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Advance on the diagnosis and typing of amyloidosis

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Abstract: Amyloidosis is a protein misfolding disorder, in which normally soluble proteins undergo conformational changes and are aggregated abnormally as insoluble fibrils deposited in the extracellular space, and progressively caused the structural and functional damage of multiple organs. So far, at least 36 different precursor proteins can cause amyloidosis. The most common type of systemic amyloidosis is immunoglobulin light chain amyloidosis (AL) , which accounted for 80% to 90% of amyloidosis in developed countries, the followed types are amyloid A amyloidosis (AA) and leukocyte chemotactic factor 2 (Lect2) amyloidosis; while hereditary amyloidosis including transthyretin, fibrinogen Aα chain, apolipoprotein A-I /A-II , lysozyme , gelsolin, and cystatin C were very rare. The incidence of AL amyloidosis affected 12 individuals per million per year in the USA. Patients with systemic AL amyloidosis in late stage with cardiac involvement have a survival of 3-6 months. Hence early diagnosis and accurate typing of amyloidosis are necessary for its therapy and prognosis.

The clinical manifestation of amyloidosis is heterogeneous depending on the severity of amyloid deposition and the organs involved. The diagnosis of amyloidosis was established by tissue biopsy and pathological analysis, in which the presence of apple-green birefringence under a polarized light after Congo red staining and the fibrils in a diameter of 8-12nm identified by electron microscopy of biopsy specimens are the main diagnostic evidence. The less invasive investigations including subcutaneous fat, bone marrow, tongue, rectum or lip biopsy can lead to diagnosis in 50–85% of systemic AL amyloidosis; the direct biopsy of an affected organ including kidney, heart or liver will get high sensitivity of amyloid deposit detection. The identification of precursor protein of amyloid is generally performed by immunofluorescence(IF) or immunohistochemistry(IHC) using antibodies to the common types of precursor proteins. However, due to the false negative or positive staining of IF or IHC in the typing of amyloidosis, there were potential diagnostic pitfalls caused misleading results. A detailed observation of electron microscopy combining with immuno-electron microscopy using colloid gold labeling can get high sensitivity and specificity in the diagnosis and typing of early stage of amyloidosis. The recently developed proteomic analysis based on mass spectrometry is proved to be a powerful tool in typing of amyloidosis, especially for identification of rare types of hereditary amyloidosis. Although

there is no specific biomarker of blood or urine for amyloidosis, a screening for NT-proBNP level and albuminuria in patients with a monoclonal gammopathy and/or an abnormal free light chain ratio is essential for detection of pre-symptomatic systemic AL amyloidosis, and can get good prognosis with early intervention to curb monoclonal Ig production effectively.