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INVITED LECTURE – IL3

FROM MUMMIES TO PATHOGENS: PALEOGENETICS AND ANCIENT HUMAN REMAINS

Albert Zink[1,2,3]

[1]SNSB, State Collection for Anthropology, Munich, Germany; [2]Department of Biology II, Ludwig Maximilian University (LMU) Munich, Germany; [3]Institute for Mummy Studies, Eurac Research, Bolzano, Italy

The application of next-generation sequencing (NGS) to ancient human remains has revolutionized the study of past populations by enabling the recovery of ancient human and microbial DNA. Paleogenetic analyses of mummies and archaeological skeletons provide unique insights into human history while transforming our understanding of the origins, evolution, and spread of infectious diseases. By identifying ancient pathogens and reconstructing their genomes, ancient DNA has become an indispensable tool for investigating the health, evolution, and disease burden of past populations.

One of the earliest and best-known applications of palaeogenetics is the investigation of the Tyrolean Iceman, Otzi. Using metagenomic diagnostics and targeted genome capture, we identified the gastric pathogen Helicobacter pylori and reconstructed its complete genome. Comparative genomic analyses revealed a virulent strain belonging to a predominantly Asian ancestral lineage, providing new insights into the evolutionary history and dispersal of H. pylori in Europe. Naturally mummified human remains from Vac, Hungary, have proven to be an exceptional source for the molecular detection of ancient pathogens. The reconstruction of complete Mycobacterium tuberculosis genomes from these mummies has greatly advanced our understanding of the long-term evolution, diversity, and adaptation of the tuberculosis pathogen while demonstrating the enormous potential of ancient DNA for investigating historical infectious diseases. Our investigations of mummies from Pharaonic Egypt combine morphological, radiological, and molecular methods to reconstruct health and disease in ancient populations. High-resolution computed tomography together with ancient DNA analyses has enabled the identification of infectious diseases, vascular pathologies, and genetic predispositions. A notable example is the detection of ancient Plasmodium falciparum DNA in Tutankhamun, providing direct molecular evidence for malaria tropica in ancient Egypt.

Together, these examples demonstrate how palaeogenetics has transformed the study of ancient human remains by enabling the direct detection of pathogens and providing unique insights into the long-term co-evolution of humans and infectious diseases.

Keywords: Ancient DNA; Paleogenetics; Mummies; Ancient pathogens; Paleopathology.