A new approach to measure the severity of NF1: a preliminary report

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Neurofibromatosis type 1 (NF1) affects quality of life through its clinical complications and cosmetic features. Riccardi & Kleiner (1977) have originally ranked NF1 severity and Ablon (1996) presented a scale to rank the visibility of NF1. Wolkenstein et al (2001) hypothesized that the severity of NF1 would be better quantified if severity and visibility scales were used together. **Methods**: 172 NF1 patients presented at the CRNF were classified according to Riccardi´s severity scale (R) and Ablon´s visibility scale (A) and to a new modified Riccardi´s plus Ablon´s scale (R+A) as folows: typed **in red, the R scale**, **in green the A scale and in blue the modified itens for the new R+A scale**:

Very mild form

café-au-lait spots, limited neurofibromas, Lisch nodules - features apparently unremarkable when the patients were observed fully dressed.

Mild form

multiple neurofibromas, pubertal abnormalities, cognitive and learning disabilities, chronic headache, muscular hipotony, short stature, voice and speech disturbances and dermatological or orthopedic problems - features observed hardly at a glance when the patients were fully dressed.

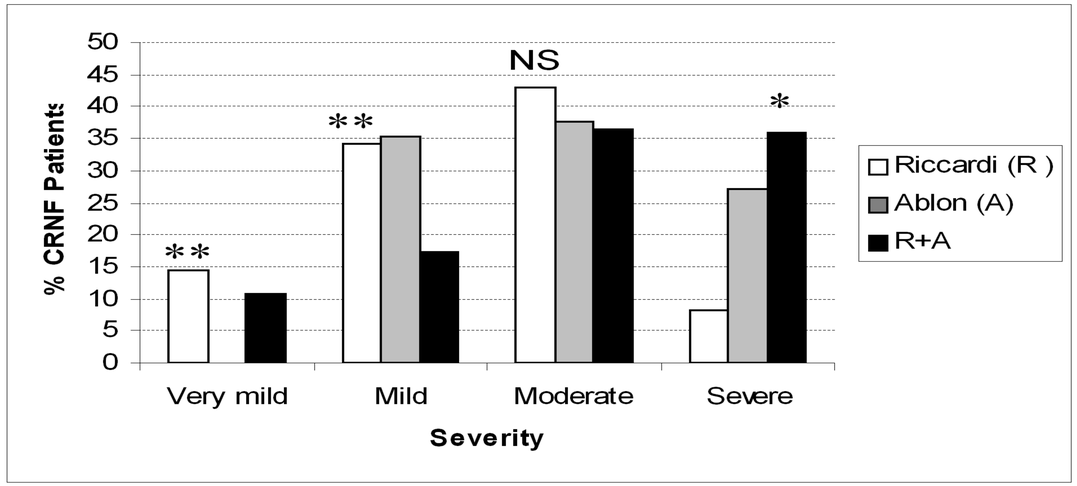
Moderate form

plexiform neurofibromas, controled seizures, pseudoathrosis, hypertension, scoliosis - features were visible at first glance

Severe form

problems leading to major health impairments, often requiring surgical intervention, intracranial and spinal tumors, malignancy, pheochromocytomas, uncontroled seizures, mental retardation, hydrocephalus, progressive e diffuse hypertrophic growth - features were easily found at a first glance.

Results: When the modified Riccardi plus Ablon criteria were used, a greater number of NF1 patients were classified in the severe form (35. 9%) than when Riccardi´s or Ablon´s scales were used separately. Nevertheless the scale used, most of the CRNF patients were classified in the moderate and severe forms.



Conclusion: The modified R+A criteria seem to be reasonable and useful to evaluate the severity of NF1 patients. Using these criteria NF1 apparently is more severe and probably should have a greater impact on patient's quality of live than it is usually recognized .

This study was supported by CNPq, FAPEMIG and AMANF