Evaluation of endothelial function using digital tonometry in NF1 patients: a preliminary report

Luiza de Oliveira Rodrigues, Luiz Oswaldo Carneiro Rodrigues, Miguel Tannus,
Luisa Lima Castro, Nilton Alves de Rezende and Antonio Luiz Pinho Ribeiro
Institution/University: Faculdade de Medicina e Hospital das Clínicas da Universidade Federal de Minas Gerais, Belo Horizonte, Brazil.

Background: Neurofibromatosis type 1 (NF1) is known to cause a reduction in life expectancy that has been attributed to malignancies. Recent studies have suggested that cardiovascular disease (CD) could be the main cause of death in these patients. Neurofibromin has an important role in endothelial cells and endothelial dysfunction (ED) has been studied as a possible factor associated to, or responsible for, a higher incidence of CD and premature death in NF1 patients.
Aim: To assess endothelial function (EF) through a non-invasive method in young adult NF1 patients, and matched controls, with no CD risk factors.

Methods: EF was measured in 16 NF1 patients and in 19 healthy subjects, with the Reactive Hyperemia Peripheral Artery Tonometry (RH-PAT) technology, using the EndoPAT2000® equipment (Fig.1). The presence of ED was defined by reactive hyperemia index (RHI) lower than 1.67. The NHI criteria for diagnosis of NF1 were used. In both groups the exclusion criteria were any risk factor for CD (such as hypertension, dyslipidemia, smoking, diabetes, family history of CD) as well as any organ dysfunction (liver, kidney, thyroid) or cancer, recognized to compromise endothelial function. Patients without definite diagnosis of NF1, such as the possibility of Legius syndrome or segmental NF1, were excluded.

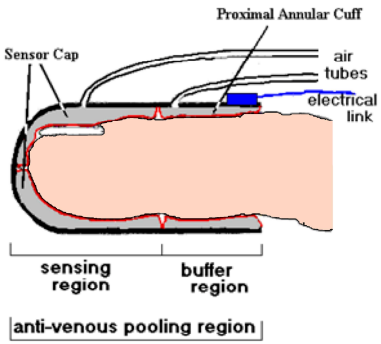

Fig.1 - EndoPAT2000 finger probe

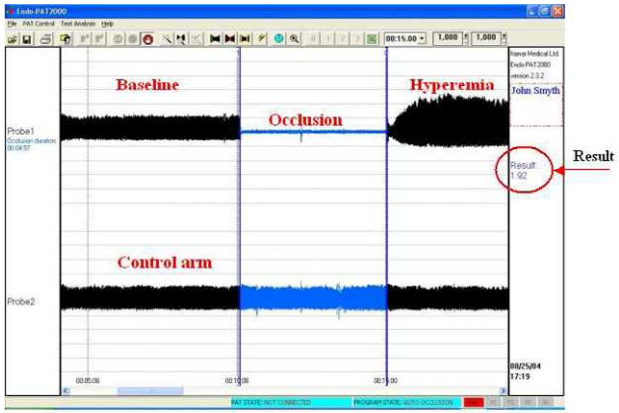

Fig.2 - EndoPAT2000 sofware results

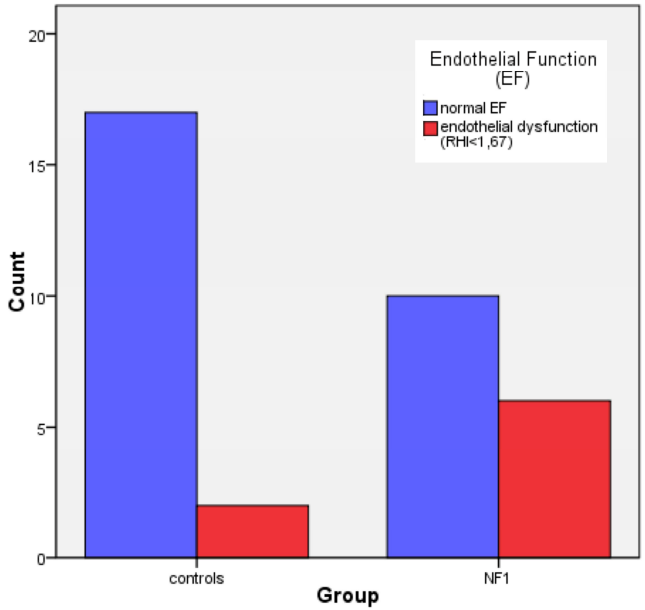

Fig.3 – Subjects with Endothelial Dysfunction in NF1 and Control Groups

Results: The mean age, body mass index, blood pressure levels, habitual physical activity levels and the male/female proportions were similar in both groups. The use of hormonal contraceptives was less prevalent (p=0.000) in NF1 women (8%) than in control group women (71%). The mean heart rate during the test was greater (p=0.044) in NF1 patients (76.5±8.48 bpm) than in controls (70.21±9.24 bpm). Reduced RHI characterizing ED was found in 6 (37.5%) of the patients of NF1 group and 2 (10.5%) of the patients in the control group (p=0.105).

Conclusion: These preliminary data suggest that NF1 patients have an increased prevalence of ED. This study is ongoing and by expanding the number of participants it might be possible to recognize a stronger association between NF1 and endothelial dysfunction.

References:

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