Hyperautofluorescent parafoveal ring in children with rod-cone dystrophy and literature review

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BACKGROUND

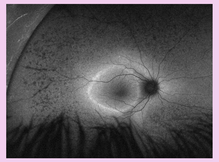
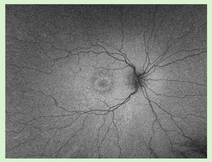
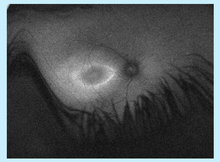
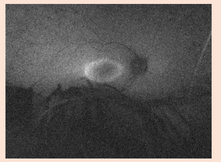
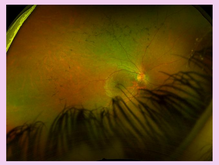
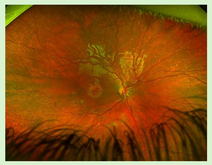
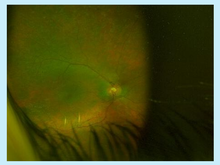
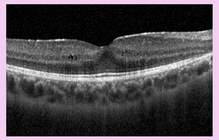
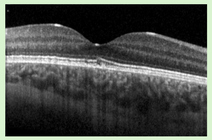
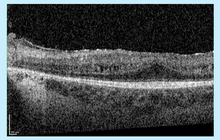
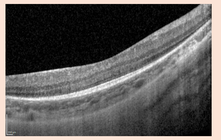
- Autofluorescence is an increasingly useful tool
- A blue light is used to excite natural fluorophores in the retina which then further emit light
- Patterns of autofluorescence can aide diagnosis and determine prognosis
- We report on a series of four children with hyperautofluorescent parafoveal rings with four different genetic variants for retinal dystrophy

METHODS

This is a retrospective review of 4 children from a paediatric ophthalmology clinic in the UK. Clinical data and results of retinal imaging including autofluorescence and OCT macula are reported.

KEY FINDINGS

- All children exhibited a similar pattern and location of parafoveal hyperautofluorescent rings, around the central macula but non-foveal involving
- Vision ranged between -0.14 to 0.42 LogMAR in the better seeing eye of each patient
- Retinal findings on fundoscopy ranged from bone spicule retinopathy, to a normal retina on examination

	Patient 1 Age 16	Patient 2 Age 12	Patient 3 Age 12	Patient 4 Age 9
Autofluorescence				
Colour fundus				
OCT macula (B-scan)				
Best corrected visual acuity (Better eye)	-0.08 LogMAR (unaided)	0.70 LogMAR (pinhole)	0.52 LogMAR (glasses)	0.34 LogMAR (glasses)
Peripheral retinal findings	Bone spicule pigmentation	No peripheral changes – yellow flecks on macula	No peripheral changes	Bone spicule pigmentation

LITERATURE REVIEW

- A hyperautofluorescent parafoveal ring is a recognised fundus autofluorescence pattern reflecting a metabolic transition zone between relatively preserved central retina and surrounding photoreceptor degeneration
- Recent reports have shown hyperautofluorescent rings as a prognostic marker for slower progression in the context of retinitis pigmentosa
- These rings can sometimes be the earliest or sole abnormality, preceding fundoscopic change, visual field loss, or electroretinographic abnormalities

GENETIC VARIANTS

Patient	Gene	Inheritance	Condition
1	CNGB1	Autosomal recessive	Retinitis pigmentosa
2	PRDM13	Autosomal dominant	North Carolina macular dystrophy
3	RNU4ATAC	Autosomal recessive	Roifmann syndrome
4	IDH3A	Autosomal recessive	Retinitis pigmentosa

CONCLUSION

Our report demonstrates the utility of autofluorescence in children. This is particularly useful in the cohort who report unexplained reduced vision in dark or light conditions with minimal retinal findings. Recognition of the hyperautofluorescent parafoveal ring can support earlier diagnosis, guide prognosis, and facilitate genetic testing and counselling.

References