

Akhenaten, a unique pharaoh

To the Editor: Retief and Cilliers¹ suggest an interesting new theory about Akhenaten's physical appearance, but omitted to refer to previously suggested differential diagnoses: for example, schistosomiasis,² myotonic dystrophy,³ elephantiasis⁴ and Antley-Bixler syndrome have also been suggested.⁵ More recently, we proposed homocystinuria as a possible cause.⁶ Since Kallman's syndrome is a hereditary disease, it is important to see how this diagnosis fits into Akhenaten's family tree. The fact that Akhenaten's wife, Nefertiti, and their children were similarly depicted implies that they suffered from the same disease. Akhenaten's parents, Amenhotep III and Tiye, were most probably healthy. The genetics of Kallman syndrome are still not fully understood.⁷ However, two of the best-described forms of Kallman syndrome are inherited autosomal dominant and X-linked disorder.⁷ An obvious problem with this diagnosis is the healthy father, although it cannot be fully rejected because the phenotype of these patients varies from partial to complete. However, as we explained in our article, an autosomal recessive disease, such as homocystinuria, is the most probable explanation. Although, admittedly, all proposed diagnoses have their advantages and drawbacks, it is important to note that all of them are made under one assumption: that art from the Amarna period was realistic, i.e. that this was not just a form of artistic expression creating a distance from traditional Egyptian art. The authors do not cite an investigation by Hawass *et al.*, in which it was suggested that a mummy found in KV55 was actually Akhenaten,^{8,9} they also suggest that the depictions of Akhenaten exaggerate his actual appearance. However, the definitive confirmation of both the identity of the KV55 mummy and a possible diagnosis can be made only by DNA analysis. Unfortunately, the method for DNA sampling is the subject of heavy criticism, and the results do not appear to be totally trustworthy.¹⁰

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