Risk factors for disseminated protothecosis include human immunodeficiency virus, leukemia, malignancies, hemodialysis, corticosteroid therapy, and catheterization. The patient in the current study was receiving long-term immunosuppressive therapy for graft-vs-host disease. These factors likely led to the development of disseminated protothecosis.

In summary, we present, to our knowledge, the first human case of protothecosis with ocular involvement in a dog.

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Retinal Ischemic Syndrome, Digestive Tract Small-Vessel Hyalinosis, and Diffuse Cerebral Calcifications: A Pediatric Observation of a Rare Syndrome

We describe a pediatric case of a multisystem disorder involving the retina, the brain, and the digestive tract.

Report of a Case. A male child born of unrelated parents in 1985, with 2 healthy brothers, developed normally until age 27 months, when he experienced a hemiconvulsive attack. Major bilateral calcifications were seen on the computed tomographic brain scan (Figure 1A). Carbamazepine was prescribed for 8 years; the patient had no relapses, but physical and psychomotor development was subnormal. Developmental fetopathy was diagnosed.

At age 11 years, the patient had several grand mal seizures followed by severe digestive tract hemorrhages. At this point, ocular fundus examination was performed and mild peripheral retina ischemic changes were observed, with microaneurysms and shunt vessels developing. A computed tomographic brain scan was unchanged from the first one. Magnetic resonance imaging showed diffuse high signal intensity (Figure 1B). Abdominal sonography–Doppler examination showed moderate splenomegaly and portal hypertension. Endoscopic examination showed a watermelon stomach (diffuse antral vascular ectasia). Biopsy specimens from the digestive tract showed basement membrane thickening and thickening of small vessels by hyalin deposits in the endothelial cells. A skin biopsy specimen showed accumulation of deposits of membranelike osmiophilic material beneath endothelial cells and around pericytes and smooth muscle cells. A liver biopsy specimen showed macronodular cirrhosis. The patient was fully studied for coagulopathy and hemoglobinopathies, and results were negative. Empirical therapy with β-blockers and octreotide allowed

Figure 5. Agarose gel electrophoresis of polymerase chain reaction analysis of right macula confirms presence of Prototheca wickerhamii DNA. MW indicates molecular weight standards; NC, water-only control; PT, patient sample; PC, positive control (from P wickerhamii serum culture). Two independent, nonoverlapping primer sets were used.

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clinical improvement, with fewer digestive tract hemorrhages. Growth was stunted (−3 SDs) and the patient’s hair became thinner, grayish, dry, and fragile. His skin was pale and atrophic with areas of poikiloderma.

After 4 years of relative clinical stability, ophthalmologic examination showed visual acuity of 20/40 OD and 20/30 OS. Results of slit-lamp examination and intraocular pressure were normal in both eyes. Fundus examination showed sharp but slightly pale optic nerves and major retinal ischemic changes, with focal preretal hemorrhages. Fluorescein angiography showed mild cystoid macular edema in the right eye and normal macula in the left eye (Figure 2A and B), along with major vessel closure in the retinal periphery (Figure 2C and D) and focal neovascularization (Figure 2D). Peripheral retinal laser ablation was performed to decrease the risk of vitreous hemorrhage. Three years later, visual acuity remained stable at 20/40 OD and 20/30 OS, with no funduscopic macular changes. New peripheral preretal hemorrhages caused by new areas of ischemia prompted further photocoagulation.

The patient underwent circulating endothelial cell and endothelial microparticle enumeration. Circulating endothelial cell count was performed with 1 mL of whole blood and immunomagnetic beads (Dynal-SHAM-M 450; Biosys, Compiègne, France) coated with S-Endo I, a specific antiendothelial monoclonal antibody developed in our institution. For the endothelial microparticle count, 30 µL of plasma was incubated with fluorescein isothiocyanate–annexin V, which binds to phosphatidylerine present on the microparticle surface (Flow-Count Fluorospheres; Beckman Coulter, Fullerton, Calif) in peripheral blood. Results showed circulating endothelial cell values significantly higher than normal (110 vs 5 cells/mL) but normal endothelial microparticle values (3/µL).

Comment. This appears to be the first pediatric case of a rare syndrome, previously described in only 3 members of 1 family in western France and 1 unrelated patient from the same region. This syndrome comprises digestive tract and renal small-vessel hyalinosis, idiopathic nonarteriosclerotic intracerebral calcifications, retinal ischemic syndrome, and phenotypic abnormalities. Two of these patients died at ages 27 and 36 years of cachexia and subarachnoid hemorrhage, respectively, and all 4 manifested kidney failure due to glomerular endothelial cell alterations and mesangiolyysis, a complication that our patient does not show at present. These patients, along with ours, all had digestive tract lesions with hyalinosis.

Our patient showed a high level of circulating endothelial cells in peripheral blood, suggesting desquamation of cells, which then appeared to be causing focal occlusions. Normal endothelial microparticle count suggests a mechanical abnormality rather than an inflammatory one (ie, vasculitis).

This syndrome shares some signs with other known syndromes, including cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy and hereditary endotheliopathy with...
retinopathy, nephropathy, and stroke, but it is distinct in that the digestive tract is not involved in those 2 syndromes, which have been mapped to 19q12⁴ and 3p21, respectively. In our patient, genetic study showed a normal karyotype (standard metaphase spread). Because the patient is the only member of his family to manifest the syndrome, no linkage mapping has been performed to date.

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Maculopathy and Retinal Degeneration in Cobalamin C Methylmalonic Aciduria and Homocystinuria

The cobalamin C form of methylmalonic aciduria and homocystinuria (Cc-CMAH) is an inherited deficiency of the 2 coenzymatically active vitamin B₁₂ derivatives, methylcobalamin and deoxyadenosylcobalamin (Figure 1). Its heterogeneous clinical manifestations include feeding difficulties, neural dysfuncion, and ophthalmic abnormalities. The ophthalmic features are visual impairment, nystagmus, and retinopathy with conspicuous maculopathy.¹² The mechanism by which the biochemical abnormalities cause retinal disease has not been defined. We analyzed photoreceptor cell and postreceptorial...