

TRANSCRIPTIONAL BLOOD SIGNATURES OF SARCOIDOSIS, SARCOID-LIKE REACTIONS AND TUBERCULOSIS AND THEIR DIAGNOSTIC IMPLICATIONS

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There is an increasing evidence of genetic overlap between sarcoidosis (SA) and tuberculosis (TBC) (1), two conditions that differ often, but not always, on clinical and histopathological features in their different forms and type of organ involvement (2). These disorders reveal sometimes common etiologies and similar genetic and pathogenetic profiles (2). In particular, SA and TBC share similar, over-abundant blood transcriptional profiles induced by interferon, unlike conditions such as pneumonia and lung cancer where inflammatory transcriptional activity dominates (1). On this basis, the analysis and comparison of blood transcriptional signature of separate disorders could help to distinguish one from the other (1,4). Bloom et al. have found a high degree of similarity between blood's genetic signatures of SA and TBC patients (1). However, it should be considered that the diagnosis of sarcoidosis is sometimes not achieved with consistent and rigorous criteria (5), leading often to cases of sarcoid-like reactions (SLRs) classified erroneously as SA. This claim is sufficiently alarming, and some efforts have been made to establish new criteria giving a clear differentiation between SA and SLRs (5).

The additional unraveling of the pathogenesis of diseases such as early-onset sarcoidosis (EOS)

and Blau syndrome has allowed their classification into the group of monogenic autoinflammatory syndromes caused by mutations in the CARD15 / NOD2 gene. These conditions are substantially different from the “classical” adult form of SA (6).

Comparison between genetic signatures of recently identified EOS and Blau syndrome with those of patients with TBC and late onset SA could be a good start for future investigations. If substantial differences in blood genetic signatures are found, they could lead to reconsider the pathogenesis of SA as a whole and could give additional clues in the diagnosis of SA and differentiation with other similar conditions such as TBC and SLRs.

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Received: 9 December 2015

Accepted after revision: 11 January 2016

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