Author Response: Choroidal Abnormalities Detected by Near-Infrared Imaging (NIR) in Pediatric Patients With Neurofibromatosis Type 1 (NF1)

We appreciate the comments by Vagge et al.[1] on our article on choroidal abnormalities related to neurofibromatosis type 1 (NF1) in pediatric patients, aimed at evaluating the diagnostic performance of this new clinical sign as a diagnostic criterion for NF1.[2] The authors have published an interesting paper on this topic, not included in our discussion because it was unavailable at time of manuscript preparation.[3] The results of Vagge et al.[1] agree with our data. Both studies conclude that NF1-related choroidal abnormalities have a higher diagnostic accuracy compared with many of the standard National Institutes of Health diagnostic criteria, including Lisch nodules.[4] Although Vagge et al. reported that the detection of choroidal abnormalities was possible also in children 2 years of age, in our paper we have demonstrated that the main technical issue in the detection of NF1-related choroidal abnormalities is the amount of cooperation required to obtain adequate choroidal images in very young children.[2] Notwithstanding, the main advantage of this sign seems to be the possibility to anticipate and make more sensitive the diagnosis of NF1, whereas the main obstacle is age-related cooperation.

In recent years, many new clinical clues related to NF1 have been reported, including cutaneous signs (anaemic nevi, juvenile xanthogranulomas, mixed vascular hamartomas and cherry angiomas, hypochromic macules, "soft touch" skin, hyperpigmentation) and extracutaneous findings (choroidal hamartomas, a large head circumference, unidentified bright objects on brain magnetic resonance imaging, and the typical neuropsychological phenotype).[5] When validated, these new diagnostic findings may improve the diagnosis of NF1.

We have also recently demonstrated that the ophthalmoscopic assessment of fine optic disc alterations (or the challenging evaluation of fluctuating visual acuities) in small children affected by NF1, to detect the presence of optic pathway glioma, may be substituted by the more precise and repeatable retinal nerve fiber layer assessment by optical coherence tomography.[6] We hope that also the challenging evaluation of Lisch nodules will be shortly replaced by simple and unambiguous evaluation of NF1-related choroidal abnormalities detected by near-infrared imaging modalities. Therefore, we agree with Tadini et al.[5] that the time has come for a new consensus conference of clinicians and molecular biologists, in order to establish a new list of age-related diagnostic criteria that can be formally approved by all specialists involved in managing NF1 patients.

Raffaele Parrozzani[1]
Edoardo Midena[1,2]

[1]G. B. Bietti Foundation, IRCCS, Ocular Oncology and Toxicology Research Unit, Roma, Italy; and the [2]Department of Ophthalmology, University of Padova, Padova, Italy.
E-mail: edoardo.midena@unipd.it

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