1. Abstract

1.1. Background: Neurofibromatosis type I (NFI) is a common genodermatosis, with an incidence of 1 new case to 3000-3500 live births. Described in 1882, it occurs from gene mutations of the encoding protein called neurofibrinin. NFI patients are at increased risk for neoplasms. Aims: We report a case of a patient with neurofibromatosis with a recent diagnosis of pheochromocytoma (PHEO), due to the rarity of this association. 1.2. Case Report: Forty-six years old female patient with NF1 and hypertension since the age of 25, developing difficulty in controlling blood pressure levels. During the investigation, a left adrenal mass was identified in tomography, with a hyper capturing pattern on the scintigraphy. After preparation, adrenalectomy was performed. Conclusions: Thirty percent of patients with NF1 develop hypertension, usually primary. A lower percentage may be secondary, usually due to renovascular causes. About 1% of cases hypertension is caused by PHEO. It’s a rare neoplasm that occurs between 20 and 50 years of age, with no gender predilection. Refractory hypertension, associated with paroxysms of headache, intense sweating and palpitations suggests PHEO. The treatment is surgical. Long-term follow-up of NF1 patients is common in dermatological practice. With the presentation of this rare association, we call attention to the adequate follow-up of these patients the present with hypertension.
this context, during investigation, she presented a left adrenal mass on a CT-exam. (Figure 4) An addition scintigraphy showed hypercaptation in the upper region above the left kidney. With a probable diagnosis of PHEO, she was hospitalized to terminate investigation, clinical compensation and surgical treatment. Vidéo laparoscopic adrenalectomy was performed, without complications. Patient is still being followed, now with 6 months post treatment, with no evidence of recurrence of the disease, requiring no more anti-hypertensive treatment.

6. Discussion

NFI is known to be a disease whose management requires a multidisciplinary team, being fundamental its knowledge in several areas, even at the primary care level, in view of its significant prevalence1. However, in general, the patients are at least accompanied by the dermatologist because of the exuberance of the cutaneous lesions. The importance of follow-up, since effective treatment is not available, lies mainly in the higher incidence of tumors, including malignant behavior ones. A cohort of 448 patients with NFI, with a follow-up of 5705 patients / year, found a 2.7-fold higher risk of malignant neoplasia in relation to the general population. It is postulated that the mutation involved inactivate NF1 genes with tumor suppressor activity, possibly explaining the increased frequency of neoplasias [2].

About 30% of NFI patients develop hypertension, and it is therefore essential that these patients have their blood pressure checked. Most cases are primary or essential, but in a lower percentage it may be secondary to a renovascular component and, even more uncommonly, can be justified by the presence of PHEO (about 1% of cases) [3].

PHEO corresponds to a rare neoplasm of chromaffin cells, with prevalence in the general population of about 2 to 8 cases: 1,000,000. It affects patients between 20 and 50 years of age, and there is no predilection for sex. Ninety percent of the cases are benign, unilateral, located in the adrenal gland [4,5]. About 10% of them are family members associated with dominant autosomal disorders, such as NF1. Tuberous sclerosis, Stuge-Weber syndrome and Carney syndrome are also of dermatological importance [5].

Refractory hypertension associated with paroxysms of headache, intense sweating and palpitations suggest FEO [4]. Diagnosis involves the measurement of serum and urinary metanephrines and catecholamines, as well as magnetic resonance imaging or scintigraphy for localization of the tumor. Surgical excision is the appropriate treatment and, in general, it is via laparoscopic surgery after adequate preoperative preparation [4,5].

Screening for PHEO in patients with NF1 is generally recommended at the onset of hypertension [6,7]. However, a study in patients with these two conditions showed that, in most cases, there was no change in blood pressure. Thus, the suspicion of PHEO should not be restricted to the onset of hypertension, but also to other signs and symptoms that may indicate the presumption of this diagnosis [4]. However, the plan to screen only symptomatic patients has led to an underestimation of the prevalence in these individuals. Thus, screening for PHEO in patients with NF1 is recommended for all patients over 40 years of age through
the quantitative analysis of metanephrines and imaging tests such as CT or RM [8]. Additional examinations before pregnancy and any surgeries are also recommended, although there is still a need to determine the cost-benefit of this strategy [9].

7. Conclusions
Among genodermatoses, NF1 is the most common, so the care of these patients is common in the dermatological practice. In addition, the survival of its patients is close to the general population, in such a way that these patients will be followed in the long term. We report this case due to the rarity of this association with PHEO, and the need for adequate follow-up of these patients.

References