Coexistence of Wilson’s disease and neurofibromatosis type 1 in a 14-year-old boy

Sir,

We herein report a 14-year-old boy admitted to our hospital, with a single episode of secondary generalized tonic-clonic convulsion. He had history of mild learning difficulty at school from two years prior to admission [Figure 1].

On physical examination, we found more than 12 café-au-lait spots (most \( \geq 2 \) cm), re-established with dermatologic consultation using biopsy, over the body and axillary freckling, without any plexiform neuroma or neurofibroma, which raised the possibility of neurofibromatosis Type 1 (NF1).

Routine blood tests were normal and brain magnetic resonance imaging (MRI) showed signal changes in basal ganglia and cerebral cortex with hydrocephaly.

He developed bradykinesia, cogwheel rigidity and mental impairment during follow-up treating with carbamazepin and reevaluated with impression of young onset Parkinsonism. Another precise physical examination showed KF rings and lab tests were all compatible with WD [marked decreased serum ceruloplasmin and copper, increased urinary copper].

The patient was started on Trientine and zinc sulfate and we discontinued trientine after eight weeks.

This patient presented with café-au-lait spots and axillary freckling. According to clinical criteria,\(^1\) he fulfilled the criteria of NF1, but had no positive family history. During follow-up, Parkinsonism had added to the patient’s problems, a finding that is not compatible with NF1.

Although it is advised to explain all the signs and symptoms with one disease, there are instances in which another concomitant disease should be considered.

The most common features of NF1 are pigmentary abnormalities, such as macules, skin folds freckling and iris hamartomas (Lisch Nodules).

They are prone to the development of a wide variety of nervous system abnormalities like benign and malignant tumors of the peripheral and central nervous systems.\(^2\)

Diagnosis of NF1 is based on clinical criteria,\(^3,4\) although genetic study can be used for finding the mutations.

This case may be interesting with having two different genetic diseases with diverse neurological complications, although estimating statistically, this may be the only case in the world, but it is possible that perhaps 60 other similar examples exist.

Two different genetic diseases may be present at the
same time and during practice, we should avoid early closure of diagnosis. This may result from premature fixation on some items in the history or examination.

The first diagnostic consideration should be rearranged to modification when new items of information are secured.

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References


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