Specifieke aanbevelingen over naamgeving van de verschillende typen amyloïdose en nomenclatuur

- Amyloid fibrils are not uniform and several “classes” can be recognized. When the word “amyloid” is used a more precise definition is therefore necessary[1].
- AL, ATTR, etc. are amyloid protein names. Corresponding diseases are AL amyloidosis, ATTR amyloidosis and so on[1].
- Diseases depending on amyloid protein gene mutations are “hereditary” and should not be called “familial”. The designations “familial amyloid polyneuropathy, FAP”, or “familial amyloid cardiomyopathy, FAC” should be not used[1].
- A variant amyloid fibril protein is best defined by exact description of the mutation. The variant should be defined by one-letter-code and be numbered from the mature protein, e.g. ATTRV122I or ATTRV30M. Instead of the exact mutation the designation ATTRv (variant) can be used and is preferred to ATTRm (mutant)[1].
- Livers from patients with hereditary ATTR amyloidosis have been used for transplantation to non-ATTR patients with liver failure and subsequently ATTR amyloidosis has been observed in some recipients [18]. The term “Iatrogenic ATTR” has been used to describe these clinical situations. The Nomenclature Committee considers that authors may use the term Iatrogenic ATTR amyloidosis to describe ATTR amyloidosis as they wish and consider appropriate[2].
- In 1999, the ISA Nomenclature Committee recommended that the mouse Saa2 locus be renamed to mouse Saa1, based on the correspondence of its chromosomal mapping to that of human SAA1 [19]. The National Center for Biotechnology Information (NCBI) database retains the original Saa2 designation for the locus now designated as mouse Saa1. Investigators will need to keep this in mind when using the NCBI databases. It is recommended that the 1999 ISA nomenclature for mouse genes and gene products continue to be used consistently[2].
- The terms “hereditary amyloidosis” and “familial amyloidosis” refer to different entities. The term “hereditary amyloidosis” should be used when there is a mutation in the fibril protein gene itself, e.g. ATTR, ALys or AFib. The term “familial amyloidosis” should be used when the syndrome occurs in a familial setting due to mutations in genes expressing non-amyloid proteins, e.g. AA amyloidosis[2].
- Synonyms and terminology used in the past when the chemical diversity of amyloid fibril proteins was unrecognized remain in use today, but are not particularly useful and may be confusing. For example, synonyms for hereditary ATTR amyloidosis include Familial Amyloid Polyneuropathy Type I (Portuguese-Swedish-Japanese Type), Familial Amyloid Polyneuropathy Type II (Indiana/Swiss or Maryland/German Type), Leptomeningeal Amyloidosis, Familial Amyloid Cardiomyopathy, Familial Oculoleptomeningeal Amyloidosis (FOLMA). It is strongly recommended that the use of these terms be discontinued and that the syndrome be designated by the name of the protein. While the term familial amyloid polyneuropathy (FAP) is widely used in the neurology literature and may be included as appropriate, it is recommended that it be accompanied by ATTR[2].
- The designation senile systemic amyloidosis (SSA) was coined when it became clear that this is a systemic disease with a specific amyloid protein and not only a cardiac disease that occurs particularly in old age [20]. It was later shown that the fibril protein in SSA is derived from WT TTR [21] and that the disease can occur at younger age. It is therefore the committee’s recommendation to use “wild-type ATTR (ATTRwt) amyloidosis” instead of SSA. In a transition period both SSA and ATTRwt amyloidosis can be used simultaneously in order to avoid confusion[2].

- The clinical classification of AL amyloidosis as primary amyloidosis or amyloidosis secondary to myeloma and AA amyloidosis as secondary amyloidosis is ambiguous and outdated and it has long been recommended that these designations not to be used[2].
- The committee recommends that amyloid β precursor protein (AβPP) be used for the precursor of Aβ instead of amyloid precursor protein (APP)[2].

Bron:

1. Benson MD, Buxbaum JN, Eisenberg DS, Merlini G, Saraiva MJ, Sekijima Y, Sipe JD, Westermark P. Amyloid nomenclature 2018: recommendations by the International Society of Amyloidosis (ISA) nomenclature committee. Amyloid 2018; 25:215-219 (https://www.tandfonline.com/doi/full/10.1080/13506129.2018.1549825).
2. Sipe JD, Benson MD, Buxbaum JN, Ikeda SI, Merlini G, Saraiva MJ, Westermark P. Amyloid fibril proteins and amyloidosis: chemical identification and clinical classification International Society of Amyloidosis 2016 Nomenclature Guidelines. Amyloid 2016; 23:209-213 (https://www.tandfonline.com/doi/full/10.1080/13506129.2016.1257986)