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Abstract (oral session)

Molecular detection of Tropheryma whipplei in clinical specimens: eight years of experience from the German Reference Laboratory for Tropheryma whipplei

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Objectives: Whipple´s disease is a chronic infectious disease caused by the bacterium Tropheryma whipplei. If diagnosed and treated timely, the disease can usually be cured with long-term antibiotic therapy. Yet if left untreated it frequently follows an invariable fatal course with multiorgan involvement. Typical clinical manifestations of Whipple's disease include mainly diarrhea, weight loss, abdominal pain and polyarthralgias; but atypical symptoms such as neurological involvement may develop. Classical diagnosis relies on histopathology (PAS staining). However, since the advent of molecular methods and techniques for the specific detection of this pathogen, T. whipplei has been detected in a number of other extraintestinal sites, as the cause of a myriad of clinical manifestations. Herein, we present an eight-year experience as a German Reference Laboratory for the specific detection of T. whipplei using diverse molecular platforms. Methods: For specific detection of T. whipplei we used 16S rRNA-gene PCR with sequencing and real- time PCR of the rpoB gene. We overall investigated 1273 specimens from 977 patients. Additionally, in order to obtain information about number and spatial distribution of the microorganisms we developed a tissue-based fluorescence in situ hybridization (FISH) approach for sections of selected samples. Results: During the eight-year period T. whipplei DNA was detected by PCR in 76 out of 977 cases (7.8%). Later and since the incorporation of real-time PCR for the diagnosis of T. whipplei, we found the organism in a variety of other clinical samples including cerebrospinal fluid, synovial fluid, bone, skin, lymph nodes, and bronchoalveolar lavage. Within heart valves and lymph nodes, infection was additionally confirmed by FISH. Furthermore we diagnosed cases without the typical intestinal involvement of Whipple's disease, enabling accurate identification of the agent in the context of diverse non-classic clinical scenarios that would have otherwise remained undiagnosed. Conclusions: This study highlights the use of molecular-based detection methods to detect T. whipplei infection in clinical specimens, increasing our ability to identify this agent. In this case series, a molecular approach served to unravel the broad spectrum of clinical manifestations of Whipple's disease, while at the same time highlighted the valuable role of FISH confirming the infection in extra-intestinal cases.