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Genetic and Neuro-Imaging Aspects in Neurofibromatosis 1

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Short Communication

Neurofibromatosis 1 (NF1) is the most common genetic diseases. It is transmitted by dominant autosomal mode. But there is 50% of de novo mutation. It is a predisposing condition for benign and malignant tumors.

Cutaneous manifestations are important in diagnosis. According to the NIH, diagnostic of NF1 is established if two or more of these following criterias are met in an individual:

- Six or more "café au lait" macules over 5 mm in prepubertal individuals and over 15 mm in post pubertal individuals (Figure 1),
- Two or more neurofibromas of any type or one plexiform neurofibroma (Figure 2),
- Freckling in the axillary or inguinal régions
- Optic glioma
- Two ore more lisch nodules (iris hamartomas)
- A distinctive osseous lésion such as sphenoid or tibial pseudarthrosis
- A first-degree relative with NF1as defined by the above criteria [1,2].

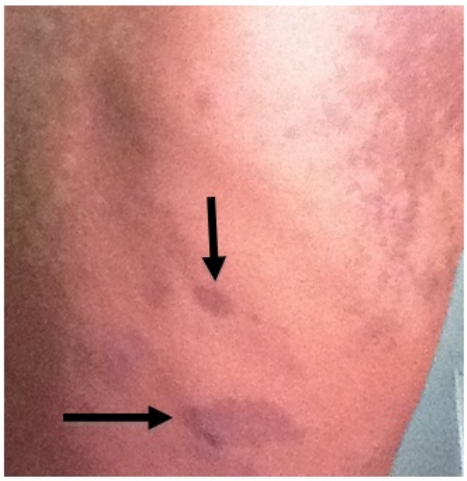

Figure 1: Showing "café au lait" macules (arrows) in a 18 years old woman.

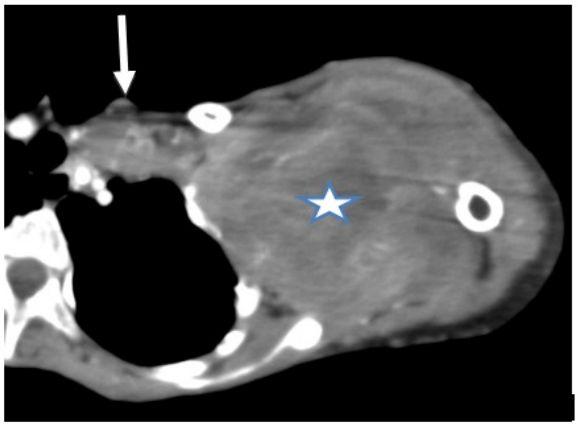

Figure 2: Shows a malignant tumor of axillary left region (star) associated with sub clavicular cutaneous neurofibroma (arrow) in a 42 years old woman.

NF 1 gene's is localized on the chromosome 17 (17q11.2). It's responsible of the synthesis of neurofibromin. This protein involves in the control of cell differentiation and proliferation by inhibiting the activation pathway of the p21ras protein. Loosing expression of this gene is associated with loosing control of cellular differentiation and way to development of benign and malignant tumor. The risk of malignant tumors occurrence is estimated 5 to 10%. However, authors note that alteration of other genes encoding proteins p53, p16, p27-Kip1 that are involved in cell cycle regulation is necessary for the genesis of malignant tumors. Malignant tumors in NF1 are the most frequent cause of death around the age of 40 (Figure 2).

Neuro-imaging involves in all steps in NF1; from the initial diagnosis, assessment and monitoring of evolving lesions that can degenerate or clinically difficult to assess. For example we cite unidentified bright objects (UIBO). They correspond to dysplastic lesions with heterotopias or melanocytic cells. It joins in vacuolation myelinopathy that generates swelling and the hight T2 signal on MRI examination. These UIBO can develop into glioma and need neuroimaging monitoring. The figure 3 shows an evolutionary appearance with UBIO and glial lesions in the same patient. Ependymomas and meningiomas are also reported as showed in the Figure 3.

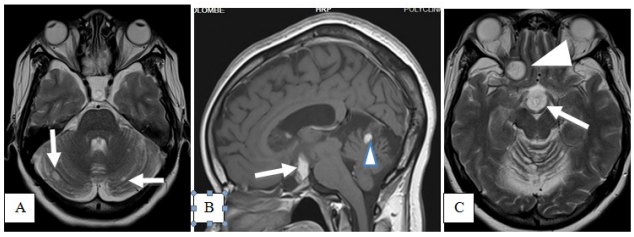

Figure 3: Shows multiples UIBO in cerebellum parenchyma and the vermis (arrows in A). One of them is enhanced after gadolinium injection indicating a transformation into glioma (arrowhead in B). Enlargement and hypersignal of the V3 with enhancement after injection indicating an ependymoma (arrows in B and C). Meningioma of orbitary apex showed on C (arrowhead in C).

References

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