

## **Evaluation of clinical, laboratory and radiographic characteristics of Patients With Alkaptonuria ochronosis in Ardabil Province**

### **Abstract**

**Background:** Alkaptonuria is a rare recessive autosomal metabolic disease that occurs due to a deficiency of the homogentetic acid oxidase hepatic enzyme. Symptoms usually appear as discoloration of the tissues and degenerative joint disease usually after 1 to 2 years of age.

**Aim:** evaluate the clinical manifestations and radiographic and laboratory findings of patients with Alkaptonuric Akronosis in Ardabil province.

**Patient and methods:** In this cross-sectional, descriptive-analytical study, all patients with Alkaptonuronic Acronosis in Ardabil province who referred to rheumatology clinic after obtaining informed consent were enrolled. A questionnaire including demographic information, height, weight, history of smoking and alcohol, educational level, occupation, clinical presentation, laboratory and radiographic findings were completed for each patient. The data were recorded in the relevant checklists and the results were statistically evaluated.

**Results:** In this study, 28 patients were eligible for inclusion, 7 of whom were in alkaline phase and the rest had clinical symptoms. The mean age of patients was  $49.82 \pm 17.2$  years and mean age of onset was  $42.13 \pm 11.51$  years. The mean age of patients with clinical symptoms was  $57.10 \pm 10.21$  years and mean age of onset of clinical symptoms was  $42.05 \pm 11.43$  years. The minimum age of onset of clinical symptoms was 20 and maximum 68 years.

The average duration of clinical symptoms to admission was  $9/45 \pm 6/60$  years. Of the 27 patients studied, 12 were male and 16 were female. The most common symptoms in symptomatic patients were back pain and skin symptoms, cartilage involvement (pigmentation and discoloration of the ear), ocular symptoms, pelvic, nail and shoulder involvement respectively. In terms of radiographic findings in patients with clinical symptoms, the most common findings were

involvement, which was observed in 90% of patients.

**Conclusion:** Investigation of clinical manifestations and radiographic and laboratory findings of patients with Alkaptonuric Akronosis in Ardabil showed that back pain is the most common complaint in these patients and it is necessary to obtain more detailed history and examination in case of low back pain.

**Keywords:** Alkaptonuria, Akronosis, spondyloarthropathy