Genetics and clinical diagnostics of mycoses

Several studies show that the risk for immunocompromised patients to suffer from an invasive mycosis can also be determined through genetic factors. Fungal Septomics coordinates the international multi-centre-study AspIRS (Aspergillosis intrinsic risk stratification) – the first systemic and genome-wide approach to help identifying genetic polymorphisms that are associated with invasive aspergillosis (IA). In addition to the genome-wide analyses, a risk marker for IA has been identified in petraxin 3 locus in cooperation with A. Carvalho und L. Romani (Perugia). Pentraxin 3 is of central importance with regard to the immune response against *A. fumigatus*. The risk haplotype may lead to a substantially decreased expression of pentraxin 3 in the neutrophile granulocytes and hence increases the risk for patients benefiting from a stem cell transplantation to contract an IA. (Cunha et al., 2014). These projects aim at identifying potential bio-marker molecules in the medium term, which will enable better molecular diagnostics of invasive mycoses. We are also working on improvements of diagnostic possibilities for invasive mycoses in other areas. In a prospective study, we have evaluated a new protocol for the DNA-enrichment for IA diagnostics that we have evaluated by means of PCR. (Springer et al., 2011). This work is carried out within the frame of the National Reference Centre for Invasive Mycoses (NRZ-Myk), which is available to physicians from all over Germany.

National Reference Center for Invasive Fungal Infections