

## Ancestry and Pathology in King Tutankhamun's Family

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The New Kingdom in ancient Egypt, comprising the 18<sup>th</sup>, 19<sup>th</sup>, and 20<sup>th</sup> dynasties, spanned the mid-16<sup>th</sup> to the early 11<sup>th</sup> centuries BC. The late 18<sup>th</sup> dynasty, which included the reigns of pharaohs Akhenaten and Tutankhamun, was an extraordinary time. The identification of a number of royal mummies from this era, the exact relationships between some members of the royal family, and possible illnesses and causes of death have been matters of debate.

The objectives of this study were: To introduce a new approach to molecular and medical Egyptology, to determine familial relationships among 11 royal mummies of the New Kingdom, and to search for pathological features attributable to possible murder, consanguinity, inherited disorders, and infectious diseases.

From September 2007 to October 2009, royal mummies underwent detailed anthropological, radiological, and genetic studies as part of the King Tutankhamun Family Project. Mummies distinct from Tutankhamun's immediate lineage served as the genetic and morphological reference. To authenticate DNA results, analytical steps were repeated and independently replicated in a second ancient DNA laboratory staffed by a separate group of personnel. Eleven royal mummies dating from circa 1410-1324 BC and suspected of being kindred of Tutankhamun and 5 royal mummies dating to an earlier period, circa 1550-1479 BC, were examined.

Main outcome measures comprised microsatellite-based haplotypes in the mummies, generational segregation of alleles within possible pedigree variants, and correlation of identified diseases with individual age, archaeological evidence, and the written historical record.

Genetic fingerprinting allowed the construction of a 5-generation pedigree of Tutankhamun's immediate lineage. The KV55 mummy and KV35YL were identified as the parents of Tutankhamun. No signs of gynecomastia and craniosynostoses (eg, Antley-Bixler syndrome) or Marfan syndrome were found, but an accumulation of malformations in Tutankhamun's family was evident. Several pathologies including Köhler disease II were diagnosed in Tutankhamun; none alone would have caused death. Genetic testing for *STEVOR*, *AMA1*, or *MSP1* genes specific for *Plasmodium falciparum* revealed indications of malaria tropica in 4 mummies, including Tutankhamun's. These results suggest avascular bone necrosis in conjunction with the malarial infection as the most likely cause of death in Tutankhamun. Walking impairment and malarial disease sustained by Tutankhamun is supported by the discovery of canes and an afterlife pharmacy in his tomb.

Using a multidisciplinary scientific approach, we showed the feasibility of gathering data on Pharaonic kinship and diseases and speculated about individual causes of death.

**Key words:** Egyptian mummies – paleogenetics – malaria

