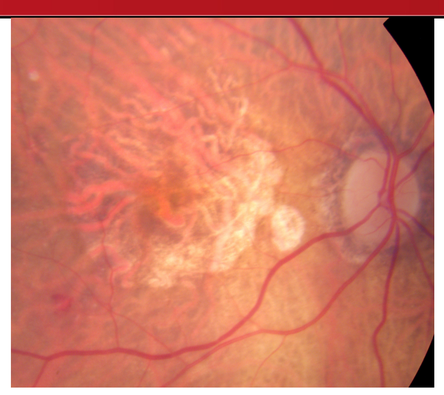
Diagnosis of maculopathies in a private tertiary practice in Australia: retrospective clinical audit

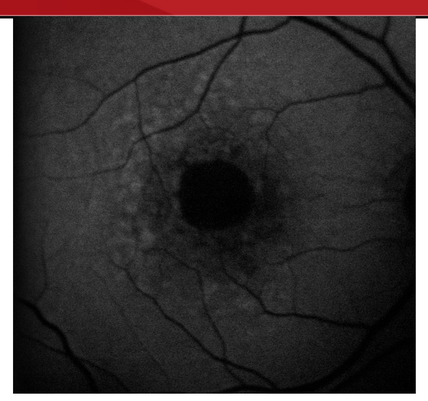
Demi Markakis,[1,2] Alexis Ceecee Britten-Jones,[3,4,5,] Robyn H Guymer,[3,4] Ming-Lee Lin,[6] Simon Skalicky,[3,4,6] Lauren N Ayton,[3,4,5] Heather G Mack[3,4,6]

Background

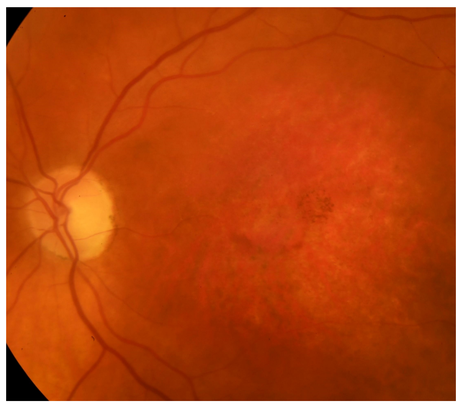
Pegetacoplan[1] and avacincaptad pegol[2] are complement-based therapies recently approved in the USA for treatment of geographic atrophy secondary to age-related macular degeneration (AMD). Accurate diagnosis is imperative for accessing these novel therapies. Currently, diagnosis of AMD relies on nuanced pattern detection of drusen, pigmentary changes or later signs (atrophy and/or neovascularisation) from clinical examination and imaging of the macula. Geographic atrophy secondary to AMD can present similarly to some inherited retinal diseases (IRDs), particularly macular dystrophies[3]. Ocular gene therapy clinical trials for IRD are also underway and in the absence of a chairside guideline to assist in diagnosis of equivocal macular disease, further understanding of the differences between atrophic AMD and atrophy secondary to macular dystrophies is necessary to ensure patients have access to novel treatments.



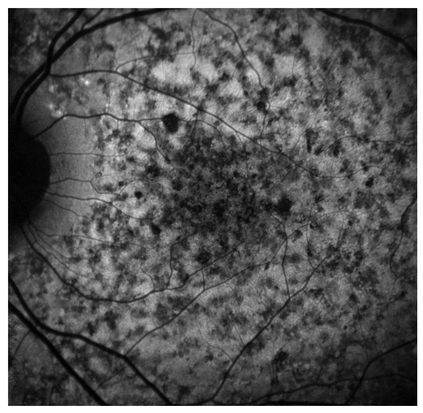
Retinal imaging of an atrophic macula secondary to age-related macular degeneration



Fundus autofluorescence demonstrating geographic atrophy



Retinal imaging of macular atrophy secondary to Retinoschisis 1 mutation



Fundus autofluorescence of macular atrophy secondary to confirmed ABC4 mutation

Aim

This study aimed to estimate the misdiagnosis rate of AMD as an IRD in a tertiary, private practice setting with general and specialist ophthalmologists, at a time where misdiagnosis represents a barrier to accessing potentially sight-saving interventions.

Method

A retrospective audit of 5715 records is being undertaken to assess the rate of misdiagnosis of a macular dystrophy as AMD in a specialist ophthalmology clinic. After screening patient files for confounding history (including retinotoxic medications, oral steroid exposure, coexistent retinal pathologies or known macular dystrophy), retrospective clinical images will be assessed for typical drusen and the presence of atrophy[4]. Cases with atrophy in the absence of typical drusen will be further appraised, including characteristation of atrophy and any other features, such as flecks on OCT. An expert panel including medical retina ophthalmologists and academic optometrists will be consulted to thoroughly assess potentially misdiagnosed cases and to confirm those which may require further genetic testing for IRDs.

Discussion

With advances in clinical treatments underway, this study will estimate the rate of misdiagnosis in a clinical ophthalmology setting. Assessment and discussion of atypical cases of atrophy by our expert panel will allow for identification of patients who may benefit from further genetic testing for potential IRD diagnosis. This information may assist in finding key phenotypical differences to distinguish between the two disease groups. Ultimately, in addition to contextualising the misdiagnosis rate of macular dystrophies as AMD in a tertiary ophthalmology clinic, this study's findings of clinical subtleties may assist specialists internationally to enhance the diagnosis of macular disease and ensure that all patients can access the best treatments available to improve their visual prognosis.

Conclusion

With clinical trials and regulatory-approved treatments for atrophic macular disease underway, an understanding of the clinical nuances between macular dystrophies and AMD is imperative. Only through accurate diagnosis in the consulting suite will patients with AMD or macular dystrophies be able to access the intervention which may prevent further vision loss.

References

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