Some under-reported clinical complications in NF1 patients

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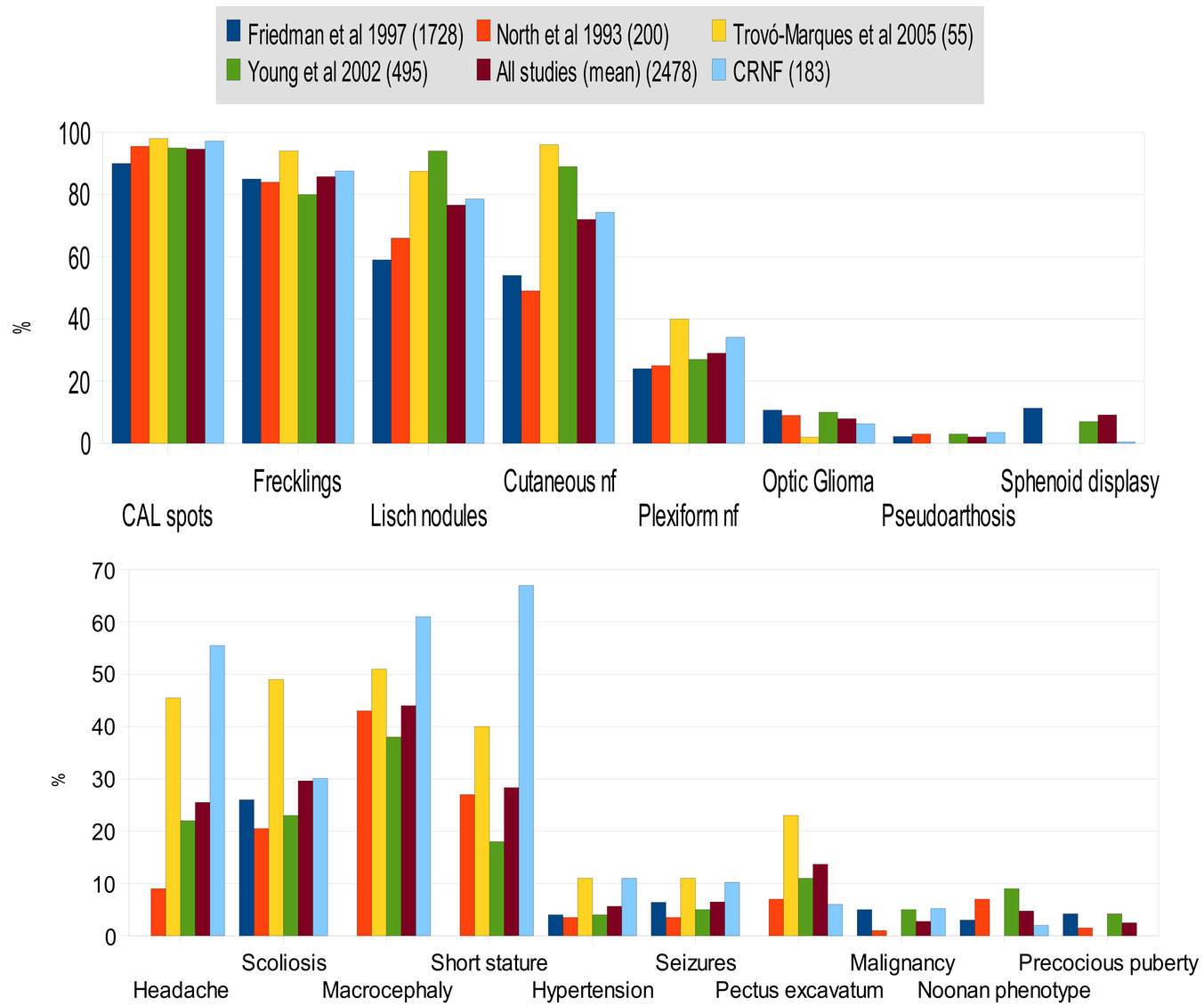
Objectives: Neurofibromatosis type 1 (NF1) had its wide variety of clinical manifestations recorded in 183 patients presented at the CRNF and the results were compared with international reports. The diagnostic criteria of NF1 was established according to the NIH, based on clinical examination and laboratory data.

Results: 58% of the patients were females and the mean age was 27 years (range: 1-67 years) and 45.2% had a first degree parent affected.The main clinical features diagnostic criteria are showed in the upper part of Figure. The commonest associated health problems are showed in the lower part of Figure.

Conclusion: The prevalence of clinical diagnostic criteria among these Brazilian NF1 patients were comparable to those reported by other studies, but some mild complications have not been described before: facial assimetry (59%), muscular hypotony (32%); or have been under-reported: chronic headache (55.5%), short stature (67% - men; 39% - women), machrocephaly (61% - men; 34% - women) and voice and speech disturbance (56.7%).

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