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Black kidney in Albinism

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Implication for health policy/practice/research/medical education:
Oculocutaneous albinism may be similar to two related syndromes (Hermansky-Pudlak and Chediak-Higashi) and could lead to more widespread lysosome excretory defects. These defects could lead to accumulation of some intracellular material, leading to the gross discoloration of the kidney.

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Brief Report

1. Case

A 27-year-old man with Oculocutaneous Albinism (OCA) underwent surgery to donate his kidney to a 16-year-old boy with end stage renal disease. Just after exposing his left kidney, transplant surgeon and his team noted a strange completely black appearance of the kidney, which was very similar to a gangrenous organ. They made some pictures from this strange finding and without taking any biopsy sutured the incision (Figure 1). Then, the recipient was returned back to the recovery unit. Before operation, the donor had undergone profound workup, and there was not any abnormal finding. His serum creatinine was 0.7 mg/dl; hemoglobin level, 14.2 mg/dL; hematocrit, 39.3%; platelets, 246000/μL; fasting blood sugar, 99 mg/dL; serum uric acid, 5.9 mg/dL; cholesterol, 204 mg/dL; triglyceride, 116 mg/dL; AST, 18 IU/L(0-31); ALT, 28 IU/L (0-31); bilirubin, total: 0.5 mg/dL (0.1-1.2); bilirubin indirect, 0.4 mg/dL (0.1-0.4); LDH, 317 IU/dL (up to 480); and serum albumin, 4.0 g/dL. Plain and color Doppler ultrasound studies of the kidneys were normal. Renal scan with 99 mTc-DTPA that was performed before surgery revealed left kidney GFR of: 50 ml/minute, and right kidney GFR of: 59 ml/minute, with good excretory function on both sides. Urine analysis was normal. Urine protein was negative, white blood cells; 1-2/ high power filed, and red blood cells: 1-2/ high power filed. Intravenous pyelography and CT-angiography which were performed before surgery were completely normal and both left and right kidneys had single artery without any accessory branches or any polar arteries. He also underwent future follow up after surgery and he had a normal clinical and laboratory findings up to one year of follow-up.

2. Discussion

Hermansky-Pudlak syndrome (HPS) and Chediak-Higashi syndrome (CHS) are two rare autosomal recessive disorders, that share the clinical findings of OCA, a disorder of lysosomal

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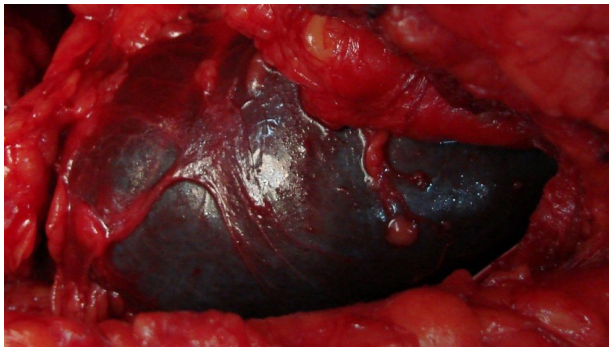

Figure 1. Black kidney was discovered in the operating room when a healthy 27-year-old man with Ocular-Cutaneous Albinism(OCA) underwent surgery to donate his kidney to a 16-year- old boy with end stage renal disease.

vesicle formation and trafficking. HPS causes abnormalities in the biosynthesis and function of melanosomes, platelets and immune cells, and storage of ceroid-like material in kidneys, heart and lungs (1). CHS is characterized by recurrent infections and death in midlife and HPS is characterized by frequent bleeding diathesis. Our patient was a case of albinism and did not have any signs or symptoms of those two syndromes. Le/le mutant (light-ear) mouse is an animal model of human OCA and there is a striking accumulation of intense green auto-fluorescent ceroid-like material in the lysosomes of the proximal tubular cells that turns to black granules after staining. An increased activity of beta galactosidase enzyme is also observed in proximal tubular cells of le/le mutants (1,2). The lysosome structure of male le/le mice accumulates more beta-galactosidase (1-3).To our knowledge, this is the first report case of a black kidney in a normal person with OCA.

Renal hemosiderosis is a known complication of chronic intravascular hemolysis, such as hemolytic anemia, paroxysmal nocturnal hemoglobinuria (PNH) and mechanical hemolysis of prosthetic cardiac valves. Our patient, however did not have any clinical or laboratory findings of hemosiderosis. We propose that, OCA may be similar to the two related syndromes (CHS, HPS), that could lead to more widespread lysosomal excretory defect, beyond the melanosomes. These defects could lead to accumulation of some intracellular material, leading to the gross discoloration of the kidney. Elevated urinary dolichol excretion in HPS that indicates the lysosomal dysfunction has been reported and the same finding could be present in simple OCA but we did not perform this measurement (3).

Author contributions

MRA was the single author of the paper.

Conflict of interests

The author declared no competing interests.

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References

1. Huizing M, Anikster Y, Gahl WA. Hermansky-Pudlak syndrome and related disorders of organelle formation. Traffic 2000; 1(11):823-35.
2. Meisler MH. Synthesis and secretion of kidney beta-galactosidase in mutant le/le mice. J Biol Chem 1978;253(9):3129-34.
3. Witkop CJ Jr, Wolfe LS, Cal SX, White JG, Townsend D, Keenan KM. Elevated urinary dolichol excretion in the Hermansky-Pudlak syndrome. Indicator of lysosomal dysfunction. Am J Med 1987;82(3):463-70.