Classification and heritability of macular pigment spatial profile phenotypes using two-wavelength fundus autofluorescence

Byki Huntjens, Irene Ctori, Chris J. Hammond, Katie M. Williams, Omar A. Mahroo

Purpose:

We investigated the frequency and heritability of macular pigment (MP) spatial profile phenotypes determined by objective and subjective profile classification based on fundus autofluorescence (FAF).

Methods:

Between scans Coefficient of Repeatability (CoR) of MP optical density (MPOD) was calculated from two FAF scans (Spectralis, Heidelberg, Germany) of 40 participants (39±8.6 years) acquired in a single session. We then analyzed two FAF scans acquired in a single session from 314 twins (157 pairs; 39±8.8 years) and classified each MP profile as exponential, ring-like or central dip by subjective visual assessment. Profiles were also classified objectively based on deviations larger than the CoR away from the exponential fit. We calculated Kappa agreement of the profiling methods, case-wise concordance of non-exponential profiles for the 88 mono- (MZ) and 69 dizygotic (DZ) twin pairs, and profile heritability.

Results:

Following visual subjective profiling, 64% showed an exponential profile, 27% presented ring-like and 9% central dip profiles; case-wise concordance was 0.80 for MZ and 0.41 for DZ twins. Following objective classification, 71% showed an exponential profile, 29% ring-like profile and no central dip profiles were identified; case-wise concordance was 0.74 for MZ and 0.36 for DZ twins. Heritability was calculated as 81.5% (95% CI 61.1 to 93.1). Between scan repeatability of profile classification showed good agreement objectively (κ=0.85, 95% CI 0.69 to 1.00; P<0.0005) and moderate agreement visually (κ=0.48, 95% CI 0.23 to 0.73; P<0.0005). Agreement of subjective versus objective profiling was low (κ=0.23, 95% CI 0.04 to 0.42; P=0.02).

Conclusions:

MP profiles showed high heritability. Compared to visual assessment, objective profile classification is a more reliable method for future experimental studies using FAF.

max. 1.850 characters. Every character and space is included

1,840 characters

	Visual assessment (Tariq et al. IOVS 2014)		Subjective spatial profiling by visual assessment		Objective spatial profiling	
Classification	%	(n)	%	(n)	%	(n)
Exponential	59	(187)	64	(201)	71	(223)
Ring-like	26	(81)	27	(85)	29	(91)
Plateau Central dip	15	(47)	9	(28)	0	(0)
Case-wise concordance	MZ 0.75	DZ 0.22	MZ 0.80	DZ 0.41	MZ 0.74	DZ 0.36