Uveal melanoma in congenital ocular melanocytosis mimicking malignant transformation of the optic disc melanocytoma (A controlled case)

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Abstract

A case of 57-year old woman with uveal melanoma of the posterior pole arising in the congenital ocular melanocytosis mimicking clinically malignant transformation of the optic disc melanocytoma is presented.

CASE REPORT

Introduction

Congenital ocular melanocytosis (COM) is a rare condition in white population (0.04%), more common in yellow and black populations. It is characterized by the pigmentation of the episclera, sclera, anterior chamber angle structures, uveal tract (iris, ciliary body, choroid) and optic nerve. Orbital fat and periosteum (orbital melanocytosis) or menings and central nervous system (leptomeningeal melanocytosis) also can be affected in some patients. When periocular skin is affected as well, the condition is termed oculodermal melanocytosis or nevus of Ota.

The tissues from the above sites are infiltrated by the dendritic melanocytes which can vary in number from so scarce up to so numerous.

The COM is nonhereditary and can be unilateral (most of cases) or bilateral.

On anterior segment in COM we find flat gray or brown areas of episcleral and scleral pigmentations, velvet-like dark brown iris and hyperpigmentation of the anterior chamber angle. The background fundus pigmentation is greater and often we can see degeneration of the overlaying retinal pigment epithelium and numerous drusen.

In white patients there is a high susceptibility for development of uveal melanoma (1–2%). They have 20-fold increased risk for uveal melanoma (UM). It usually arises in the choroid or ciliary body. None of published examples was situated on the optic disc. The melanomas in COM don´t appear at an earlier age than average [1,2,8,9].

Because of the increased incidence of the glaucoma, optic disc melanocytoma (ODMC) and UM, patients with COM must be examined regularly.

Also other conditions like iridocorneal endothelial syndrome, Fuchs iridocyclitis, juvenile xantogranuloma, melanocytic tumors of the iris and topical prostaglandin therapy can cause iris heterochromia. Other disease that can resemble COM is bilateral diffuse uveal melanocytic proliferation.

Case Report

A 57-year old Caucasian woman was presented with the normal left eye, on the right eye there was velvet-like dark brown iris (Figure 1), patchy episcleral pigmentation (Figure 2) and epipapillary, retinal and juxtapapillary choroidal pigmented lesion (Figure 3). VOD was 4/6 (spherical equivalent, SE of +0.5 Dpt), VOS was 4/4 (SE of +0.5 Dpt). Fluorescein angiography (FA) revealed hyperfluorescence of the choroidal component with its late leakage and blocked fluorescence by the retinal portion. Well demarcated blocked fluorescence of the lesion with hyperfluorescent margin was on indocyanine green angiography (ICGA) (Figure 4). It was much larger than biomicroscopical appearance of the tumor base. The dome shaped lesion with higher internal reflectivity measuring 1.3 mm in thickness was found on standardized ultrasonography (US) (Figure 5). Enlargment of the blind spot was documented on perimetry. There was subretinal fluid in the upper part of the macula on optical coherence tomography (OCT) (Figures 6, 7).

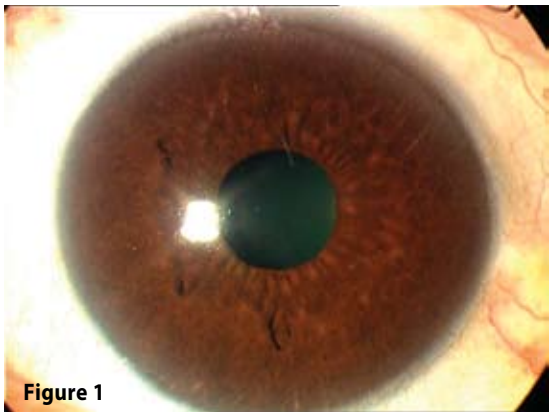
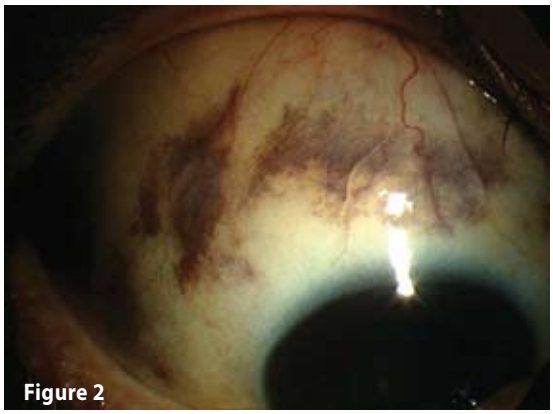


Figure 1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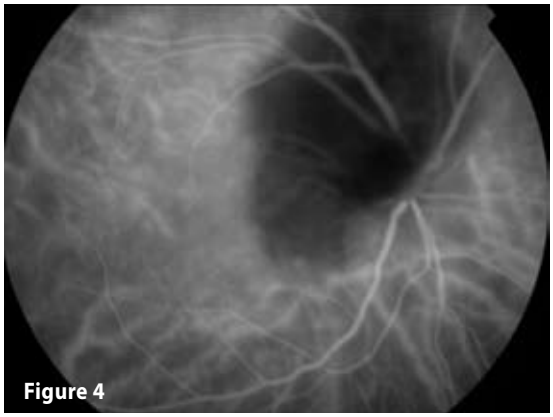
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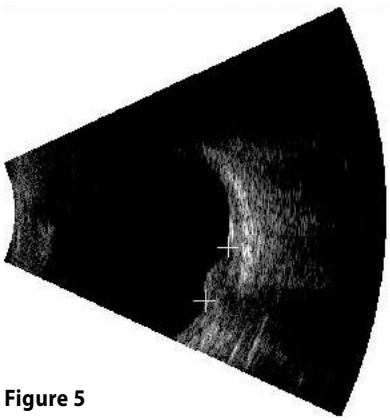
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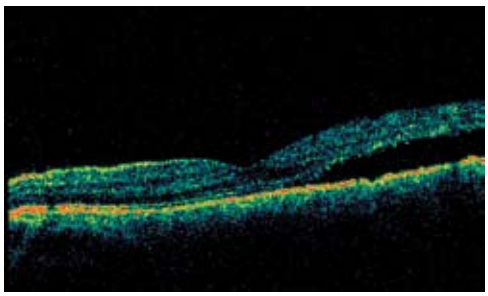
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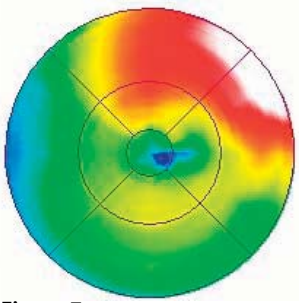
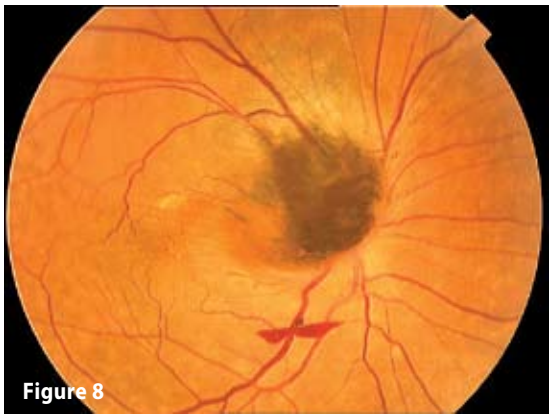


Figure 7

Figure 1. Velvet-like dark brown iris of the right eye.
Figure 2. Patchy episcleral and scleral pigmentation of the right eye.
Figure 3. Epipapillary, retinal and juxtapapillary choroidal pigmented lesion of the right eye at the first examination.
Figure 4. Well demarcated blocked fluorescence of the lesion with hyperfluorescent margin on ICGA at the first examination.
Figure 5. US B-scan: dome shaped lesion at the first examination.
Figure 6. OCT (90° scan): subretinal fluid in the upper part of the macula at the first examination.
Figure 7. OCT retinal map with apparent neuroretinal thickening in the upper part of the macula at the first examination.



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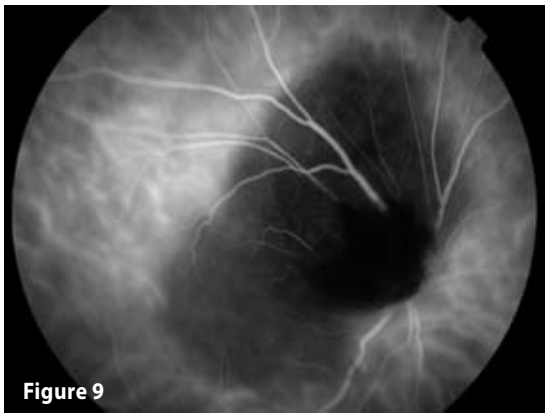

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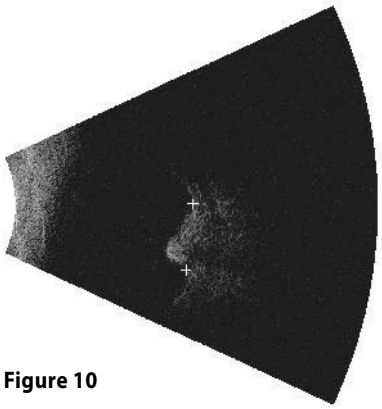

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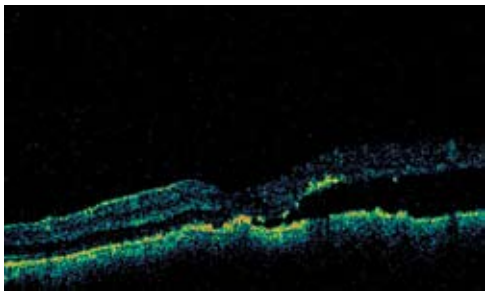

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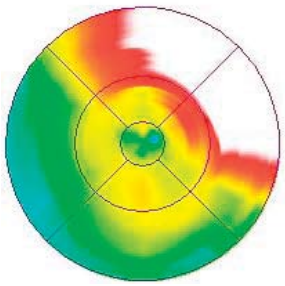
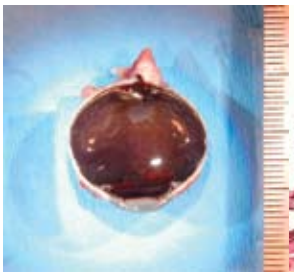


Figure 12

Figure 8. Color fundus photograph with the marked growth of the lesion and with the preretinal haemorrhage at 18-month visit.
Figure 9. ICGA at 18-month visit: enlargement of the blocked fluorescence.
Figure 10. US B-scan at 18-month visit: dome shaped lesion.
Figure 11. OCT (90° scan) at 18-month visit: pronounced neuroretinal thickening.
Figure 12. OCT retinal map at 18-month visit: extension of the neuroretinal thickenning in the upper part of the macula.



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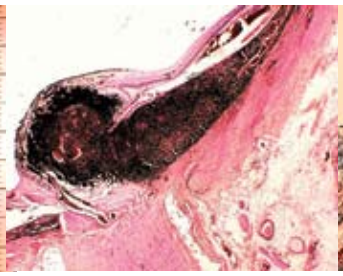

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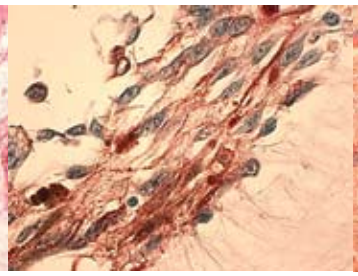

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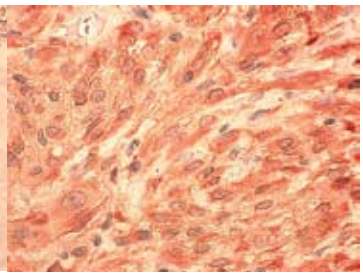

Figure 16

Figure 13. Gross photo of the fixated and sectioned enucleated eyeball. Overall heavy pigmentation of the eye consistent with diagnosis of COM is visible, as well as pigmented tumor of the posterior pole.

Figure 14. Histological section through the posterior pole tumor showing its choroidal portion, prepapillary and retinal extension (paraffin section, hematoxylin-eosin (HE) staining, magnification × 12.5).

Figure 15. Bleached section through the retinal portion of tumor with spindle B melanoma cells, with their positivity for S100 protein (paraffin section, magnification × 500).

Figure 16. Bleached section through the choroidal portion of tumor. Spindle B melanoma cells with imunohistochemical positivity for HMB 45 (paraffin section, magnification × 500).

Magnetic resonance imaging demonstrated no orbital or intracranial abnormality. A chest radiogram, abdominal ultrasonograph, liver tests, blood count and urine were within normal limits. She had arterial hypertension and had undergone thyreoidectomy 8 years ago.

There were performed following examinations: best corrected visual acuity (BCVA), slit lamp, intraocular pressure, biomicroscopy, FA, ICGA, US, perimetry, OCT and body organs testing during the 18-month follow-up regularly.

At 6-month and 12-month visits no tumor progression was found except slight enlargement of blocked fluorescence of the choroidal component of the lesion on the ICGA and blocked fluorescence of the epipapillary and retinal portions on FA. The BCVA was stable, 4/5. Marked growth of the lesion (biomicroscopically, on FA

and ICGA) (Figures 8, 9) with preretinal haemorrhage appeared at 18-month visit. BCVA decreased to 4/20 (SE increased to +1.25 Dpt). Tumor thickness was 3.6 mm on US with lower internal reflectivity (Figure 10). Centrocoecal scotoma and the defect in the upper nasal quadrant of the visual field was found on perimetry. Amount of the subretinal fluid on OCT increased (Figures 11, 12).

After the biopsy confirming suspicion from uveal melanoma the enucleated eyeball was throughly histopathologically examined (Figure 13). Melanosis was observed within the iris, ciliary body, choroid, episclera, sclera and optic nerve sheaths. The tumor of the posterior pole composed of the three main parts: choroidal portion, epipapillary nodule and retinal portion (Figure 14). Heavy pigmented spindle B malignant melanoma cells formed all three parts of the tumor, no residual structures of the melanocytoma were found. The tumor extended from its uveal portion at the temporal edge of the optic disc around the edge of Bruch´s membrane to its epipapillary and retinal portion. Immunohistochemistry revealed positivity of the tumor cells for S100 protein (Figure 15) and for HMB 45 (Figure 16). No evidence of extrascleral extension of the tumor was found.

No metastasis was found before enucleation and 9 months after that.

Discussion

ODMC is more frequently found in COM as compared to general population. The lesions are almost dormant clinically and does not demonstrate the active proliferation that characterizes most UM.

There were published several well documented cases of UM arising from ODMC. None of them had COM [3,4,5,6,7].

Our case with COM looked clinically as ODMC in the begining, then we suspected its malignant transformation. Finally on histopathology no melanocytic cells were found.

A wide range of diagnostic tools helped to monitor the disease very closely. OCT showed suspected activity of the lesion, so ICGA was indicated. Discrepancy between biomicroscopic and angiographic appearence of the tumor base extention was found.

In conclusion, uveal melanoma mimicking malignant transformation of the optic disc melanocytoma can be found also in eyes with congenital ocular melanocytosis.

This case was presented at Congress of Czech Society of Ophthalmology in 2006.

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