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Commentary

Precision Medicine: The new era in medicine

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Precision medicine, also known as stratified medicine or personalized medicine, aims to better target intervention to the individual, maximize benefit and minimize harm. Enthusiasm for the prospect of precision medicine has grown significantly in the last few years. There has been increasing interest in two streams of development in precision medicine. First is the integration of electronic medical records capturing longitudinal data and providing clinical phenotypes into decision making tools for precision medicine. Secondly, the advance in genomic medicine and pharmacogenomics research provides an expanding arsenal of genetic predictors of disease and health outcomes. Together these two streams provide a unique framework for an individualized diagnostic and therapeutic approach. Genetics has been a part of the health care system since introduction of the newborn screening in the 1960s, now it has become a priority area for prevention and treatment of common chronic diseases such as cancer and heart diseases (Auffray et al., 2016). In cancer research, personalized medicine based on genomics and pharmacogenomics is expanding rapidly. Other chronic diseases, such as diabetes and neurodegenerative diseases have widened their horizons and included precision medicine in their research strategies. Inflammation appears in a wide spectrum of diseases and has a very heterogeneous phenotype, including autoimmune diseases such as systemic lupus erythematosus and rheumatoid arthritis to autoinflammatory diseases and immunodysregulatory diseases such as secondary hemophagocytic lymphohistiocytosis. In the last 25 years, the understanding of inflammatory diseases has dramatically increased and changed our approach to treatment. Initially these under-recognized childhood and adult diseases were associated with high burden of disease and associated high mortality rates. In rheumatoid arthritis, the single discovery of the cytokine TNF-alpha has changed a joint-destructing debilitating disease refractory to treatment into a well-controlled disease (Cuppen et al., 2015). Inflammatory brain diseases are a relatively new area of inflammation, but have a vastly expanding spectrum of diseases.

Inflammatory brain diseases, including infectious meningitis, encephalitis, vasculitis, T-cell mediated inflammatory brain diseases, and demyelinating diseases are rare diseases and would benefit from a precision medicine approach. The spectrum of inflammatory brain diseases has rapidly expanded during the last decade and keeps expanding with new discoveries (Twilt and Benseler, 2014). Diseases such as small vessel primary CNS vasculitis, previously only diagnosed on autopsy, are now increasingly recognized early in the disease course, before irreversible neurological deficits are developed. One of the big challenges in inflammatory brain diseases is the overlapping clinical phenotype and absence of genetic markers and disease specific biomarkers for most diseases. Verhey et al. (2013), developed a scoring system to evaluate brain MRIs of children with demyelinating diseases. The scoring tool was tested on MRIs of patients with demyelinating diseases (acute disseminated encephalomyelitis (ADEM) and multiple sclerosis (MS)) and small vessel CNS vasculitis because these disorders share clinical features rendering accurate diagnosis challenging at onset. Although the inter-rater agreement of many features was acceptable, there were items that demonstrated acceptable inter-rater agreement but did not aid in differentiating ADEM, MS and small vessel CNS vasculitis on MRI. These authors made a significant step towards precision medicine, but other factors must to be added to the equation before barriers in diagnosing these diseases are lifted.

Due to the heterogeneity of inflammatory brain diseases, the development and implementation of one simple automated classification tool seem extremely difficult. Even so, the study by Obermeier et al. (2016), uses a tool to enable physicians to ask the correct questions at the right time, thereby classifying cases consistently and accurately, and facilitating translational research. The implemented Vienna Vaccine Safety Initiative Automated Case Classification-tool (VACC-Tool) was used to identify early common infectious and rarer inflammatory brain diseases (demyelinating diseases). Although the tool left some cases indeterminate (when doctors in routine care failed to obtain the necessary laboratory tests), this is the first step in classification of inflammatory brain diseases using a precision medicine approach, and could potentially be expanded to the whole spectrum of brain diseases. The next step would be to include biomarkers, proteomics and genomics to further develop precision medicine strategies leading to personalized medicine for patients with inflammatory brain diseases. Currently most treatment strategies include potent immunotherapy with severe side-effects. Inflammatory brain diseases are not the only neurological area precision medicine holds a future. Several studies

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have looked at the role of personalized medicine in mood disorders, autism and related neurodegenerative disorders (Alhajji and Nemeroff, 2015; Sahin and Sur, 2015). All studies have one thing in common, they all combine clinical phenotypes with genomics and proteomics, feeding back to the two big streams in precision medicine.

In the next coming years many genomic discovery platforms in combination with clinical phenotyping based on electronic medical record data will lead to more insight with respect to disease processes and lead to more individualized targeted treatment (Smoller et al., 2016). Overall the use of clinical phenotype and classification tools help to diagnose diseases early to allow a more rapid introduction of targeted treatment avoiding harm and increasing benefit.

Conflict of interest

I declare I have no conflict of interest.

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