from a couple that immigrated to the USA from Germany in the early 1700s.

The results showed that BAP1 mutations are transmitted across several generations. Over the course of 3 years, Carbone and colleagues travelled across the US and other parts of the world in order to find the evidence linking these 4 families, constructing a large family tree of approximately 80,000 individuals.

These findings are published in *PLOS Genetics* and the authors anticipate that the publication of their work will accelerate their research and help them to identify more descendants of these families. These descendants can be tested for BAP1 mutations, and if they are found to have inherited the mutation, they can be followed for cancer prevention and also for early detection. This has important implications, especially for cancers such as melanomas of the skin and of the eye, which are 100% curable when detected at an early stage.

More information: Michele Carbone et al. Combined Genetic and Genealogic Studies Uncover a Large BAP1 Cancer Syndrome Kindred Tracing Back Nine Generations to a Common Ancestor from the 1700s, *PLOS Genetics* (2015). DOI: 10.1371/journal.pgen.1005633

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