

# informe\_pdf



#### User info:

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### Diagnose info:

Illness Name Illness ID:

## Illness description:

what is familial atypical melanoma?

Melanoma-pancreatic cancer syndrome 404560



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### **Symptoms:**

The clinical phenotype of FAMMM syndrome shows wide heterogeneity in regards to the presence of nevi and familial predisposition to melanoma and pancreatic cancer (mainly adenocarcinoma). The disease most often presents in children and teenagers but can occur at any age. The presenting feature is usually a high total body nevi count (usually more than 50). The majority will be clinically typical but some may have an atypical appearance (asymmetrical, raised, and/or different shades of tan, brown, black, or red and often of different sizes) resembling early melanoma and most frequently occur on the back, chest, buttocks, breasts, and scalp. Melanomas can arise from atypical moles or de novo and have been reported in some FAMMM syndrome patients as early as the second to third decade of life. Those with CDNK2A mutations have a 90% risk of developing melanoma by the age of 80 and a 20% increased risk of developing pancreatic cancer by the age of 75. These mutations are also associated with a younger age of onset. Other cancers that can be rarely associated with FAMMM syndrome include breast cancer, esophageal cancer and sarcoma.

#### **Proposed solution:**

Source of information https://www.orpha.net/consor4.01/www/cgi-bin/OC\_Exp.php?Ing=EN&Expert=404560