Shafa Ortho J. 2017 May; 4(2):e11718. doi: 10.5812/soj.11718.

Published online 2017 May 14. **Commentaries**

Undiagnosed Ochronosis Presented as Severe Osteoarthritis with Concomitant Osteoporotic Fracture

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Received 2017 April 12; **Accepted** 2017 April 20.

Dear editor,

With respect to the commentary article written by Dr. Alireza Mirzasadeghi (DOI: 10.17795/soj-7533) (1), concerning the adequacy of evidence presented to support the diagnosis of ochronosis/alkaptonuria in the article of Dr. Zabihiyeganeh et al. entitled "undiagnosed ochronosis presented as severe osteoarthritis with concomitant osteoporotic fracture", (DOI: 10.17795/soj-4717) (2), we have provided our complementary answers in this report.

Regarding shortage of enough information required to establish the diagnosis of ochronosis, it should be noted that although the ochronosis is a rare disorder, the number of case reports on the diagnosis of the disease is considerable. Considering the word limit of case reports and regarding the main focus of our report, which was drawing attention towards the osteoporotic fracture, and low bone quality in spite of high bone density in such patients, we chose to place more emphasis on the fracture part and ignore some details in the diagnostic part, which in turn could be found in other reports of ochronosis.

Lack of laboratory or histopathological investigations pointed by Dr. Mirzasadeghi has been regarded in the limitation section of our study. Nevertheless, since such tests are not routinely performed in our country and considering the typical clinical and radiological presentation of the disease, these tests were ignored to perform.

Hyperpigmentation was clearly present in the patient's ear cartilage and sclera. However, since the patient was a Muslim lady, she did not agree to publish the photograph of these lesions. In addition, no history of hydroquinone and cosmetic intervention was reported by the patient. Moreover, deposition of dark pigments in cartilage and connective tissue was noticed by the due surgeons, which could be added to the original article. Nevertheless, no synovial effusion was present to allow synovial fluid aspiration.

Considering prolonged exposure to arsenic or silver, no such exposure was also reported by the patients during her clinical evaluation process.

It should be added that although degenerative joint diseases are prevalent among elderly women, presentation of dense disc calcification (a wafer like calcification) with minimal osteophytic changes is not regarded as manifestation of primary osteoarthritis, and the presence of mentioned clinical findings would be strongly in favor of ochronosis (3).

Finally, we thank Dr. Mirzasadeghi for expressing his concerns. We believe that sharing such concerns will improve our clinical knowledge and lead to better management of similar cases.

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