

An ontology for Age-Related Macular Degeneration - Poster

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An Ontology for Age-Related Macular Degeneration

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Interleaving ML with reasoning for diagnosis

- Reasoning with Description Logics and rules to diagnose retinal conditions
- Using ML for identifying biomarkers
- Focusing on formalising OCT biomarkers for Age Related Macular Degeneration
- Can large language models be used to support ontology engineering?

Towards an ontology for AMD

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
$NoAgeingChanges^C$	$\equiv \forall hasDrusen.\perp \sqcap \forall hasAbn.\neg PigmentaryAbn$
$NormalAgeingChanges^C$	$\equiv \forall hasDrusen.SmallDrusen \sqcap \forall hasAbn.\neg PigmentaryAbn$
$EarlyAMD^C$	$\equiv AMD \sqcap \exists hasBiomarker.MediumDrusen \sqcap \forall hasAbnormalities.\neg PigmentaryAbnormalities$
$IntermediateAMD^C$	$\equiv AMD \sqcap (\exists hasBiomarker.LargeDrusen \sqcap \exists hasAbnormalities.\neg PigmentaryAbnormalities)$
$LateAMD^C$	$\equiv NeovascularAMD \sqcup GeographicAtropy$
	AREDS simplified severity scale points
$Severity_0$	$\equiv \forall hasBiomarker.\neg LargeDrusen \sqcup \forall changes.\neg Pigment$
$Severity_1$	$\equiv \exists hasBiomarker.\neg LargeDrusen \sqcup (= 1)changes.Pigment$
$Severity_4$	$\equiv (> 1)hasBiomarker.LargeDrusen \sqcap (> 1)changes.Pigment$

Using language models for ontology?

Extracting structured information on morphological features

Prompt: "A table summarizing the associations of morphological features with disease activity"

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

Manually checking provenance:

"As shown in Table 1, intraretinal fluid has a negative and subretinal fluid a positive impact on visual outcome"

"patients with subretinal fluid derive larger visual acuity benefits from antiangiogenic treatment"

Identifying OCT biomarkers for AMD

Prompt: "OCT biomarkers for neovascular AMD diagnosis.", intro = "A ranked list of OCT biomarkers for neovascular AMD"

- Foveal Thickness
- Foveal Avascular Zone Area
- Subretinal Hyperreflective Material
- Subretinal Fluid
- Retinal Pigment Epithelium Detachment
- Choroidal Thickness
- Macular Volume
- Macular Edema
- Macular Ischemia
- Macular Atrophy
- Subretinal Fibrosis
- Subretinal Neovascularization
- Subretinal Hemorrhage
- Retinal Pigment Epithelium Atrophy
- Retinal Nerve Fiber Layer Thickness
- Retinal Vessel Density
- Retinal Vessel Tortuosity
- RPE Hyperreflectivity
- Photoreceptor Integrity
- Outer Retinal Thickness

Generating axioms in Description logic

Prompt: "A definition in description logic of Early AMD"

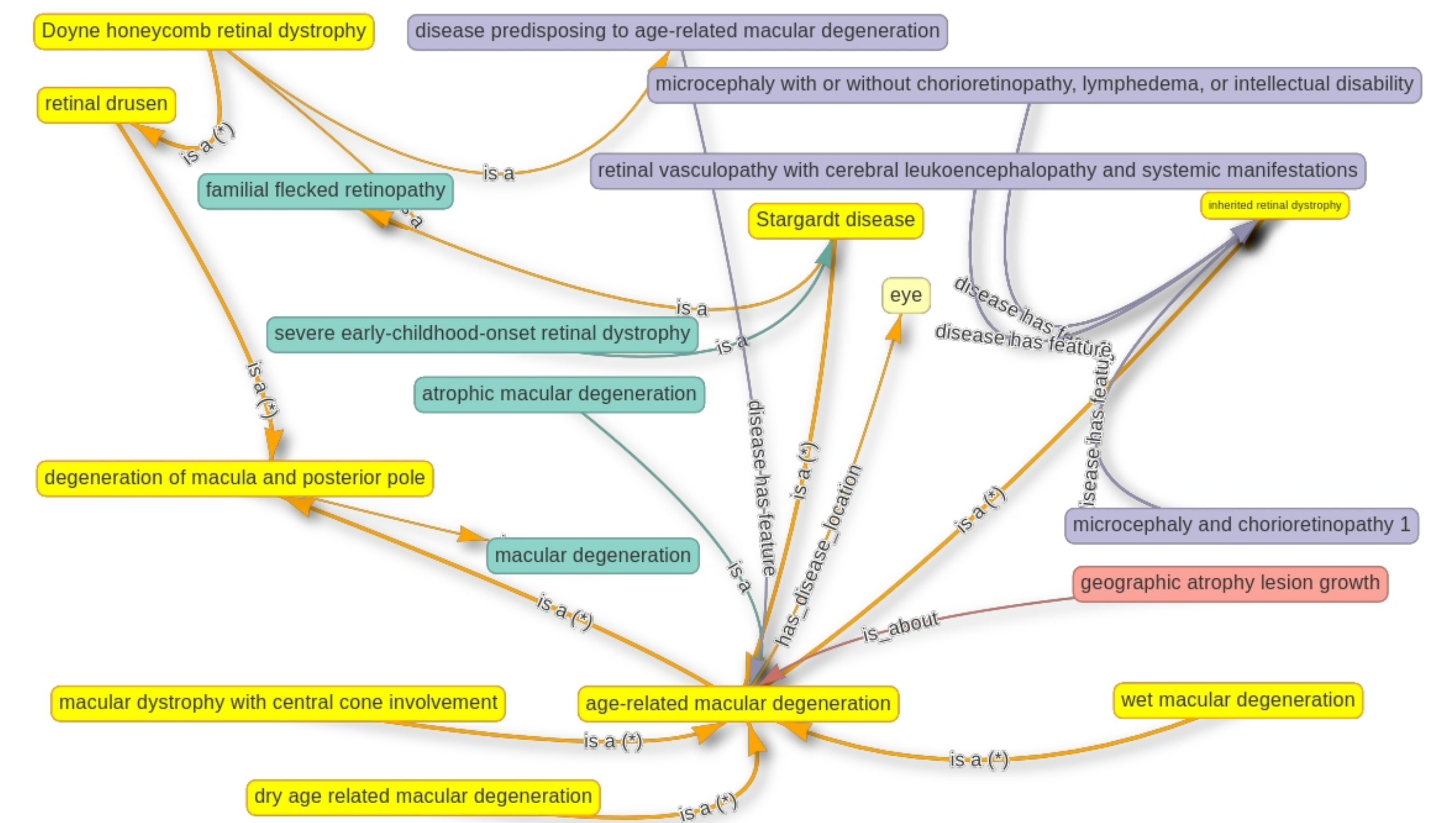
$EarlyAMD \sqsubseteq \exists hasDrusen.Drusen \sqcap \exists hasPigmentaryChanges.PigmentaryChanges$

Prompt: "A definition in description logic of Intermediate AMD"

$IntermediateAMD \sqsubseteq AMD \sqcap \exists hasStageValue.IntermediateStage \sqcap \exists hasSymptom.BlurredVision \sqcap \exists hasSymptom.DistortedVision \sqcap \exists hasSymptom.ReducedCentralVision \sqcap \exists hasSymptom.ReducedColorVision \sqcap \exists hasSymptom.DarkSpotsInVision$

Reusing biomedical ontologies

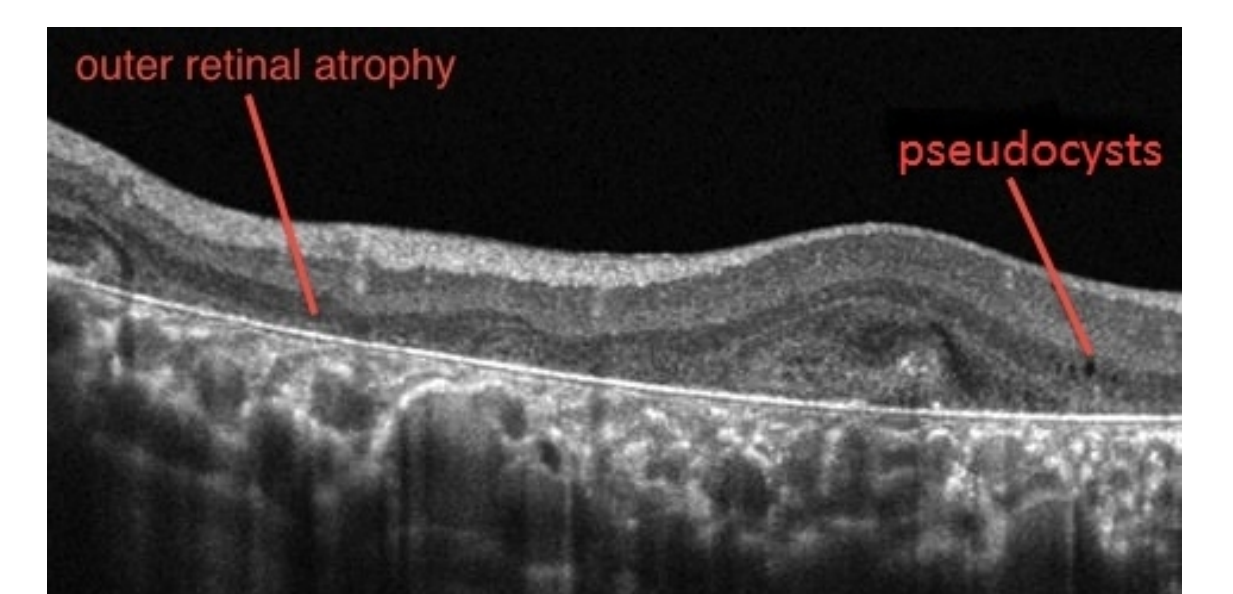
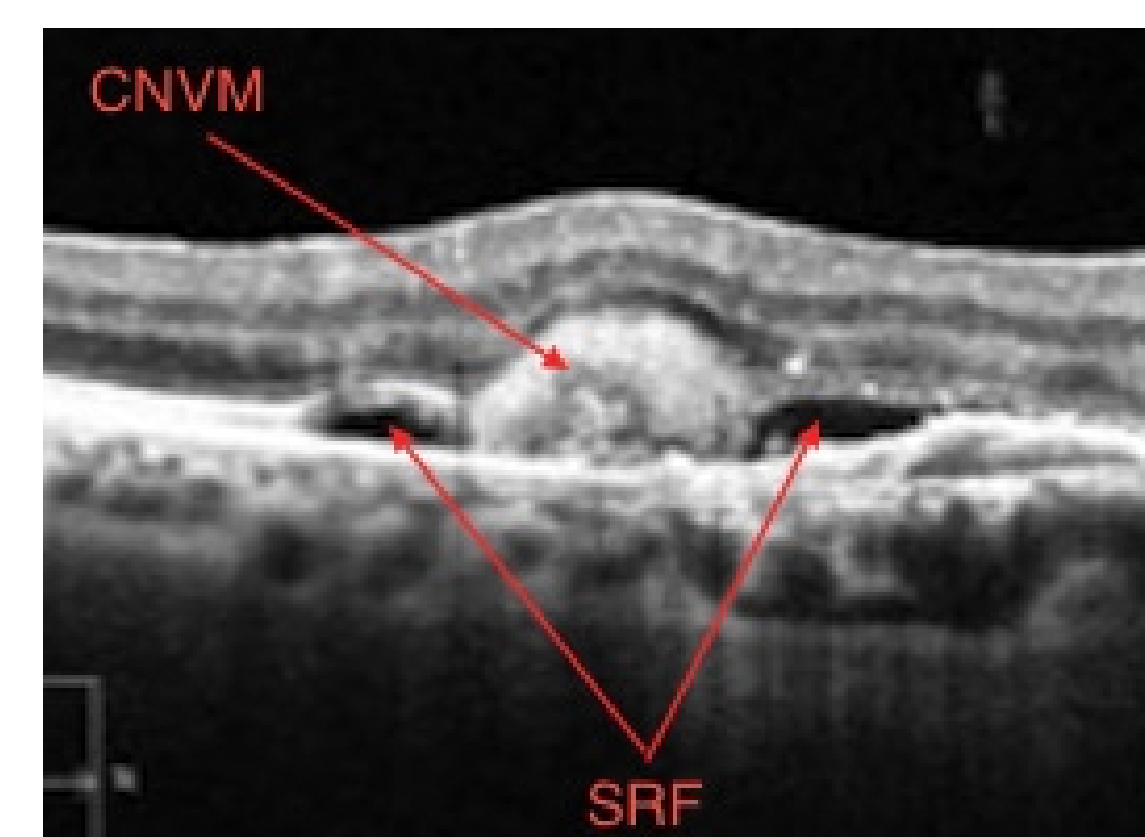
Anatomy ontology, Human Disease, Experimental Factor Ontology, SNOMED, Biological Spatial Ontology, Relation ontology, Symptom ontology



Formalising OCT biomarkers for AMD

$f_3 : hasDisease.WetAMD \sqcap \exists hasBM.(Type2CNVM \sqcap isAbove.RPE \sqcap \exists hasAdjacentBM.SRF \sqcap \forall hasAdjacentBM.SRF$ (1)

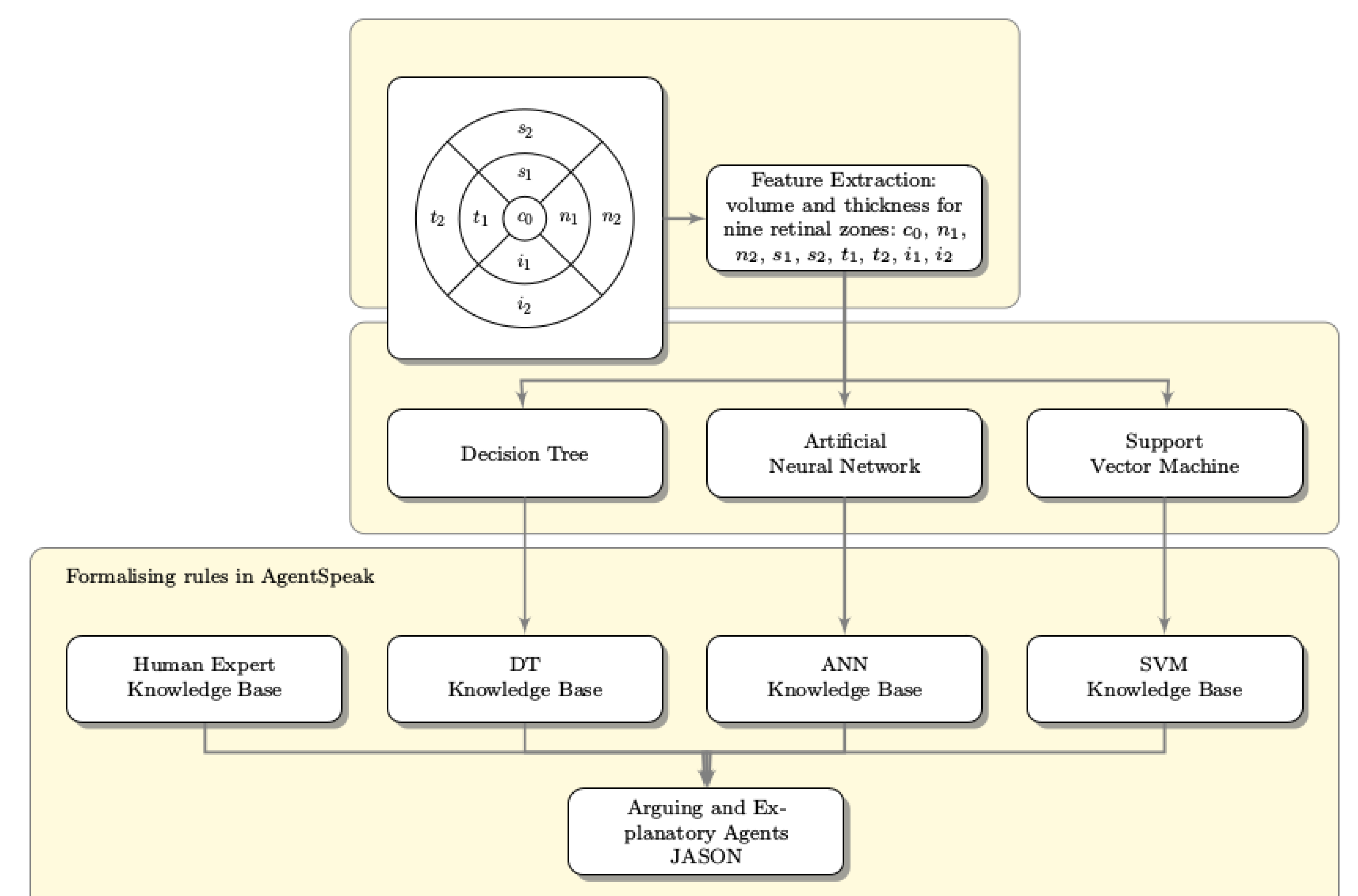
$f_3 : \exists hasBM.(Exudate \sqcap isLocated.Nasal) Type1CNVM \sqsubseteq CNVM \sqcap isBeneath.RPE \sqcap \exists appear.(Fibrovascular \sqcup HemorrhagicPigmentEpithelialDetachment) Type2CNVM \sqsubseteq CNVM \sqcap isAbove.RPE \sqcap \exists hasAdjacentBM.SRF$ (2)



$Pseudocysts \sqsubseteq \exists hasShape.Circular \sqcap \exists hasReflection.hyporeflexive$

Reasoning with rules for AMD diagnosis

Interleaving expert rules with learned rules



$R_1^{DT(a=.97)}: t(s_1) \leq .35 \wedge v(s_1) \leq .51 \rightarrow^{69} \langle 1, 0, 0 \rangle$
 $R_2^{SVM(a=.7)}: t(n_2) \leq .45 \wedge t(t_2) > .41 \wedge v(n_2) < 2.41 \wedge v(n_2) > 1.94 \rightarrow \langle .02, .54, .44 \rangle$
 $R_3^{ANN(a=.75)}: v(t_2) \leq 1.28 \rightarrow \langle .0045, .0856, .9099 \rangle$
 $R_4^E: t(c_0) = 280.1 \pm 17.5 \rightarrow^{200} \langle 0, 0, 1 \rangle$

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