A RARE PUPILLARY DEFECT
Inherited by Identical Twins

BENJAMIN V. WHITE, JR. and MARSHALL N. FULTON
Medical Clinic, Peter Bent Brigham Hospital, Boston, Mass.

On the basis of a number of close similarities these twins appear to be identical. They have a very rare eye defect, in which the pupils are not able to accommodate normally to light by contraction of the iris (see Figure 10).

ALTHOUGH many characteristics of the human eye are transmitted by heredity, notably various degrees of coloboma, refractive errors, and iris color, inherited defects of the pupil alone are distinctly less common. Congenital miosis (pin point pupil) and ectopia pupillae are generally recognized in treatises on ophthalmology and on heredity. Membrana pupillaris perseverans, coloboma iridis, and aniridia are also mentioned by Fuchs, while de Schweinitz notes as hereditary the cat-like pupil originally described by Greef. In this deformity the pupils constrict medially so as to form an elongated slit. De Schweinitz also states that slight inequality of the pupils may be hereditary.

Pupils with multiple irregular cup-shaped indentations into the margin of the iris are also described. Lindberg reports a series of four cases with this type of deformity. Gilbert describes a single case and notes that the affected pupil is larger than the normal one, but reacts to light and on accommodation in the normal manner. Bernheimer also reports a single case. All of these defects were unilateral.

The purpose of this communication is to report the finding of large irregular pupils in a pair of homozygous twins, a syndrome which Macklin did not encounter in a review of over 3,000 case reports of hereditary abnormalities. The malformation observed in these twins is also said to have been present in the
EGG-SHAPED PUPILS

Detailed view of eyes of twins S.L. (A) and C.L. (B). In the presence of a bright light, the pupils can contract only in certain regions, giving an egg-shaped outline. The right eye of C.L. (B) has accommodated more than the other three eyes.

Of the twins reported in this study, one, S.L., has been a patient on two occasions in the Peter Bent Brigham Hospital. The other, C.L., has been observed during her visits to her sister. The detailed history of our patient reveals no symptoms having an obvious bearing on the optic defect which is the subject of this report, but the history can be obtained by workers who are interested.*

On her second admission to the hospital (June 3, 1935), the patient’s pupils were carefully observed. They were found to be extremely large, irregular, and to respond to light and during accommodation only in the superior temporal, and very slightly in the inferior nasal quadrants. Peripheral visual fields and visual acuity were essentially normal as was the ophthalmoscopic examination of the eye grounds.

C.L. (Figure 9B) was not a patient at this hospital but was observed on visits to her sister. It was noted that her pupils in the resting state were in every respect identical with those of her sister. Under the strain of strong illumination, however, the right pupil showed a somewhat more marked constriction. (Figure 10.)

The women were of Russian Jewish

*Copies of the detailed medical history are on file with the American Genetic Association, and will be furnished with the author’s reprints. The history may be obtained by writing the American Genetic Association.—Ed.
White and Fulton: Iris Defect in Twins

Small differences in generally similar patterns characterize the twins.

1. Homolateral Characteristics: Asymmetrical pupils, deviation of nasal septa to the right, unilateral skin furrow over the bridge of the nose.
2. Heterolateral (mirrored) Characteristics: S.L.'s hair whorl, which permits parting on the right, C.L.'s, which permits parting on the left. The small mole on the right side of S.L.'s chin, on the left of C.L.'s.
3. Dental Deformities: Separation of the lower central incisor teeth in both subjects.
4. Blood Grouping: Both subjects of Moss's Group III, one of the less frequently encountered blood groups.
5. Finger Prints: Corresponding types of fingerprint patterns, on all fingers except the index fingers of the two hands. These fingerprint types are diagrammatically reproduced in Figure 11.

The characteristics of the pupils observed in these twins were: a, dilation; b, irregularity; and c, reaction to constricting stimuli only in the superior temporal and, to a lesser extent, in the inferior nasal quadrants. Slit lamp examination failed to reveal definite evidence of coloboma in either the iris or the choroid. There was, however, a small retinal naevus in the right eye of C.L., a finding often seen in the presence of colobomata. Macklin? has suggested that the deformity may represent an inherent deficiency in the sphincter iridis muscle.

The family pedigree is given in Figure 12. The presence of the deformity in the mother of the twins suggests that the character is dominant in heredity. Its absence in the five children of the F2 generation, all of whom have been examined by us, does not disprove this assumption, though it suggests that the gene for the iris deformity probably has normal overlaps and hence that it is an irregular dominant. The siblings of the twins do not show the defect so far as can be determined.

We are indebted to Dr. Clyde E. Keeler and Dr. Trygve Gundersen for valuable assistance they have given us in preparing this report.

Summary
1. The finding of large, irregular pupils in homozygous twins is reported.
2. The pupils were large, irregular, and incapable of responding to constricting stimuli except in certain quadrants, which were the same in both subjects.
3. The mechanism of inheritance seems to be irregular dominance.

Literature Cited
7. ———. Personal Communication.
Parent Fuerte avocado tree in the LeBlanc garden at Atlixco, Mexico. It was first noticed in 1908 and buds were cut from it for testing in southern California in 1911. At the present time about 75 per cent of the commercial avocados raised in California are of this variety. Its fruits mature in December and January at Atlixco and are of a greenish color. Photographed October 7, 1936.