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Case report: Choroidal abnormalities in neurofibromatosis type 1

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Neurofibromatosis type 1(NF1) is an autosomal dominant neurocutaneous disorder. It is a relatively rare disease, affecting 1 in 3000 births. It can affect almost all organs of the body, but most commonly affects the skin, bones and nervous system. The eye and adnexa are frequently involved in NF1. Iris Lisch nodules, optic gliomas, eyelid cafe-au-lait spots are diagnostic hallmarks in NF1. In recent years new manifestations have been described in the ocular system in NF1, due to recent progress in multimodal imaging in ophthalmology. These are choroidal abnormalities, hyperpigmented spots and retinal vascular abnormalities. Choroidal abnormalities (CAs) have been added to the actual diagnostic criteria for NF1. They are not added as a separate criterion but introduced as an alternative to the presence of iris Lisch nodules. They have the prevalence between 64 and 98%. CAs, as described in histopathological studies, are ovoid bodies, consisting of hyperplastic Schwann cells, neural crest-derived melanocytes and ganglion cells. CAs are asymptomatic and undetectable with conventional ophthalmoscopic examination or by means of autofuorescence and fuorescein angiography. However, they are visible as bright, pathcy nodules on near infrared reflectance images (NIR) and spectral-domain optical coherence tomography (SD-OCT). We present a patient with NF1 and choroidal abnormalities which were detected for the first time by using NIR and SD-OCT.

Prikaz primera: Spremembe žilnice pri nevrofibromatozi tipa 1

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Nevrofibromatoza tip 1 (NF1) je avtosomno dominantna nevrokutana bolezen. Je relativno redka, s pojavnostjo 1 na 3000 rojstev. Prizadene lahko skoraj vse organe, najpogosteje kožo, kosti in živčevje. Pogosto so prizadete tudi oči in adneksi. Prisotnost Lischevih vozličev šarenice, gliomi vidnega živca, cafe-au-lait lezije na vekah so diagnostični kriteriji za NF1. Razvoj slikovnih diagnostičnih metod je v zadnjih letih omogočil prepoznavo novih očesnih sprememb. To so spremembe v žilnici, hiperpigmentacije in nepravilnosti mrežničnega žilja. Spremembe žilnice so dodane k diagnostičnim kriterijem za NF1 kot alternativa za prisotnost Lischevih vozličev šarenice. Njihova prevalenca je 64- 98%. Histopatološko so spremembe žilnice ovoidna telesca, ki jih tvorijo hiperplastične Schwannove celice, melanociti nevralnega grebena in ganglijske celice. Spremembe so asimptomatske in jih z običajnim oftalmološkim pregledom, slikanjem avtofluorescence ali s fluoresceinsko angiografijo ne zaznamo. S slikanjem očesnega ozadja s svetlobo blizu infrardeče (NIR) in z optično koherenčno tomografijo (SD-
OCT) pa spremembe jasno vidimo kot svetle vozliče oziroma hiperreflektivne spremembe v žilnici. Predstavili bomo primer pacientke z znano NF1 in prvič diagnosticiranimi spremembami v žilnici.