



CHOROID FINDINGS IN VON RECKLINGHAUSEN DISEASE (NF1)

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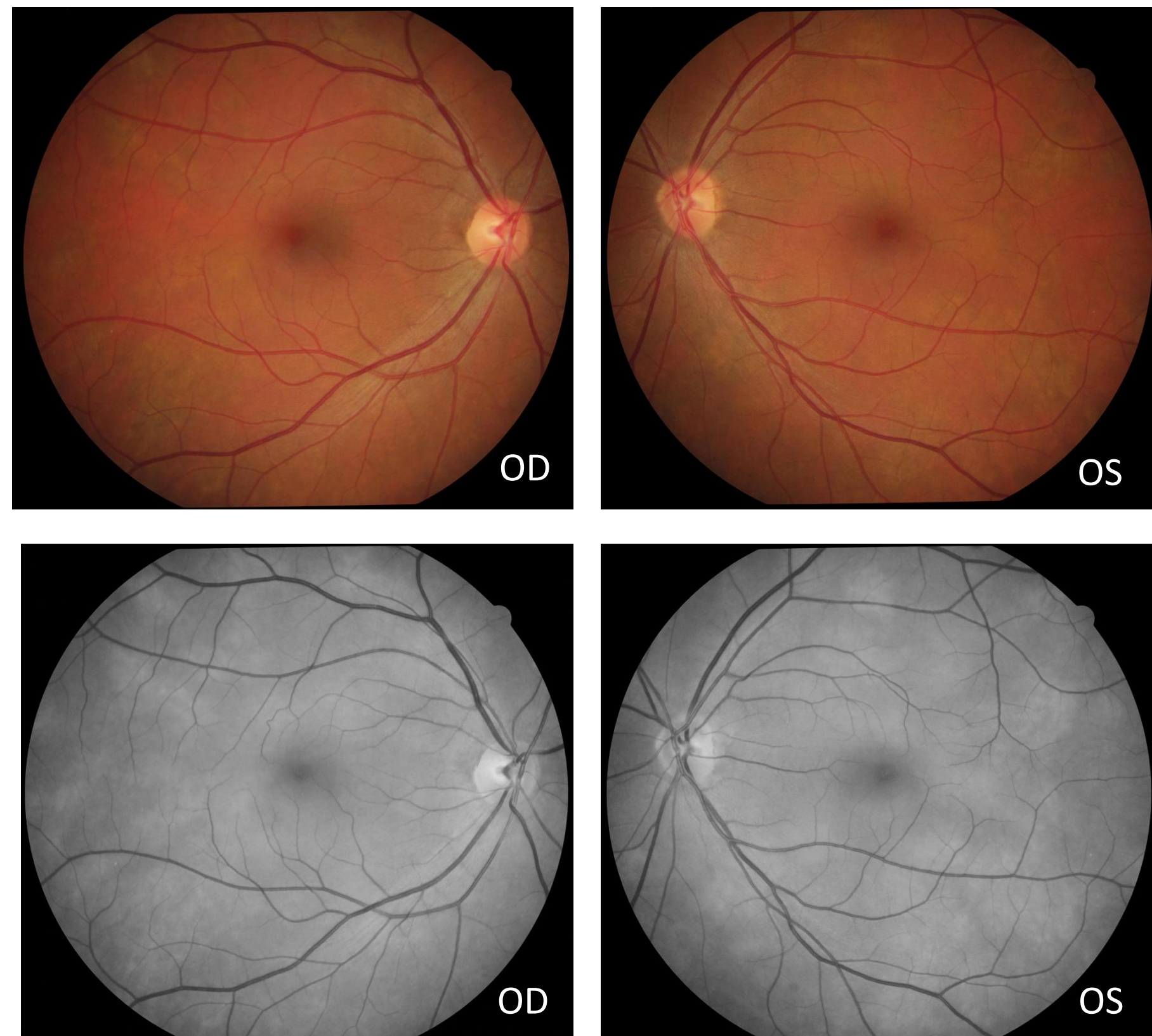


PURPOSE

Choroid nodules (Yasunari) are not identified in conventional eye examination and are rarely studied in the literature. The case report describes the findings of choroidal nodules and their importance in the diagnosis of Neurofibromatosis type 1 (NF1).

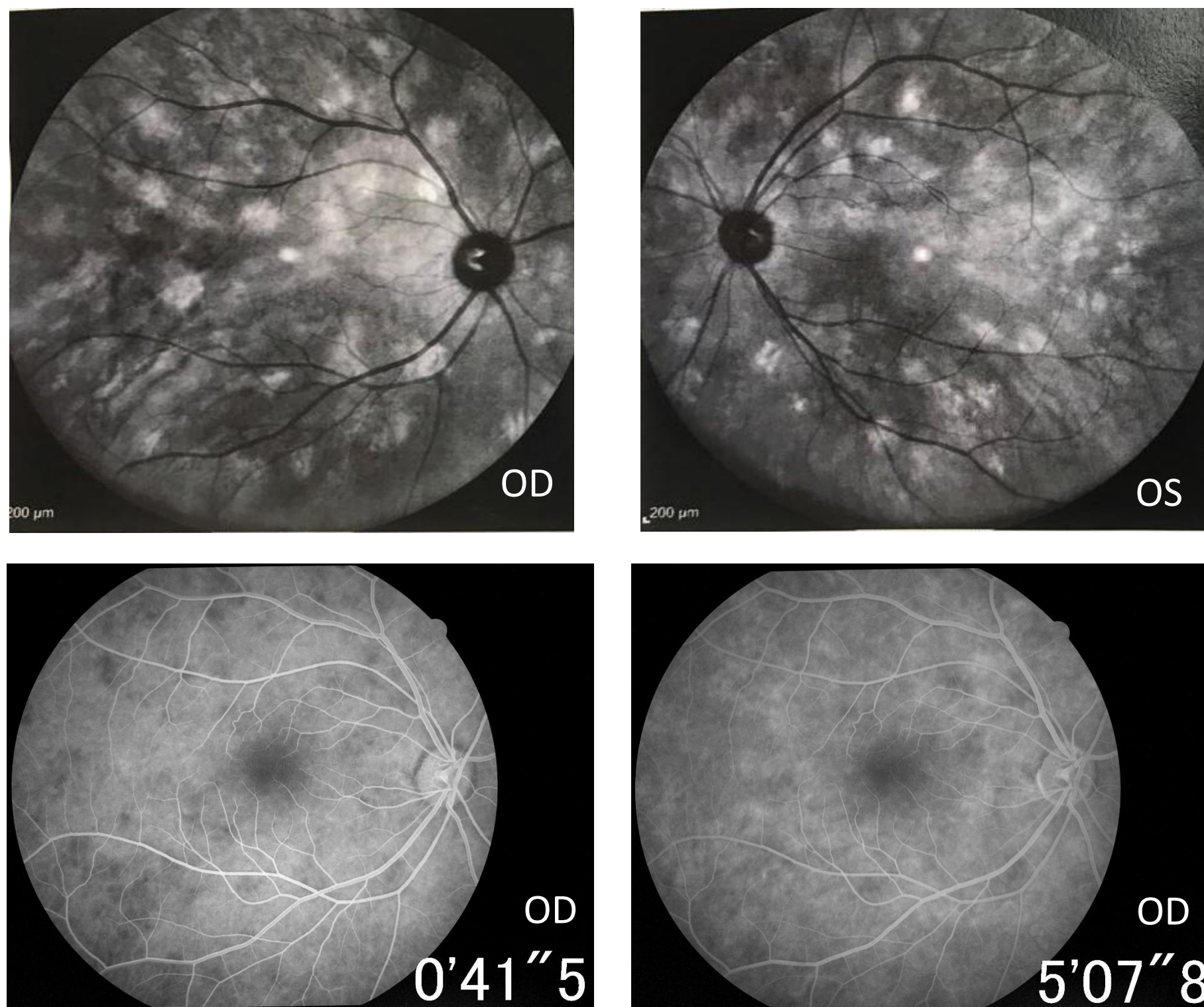
METHODS

Information was obtained through review of medical records, multimodal imaging and literature review.



RESULTS

MR, 50 years old, female, asymptomatic, mentioned a previous diagnosis of NF1, however, denied the presence of ocular manifestations. Her vision was 1.0 OU. IOP: 15/13mmHg. Biomicroscopy: cutaneous neurofibromas, multiple lisch nodules OU. Color retinography: Normal colored optic nerve, physiologica papillary excavation, intact vessels and macula, subretinal irregular pigment spots and applied retina. Red-free image: no significant changes. Near Infra-Red: spots with high reflectance. OCT: irregular hyperreflective areas in the choroid, with compression of overlying choroidal vessel. FA: multiple lesions with early hypofluorescence and hyperfluorescence by staining in the late phase.



DISCUSSION

NF1 is a genetic disease, autosomal dominant with mutation in NF1, chromosome 17q11.2, with multisystem involvement and clinical heterogeneity. The most common ocular feature is Lisch nodules. Other are optic gliomas, orbital neurofibromas, café-au-lait spots on the eyelids and congenital glaucoma. Recent studies have observed that the frequency of choroidal nodules is higher than that of Lisch nodules. They are defined as ovoid bodies of hyperplastic Schwann cells, melanocytes and ganglion cells. Due to the high melanin content, choroidal nodules are better observed through infrared reflectance, resulting in strong absorption and subsequent backscattering of light, seen as bright and irregular areas. OCT can demonstrate two morphological patterns: "dome-shaped" and placoid. After a review by an international consensus, choroidal nodules were recently added to the diagnostic criteria as an alternative to the presence of Lisch nodules.

