What is FAMMM (familial atypical multiple mole and melanoma syndrome)?

My name is Anil Rustgi, I am the chief of gastroenterology at the University of Pennsylvania. My research and clinical interests are devoted to pancreatic cancer. From a research viewpoint, my group is interested in the mechanisms that underlie the initiation, progression, and spread of pancreatic cancer; and from a clinical viewpoint, I am heavily immersed in the identification characterizations of patients and family members who are at increased risk for pancreatic cancer and who might then be amenable to genetic testing and counseling. Amongst the risk factors that are inherited on a familial basis with pancreatic cancer is something that I am asked about, and one relates to what is called FAMMM, which stands for familial atypical multiple mole and melanoma syndrome. This is an unusual or rare condition but if suspected, needs to be evaluated. In this context, individuals in the family will have melanoma, there should be at least two family members with melanoma, one family member himself or herself should have multiple melanomas, and there should be a family member with pancreatic cancer, often early in onset, sometimes as early as someone in their 20s, 30s, and 40s. In the combination of melanoma and pancreatic cancer, one should be concerned about this syndrome. Actually the genetic basis is known for this, the gene that is altered or mutated that can be identified through genetic testing is called P16, and so if one seeks the appropriate genetic testing and counseling under informed consent, then P16 genetic testing can be obtained, the results then come back in a few weeks, and then again with the gastroenterologist who serves as the geneticist and genetic counselor, then the next steps in terms of potential screening in at-risk individuals can be pursued. Also then if it is known that a particular family member has a P16 gene mutation, then other at-risk members, siblings, children, in an age-appropriate fashion, first-cousins, uncles and aunts can be screened as well to see if they either have the mutation or they do not.