Purpose: Heritability is the proportion of phenotypic variation in a population that is attributable to genetic variation among individuals. Many ophthalmic disorders and biometric traits are known to have a genetic basis and consequently much work has been published in the literature estimating the heritability of various ocular parameters. The purpose of this review was to collate and summarize the findings of heritability studies conducted in the field of ophthalmology. We also wished to compare family and twin studies and methods of heritability estimation.

Methods: We conducted a systematic search on Medline, ISI Web of Science, Scopus and Google Scholar using selected keywords with no date or language limitations. Any study that reported a heritability estimate for an ophthalmic trait or allowed one to be calculated from the data provided was included. We grouped the various studies broadly by phenotype as follows: refraction, glaucoma, age-related macular degeneration (ARMD), cataract, diabetic retinopathy and others.

Results: A total of 82 articles were retrieved from the literature relating to estimation of heritability for an ocular disease or biometric trait. Thirty-seven were concerned with glaucoma, 28 papers with refraction, four with ARMD, five with diabetic retinopathy and four with cataract. The highest reported heritability for an ophthalmic trait is 0.95 for central corneal thickness, indicating that observed variation in this parameter is largely governed by genetic factors. Conversely, corneal fluorescence has been shown to be non heritable with variation wholly attributable to environmental influences. Over 60% of the studies employed a twin study design and a similar percentage utilized variance components methods and structural equation modelling (SEM) to derive their heritability values. Using modern SEM techniques, heritability estimates derived from twin subjects were generally higher than those from family data.

Conclusions: In this systematic meta-analysis we have compiled a concise archive of heritability studies conducted in the field of ophthalmology. Many of the estimates are in the moderate to high range, but to date the majority of genetic variants accounting for these findings have not been uncovered, hence much work remains to be undertaken to fully elucidate their molecular etiology.