Prevalence of an unreported Neurofibromatosis type 1 phenotype: preliminary results

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Background - Early diagnosis of neurofibromatosis type 1 (NF1) is useful in clinical managements of learning difficulties, nervous system tumors, osseous dysplasia and neurofibromas. Most NF1 new mutation patients present at birth with only multiple café aux lait spots (CALS), only one of the seven diagnostic criteria. Any other specific congenital NF1 signal could be interesting to assure NF1 diagnose. During evaluation of nearly 600 NF1 patients, some of them presented the second toe uprighting and nearly superposing on the third toe (it was called Second Toes Signal – STS, Figure 1). We did not found STS previous report, regardless of its association with NF1. The STS was clearly different from syndactily and it was bilateral in all the cases, except in a NF1 patient with segmental NF1.

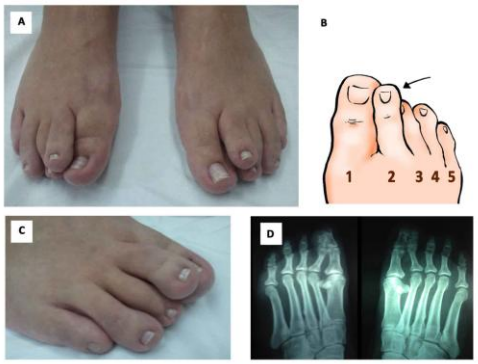

Figure 1 – A) Frontal view of a NF1 patient feet (34 y, female); B) Drawing of the Second Toes Signal; C) Lateral view of the right foot; D) Patient's feet RX.

Aim - To compare NF1 STS prevalence with healthy individuals.
Methods - A questionnaire (STS picture included) was sent by mail to 445 NF1 patients (at least three diagnostic criteria) and by e-mail to 86 non-NF1 volunteers with the question: Are your feet like that?
Results – In valid (39.2%) mail 167 answers, NF1 STS prevalence was 11.9% (11.1% men and 12.9% women, p>0.05). Of the 60 e-mail answers, only two persons answered positively (3.3%) (bicaudal chi-squared test p=0.0503): one of them actually showed halux valgus and the other one had STS exclusively in the left foot. Further studies are needed to confirm STS as a specific signal of NF1. If true, STS would be useful because it is present at birth (before the usual age of appearance of freckling, optical glioma, cutaneous neurofibromas and Lisch nodules) and it is more prevalent than osseous dysplasias. Moreover, the STS is easily diagnosed from clinical examination, without the need of further exam procedures, some of which would require anesthesia for younger patients.
Conclusion – STS is probably a specific congenital NF1signal.

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