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Toxicologic Mimic: Argyria-like Skin Discoloration Associated with Methemoglobinemia

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Background

Alkaptonuria is a rare autosomal recessive disorder of tyrosine and phenylalanine metabolism. It leads to overproduction of homogentisic acid (HGA), a tissue pigment and inducer of methemoglobin. It leads to ochronosis, a blue-gray tissue discoloration that can mimic silver deposition (argyria), usually starting after the fourth decade of life. Hypothesis In the setting of end-stage renal disease, alkaptonuria can cause skin pigmentation mimicking argyria and severe methemoglobinemia.

Methods

This is a retrospective single chart review. Hospital and autopsy records were reviewed.

Results

A 60-year-old woman with a history of worsening slate blue-gray skin discoloration, renal failure on hemodialysis, and pseudobulbar affect, on quinidine/dextromethorphan, presented to a community hospital for evaluation of weakness, where she was found to have a methemoglobin level of 10% and hemoglobin of 6.0 g/dL. After transfer to tertiary center, her repeat methemoglobin level was 24.5% with hemoglobin of 8.1g/dL. She was given 2mg/kg of methylene blue and transfused 2 units packed red blood cells. Subsequently, she became hypotensive. She underwent an 8-unit manual exchange transfusion, in addition to, pressors, continuous renal replacement and broad-spectrum antibiotics. Care was withdrawn after the patient required intubation for acute respiratory distress syndrome; she died on hospital day two. Post-mortem analysis showed findings consistent with alkaptonuria. Pre-mortem serum silver testing was negative. Urine gas chromatography/mass spectrometry did not identify other methemoglobin-inducing substances. Confirmatory genetic testing was not available.

Discussion

Alkaptonuria occurs due to an inborn error of tyrosine and phenylalanine metabolism due to dysfunction of homogentisate 1,2-dioxygenase. It leads to deposition of HGA products in the skin, causing blue-gray discoloration that presents later in life. HGA is a known inducer of methemoglobin production. It is unclear what role quinidine played in the development of methemoglobinemia or ochronosis.

Conclusion

Development of methemoglobinemia in the setting of blue-gray skin discoloration should prompt consideration of alkaptonuria as an etiology.