An ontology for Age-Related Macular Degeneration using ophthalmologists and language models

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Abstract

We aim to support monitoring of the current guidelines and scientific evidence in the management of Age-Related Macular Degeneration (AMD) in order to augment retinal specialists to develop a clinically oriented and consensual protocol for therapeutic approaches for AMD. First, we are engineering an ontology for AMD retinal condition using information from literature, related medical ontologies and domain knowledge from ophthalmologists. Second, we augment the knowledge engineer capabilities to populate and enrich the ontology using structured knowledge extracted from medical literature with the GPT-3 language model. Third, we perform reasoning to signal to the ophthalmologist differences or inconsistencies among different clinical studies, protocols or therapeutic approaches.

Keywords

medical ontologies, age-related macular degeneration, conflict detection, reasoning

First, as an example of axioms from the AMD ontology, consider the classifications of AMD in Table 1, where $LargeDrusen \equiv Drusen \sqcap \exists hasSize.\geq 125\mu m$, $MediumDrusen \equiv Drusen \sqcap \exists hasSize.\geq 63\mu m \sqcap \exists hasSize.\geq 63\mu m$, respectively $SmallDrusen \equiv Drusen \sqcap \exists hasSize.\leq 63\mu m$. One issue is that these axioms do not always correspond to clinical practice. For instance an eye with a drusen measured by an AI algorithm at 124µm (i.e. slightly below the 125µm limit) is classified according to the definition as a *MediumDrusen*, and hence an *EarlyAMD*, but the ophthalmologist still treats the disease as an *IntermediateAMD*. To map the clinical practice we are also considering axioms in Fuzzy Description Logic. The AMD ontology reuses concepts and relations from BioVerbNet (https://github.com/cambridgeltl/bioverbnet) and medical ontologies, e.g.: (i) $AMD \sqsubseteq InheritedRetinalDystrophy \sqsubseteq MendelianDisease \sqsubseteq HumanDisease$; (ii) $InheritedRetinalDystrophy \sqsubseteq InheritedVitreousRetinalDisease \sqsubseteq RetinalDisorder \sqsubseteq EyeDisorder$.

Second, we enrich the AMD ontology with structured data automatically extracted from scientific studies [1] and clinical trials. Recent advances in Natural Language Understanding can complement the studies conducted by humans on reviewing literature and recent scientific evidence (e.g. [2]). Consider querying a learned model like GPT-3 (https://bit.ly/3e3icZQ) in Table 2). The used prompt was: "A table summarizing the associations of morphological features with disease activity". One the one hand we were fascinated on the easiness to obtain such structured data. On the other hand, in line with G. Marcus (https://cacm.acm.org/blogs/

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Table 1
Sample of definitions and classifications scales for AMD

		Epidemiological classification (Wisconsin grading)
$EarlyAMD^W$	$\equiv$	$AMD \sqcap \exists hasBiomarker.(LargeDrusen \sqcup RetinalPseudodrusen \sqcup PigmentaryAbn)$
$LateAMD^W$	$\equiv$	$NeovascularAMD \sqcup GeographicAtropy$
		Basic clinical classification
$NoAgeingChanges^C$	$\equiv$	$\forall hasDrusen.\bot \sqcap \forall hasAbn.\neg PigmentaryAbn$
$NormalAgeingChanges^C$	$\equiv$	$\forall hasDrusen.SmallDrusen \sqcap \forall hasAbn.\neg PigmentaryAb$
		AREDS simplified severity scale points
$Severity_0$	$\equiv$	$\forall hasBiomarker.\neg LargeDrusen \sqcup \forall changes.\neg Pigment$
$Severity_1$	$\equiv$	$\exists hasBiomarker.\neg LargeDrusen \sqcup (=1) changes.Pigment$

Table 2
Extracting structured information on morphological features using language models (i.e. GPT3)

Review	Feature	Association with disease activity
Mowatt et al. (2014)	OCT	unlikely to be cost-effective for diagnosis/monitoring
Schmid-Erfurth et al. (2016)	CRT	inferior prognostic biomarker for guiding retreatment
Schmid-Erfurth et al. (2016)	IRF	negatively associated with VA
Schmid-Erfurth et al. (2016)	SRF	associated with superior visual benefits and a lower rate of progression towards atrophy

blog-cacm/267674-ais-jurassic-park-moment/fulltext), we are aware of the risks that such models to propagate misinformation. Our stance is that it is easier for the human agent to verify the information in Table 2 and to annotate it with provenance data, instead of manually collecting it from literature. The burden is how to feed the GPT-3 with relevant "prompts". In line with C. Baquero (https://bit.ly/3ElW1J7, prompt design was critical for querying of such language models. The job of *Prompt Designer* may become relevant in populating ontologies.

Third, we apply reasoning to signal differences and inconsistencies among the knowledge within the ontology. These differences reflect the current understanding of the AMD disease: quantitative vs. qualitative fluid assessments, intraretinal fluid vs. subretinal fluid (SRF), exudative vs. nonexudtive fluid. For instance, for SRF both negative and positive but also no-association have been reported [2]. Moreover, heterogeneity of therapeutic approaches has been increased in the context of personalised care. This heterogeneity rises the question of inconsistent information, detected in our approach by the Racer reasoning tool.

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