Seeing Spots: An Unusual Presentation of Excessive Bilateral Choroidal Nevi in a Likely Neurofibromatosis Patient

This case report of excessive bilateral choroidal nevi in a presumed neurofibromatosis type 1 patient will emphasize the importance of detailed investigation, neurological imaging and patient education. Treatment and management, including photos, will be included.

I. Case History

CC: 59 YOWF presents for doctor requested DFE.

**Ocular hx:** Recurrent subconjunctival hemorrhages; hx of “dots” in back of eyes. Patient has been informed by multiple doctors to have regular colonoscopies to screen for colon cancer.

**Medical hx:** Brain aneurysm and stroke 18 years ago; hypothyroidism

**Family hx:** No neurological conditions reported; sister is being followed for similar “dots” in her eyes.

**Meds:** Acyclovir, Levothyroxine, Naproxen, Warfarin

Patient has nevi covering both arms and both legs. Patient also has one nevi >15mm in size on her temple.

Patient denies any freckling in axillary area or groin.

Patient also denies any raised nodules on skin.

II. Pertinent findings

**BCVA 20/20 OD, OS**

Pupils, CVF, and EOMs were all normal.

Biomicroscopy unremarkable OD except irises. A couple areas of pigment clumping noticed on iris with minimal elevation, OD.

Resolving subconjunctival hemorrhage inferior nasal OS with 1.5 mm nevus superior on iris. Several areas of pigment clumping on iris with minimal elevation, OS. All other anterior segment findings unremarkable.

Posterior segment:

**OD:**
C/D ratio 0.55/0.55, large nerve with laminar dots

Macula: Flat and intact, no drusen

Background: Clear and uniform with no vasculopathy

Intact retina 360 with multiple nevi (>10) in every quadrant along arcades

**OS:**

C/D ratio 0.50/0.50, large nerve with laminar dots

Macula: Flat and intact, no drusen

Background: Clear and uniform, no vasculopathy, choroidal nevus superior nasal to ONH

Intact retina 360 degrees with multiple nevi (>10) in every quadrant along arcades

No retinal hemorrhages, OD and OS

**III. Differential diagnoses**

Neurofibromatosis Type 1

Gardner Syndrome with multiple CHRPE

Physiological variant of choroidal nevi

**IV. Diagnosis and discussion**

Potency of risk of tumor formation due to excessive bilateral nevi

Classic presentation of Neurofibromatosis Type 1

At least two Lisch nodules

At least 6 café au lait spots >15 mm in size in adults

Optic nerve glioma

Two or more skin neurofibromas

Axillary and/or groin freckling

Sphenoid dysplasia or long bone abnormalities
First degree relative with NF1

Multiple nevi versus multiple CHRPE

Comparison of nevi, CHRPE and choroidal melanoma

Mosaic neurofibromatosis only affecting specific tissues

V. Management

Genetic testing

Photo documentation

Neurological imaging

Regular follow-ups with PCP and eye care provider

VI. Conclusion

Clinical pearls of Neurofibromatosis

Recognizing ocular and dermatological signs of Neurofibromatosis

Detailed family history

Patient education about disease and possible tumor formation

Bibliography/Sources