

Assessment of Hematological, Biochemical, and Blood Mineral Parameters in Alkaptonuria Patients in Southern Jordan: A Case-control Study

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ABSTRACT

Background and Aim: Alkaptonuria is rare autosomal recessive disease. It results from a homogentisate 1,2-dioxygenase gene mutation. A major challenge for AKU researchers is the lack of a consistent approach for monitoring illness severity or therapy success. This study investigated blood minerals, hematological and biochemical characteristics, human TNF-alpha, and human Interleukin-10 in the Al-Karak region of southern Jordan.

Method: The study comprised ten people with AKU, ten carriers, and ten controls. Each participant's fasted blood was drawn twice (with and without EDTA) in the morning so that mineral levels, hematological and biochemical parameters, TNF-alpha, and human IL-10 could be analyzed.

Results: AKU patients and carriers had no abnormalities in hematological parameters (HCT, RBC, MCV, WBC, PLT, RDW-CV, and MPV) compared to controls, but HB, MCH, and MCHC were at the lower border of the normal range with no significant change, indicating mild anemia. In addition, female AKU patients and carriers had far higher lymphocyte counts than controls. Biochemical analysis (creatinine, urea, glucose, albumin, total protein, ALT, AST, Alkaline phosphatase, Bilirubin total, and uric acid) and elemental analysis (K, Na, Fe, Mg, Ca, PO₄, Zn, Cl, Cu, Ionized Ca⁺⁺, and Se) were normal regardless of gender. TNF-alpha levels were significantly higher in AKU patients and carriers than controls, but IL-10 levels were significantly lower, regardless of participant group or gender.

Conclusion: The findings of this study could serve as a springboard for further illness research, particularly in rare conditions like AKU.

Key words: Alkaptonuria, AKU, homogentisate, TNF-alpha, elements

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