Purpose: To describe the genetic analyses of two independent choroidal melanomas, a malignant melanoma and a malignant melanoma arising from a nevus, in the same eye.

Method: A 52-year-old woman presented with a symptomatic, large pigmented choroidal melanoma in her left eye. Family cancer history was negative and the right eye was unremarkable. Best-corrected visual acuity was 20/150 OS and intraocular pressure was 16 mmHg OS. Ultrasonography detected a mushroom-shaped tumor measuring 10.6mm in diameter and 11.2mm in thickness with overlying subretinal fluid and intravascular pulsations, and no extrascleral extension (lesion 1). Further ocula examination revealed a 9mm ciliary body mass superiorly (lesion 2). Enucleation and was performed and specimens were sent for immunohistochemical and genetic analyses.

Results: The enucleated specimen revealed two primary lesions confirmed by serial sections. Lesion 1 was a malignant melanoma, predominantly spindle cell type with formation of cavitation within the herniated component. Genetic studies showed GNAQ mutation and copy number gain on Chromosome 17q21-25. Lesion 2 was a malignant melanoma, predominantly spindle cell type with pronounced vasculogenic mimicry, arising from a nevus. This tumor was positive for GNAQ mutation, and copy number gain on Chromosome 17q22-25 and loss on 14q23-32. Bap1 mutation was negative.

Conclusion: Cases of two distinct uveal melanomas in one eye, with one arising from a nevus, are extremely rare. Cytogenetic analyses of these types of cases may help us better understand the pathogenesis of uveal melanomas.