RETINAL DETACHMENT ASSOCIATED WITH COLOBOMA OF THE CHOROID*

BY *Koanglu Wang,* MD (BY INVITATION) AND
George F. Hilton, MD

INTRODUCTION

A COLOBOMA OF THE CHOROID IS CHARACTERIZED BY THE CONGENITAL ABSENCE OF a portion of the retinal pigment epithelium and choroid. It is recognized clinically as a prominent white zone of the fundus which is usually located inferiorally and slightly to the nasal side. There is a very thin layer of rudimentary retina with a few blood vessels overlying the sclera, which may be ectatic. This anomaly is due to a defect in the normal fusion of the fetal fissure of the optic cup. Normally, the two layers of the optic cup close at the fissure near the equatorial zone and the fusion process then extends anteriorly to the pupil and posteriorly to the optic nerve.

Choroidal coloboma is an infrequent finding and was found in only 0.14% of a large series of eye patients.[1] It may occur as an isolated finding, but is frequently associated with other anomalies, such as iris coloboma, microphthalmus, microcornea, cataract, lens coloboma, and optic pit. It may also be associated with various systemic birth defects.[2,3] It is often found in several congenital syndromes, such as the syndromes of Goldenhar,[4] Schmid-Fraccaro,[5] Joubert,[6] Mohr-Clausen,[7] and Aicardi.[8,9] Some of these patients have been found to have defects in one or more chromosomes.[10-12]

Our review of the literature has disclosed 43 eyes with choroidal coloboma associated with detachment of the retina (Table I).[13-23] The largest previous series was ten eyes.[16] Our present paper reports 26 additional eyes.

*From the Department of Ophthalmology, University of California, San Francisco, California.

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TABLE I: COLOBOMA WITH RETINAL DETACHMENT (SURVEY OF THE LITERATURE)

YEAR	AUTHOR	NO OF EYES	NO OF EYES OPERATED	NO OF EYES REATTACHED
1922	Brinton[13]	1	0	—
1925	Wagener and Gipner[14]	2	0	—
1926	Komoto[15]	3	0	—
1961	Jesberg and Schepens[16]	10	7	4
1969	Rendahl[17]	3	2	0
1969	Gailloud et al[18]	7	*	*
1972	Limaye[4]	1	0	—
1978	Zhang[19]	4	4	2
1979	Zinn[20]	1	1	1
1980	Bao[21]	5	4	2
1981	Patnaik and Kalsi[22]	5	4	2
1983	Gonvers[23]	1	1	1
1985	Present study	26	20	7
Total		69	42	18

*Data not available.

METHODS AND MATERIALS

All patients were referred to the Tong Ren Hospital, Peking (Beijing), China, for retinal detachment surgery. The preoperative evaluation included visual acuity, motility, refraction, pupils, tension, biomicroscopy, and binocular indirect ophthalmoscopy.

Preoperative findings are summarized in Table II and Fig 1. The absolute incidence of bilaterality could not be determined, because the retina of the fellow eye in eight patients could not be visualized due to cataract or phthisis. All 26 eyes also had coloboma of the iris and the majority also had cataract, nystagmus, microphthalmus, or microcornea (Table III). Some patients also exhibited strabismus and optic nerve coloboma. Most detachments were relatively old and all four quadrants were involved in 81% of cases (Table IV). In only ten eyes could we identify retinal breaks within the rudimentary retina of the coloboma. Eight eyes had retinal breaks outside of the colobomatous area and no breaks could be found in nine eyes (Table V). All breaks in the colobomatous rudimentary retina were of the round or oval atrophic type.

Twenty of the eyes were operated. A scleral buckling procedure was used for all cases. Radial scleral buckling on both sides of the coloboma was utilized in 14 cases. These two converging lines are reminiscent of the Chinese figure for the number 8. This operation has, therefore, come to be known as the Chinese "Figure of Eight" scleral buckle (Figs 2 and 3).

TABLE II: RETINAL DETACHMENTS ASSOCIATED WITH CHOROIDAL COLOBOMA (1970-1979)

No of patients	25
No of eyes	26*
Male:female	19:6
Age range	11-29
Race	Chinese
Right:left eyes	10:16
Vision 20/400 or worse	20
Coloboma surround disc	14
Incidence	1.7% of 1536 detachments

*In eight cases the retina of the fellow eye could not be visualized.

RESULTS

Permanent retinal reattachment was accomplished in only seven eyes (Table VI). The final visual acuity was 20/200 in two eyes, 20/400 in two, and the other three cases had a final vision of counting fingers. There were very few postoperative complications, but one eye experienced a prominent vitreous hemorrhage, one developed cataract and vitreous haze, and there was one case diagnosed as sympathetic ophthalmia.

DISCUSSION

EMBRYOGENESIS

The embryonic optic vesicle invaginates to produce the optic cup and this has a cleft or fissure in the inferior aspect of the cup. This fetal fissure typically closes by a fusion process. Defective closure can be partial or complete. The spectrum extends from a very small colobomatous defect just below the optic nerve to a total coloboma extending from the pupil posteriorly to the optic nerve.

ASSOCIATED OCULAR DEFECTS

Coloboma of the choroid is often associated with coloboma of the iris. Other defects may include microphthalmus, microcornea, cataract, optic nerve coloboma, posterior scleral staphyloma, retrobulbar cyst, nystagmus, and strabismus.

HISTOLOGY

There is total absence of the retinal pigment epithelium and choroid, with the rare exception of an occasional choroidal vessel which still may be

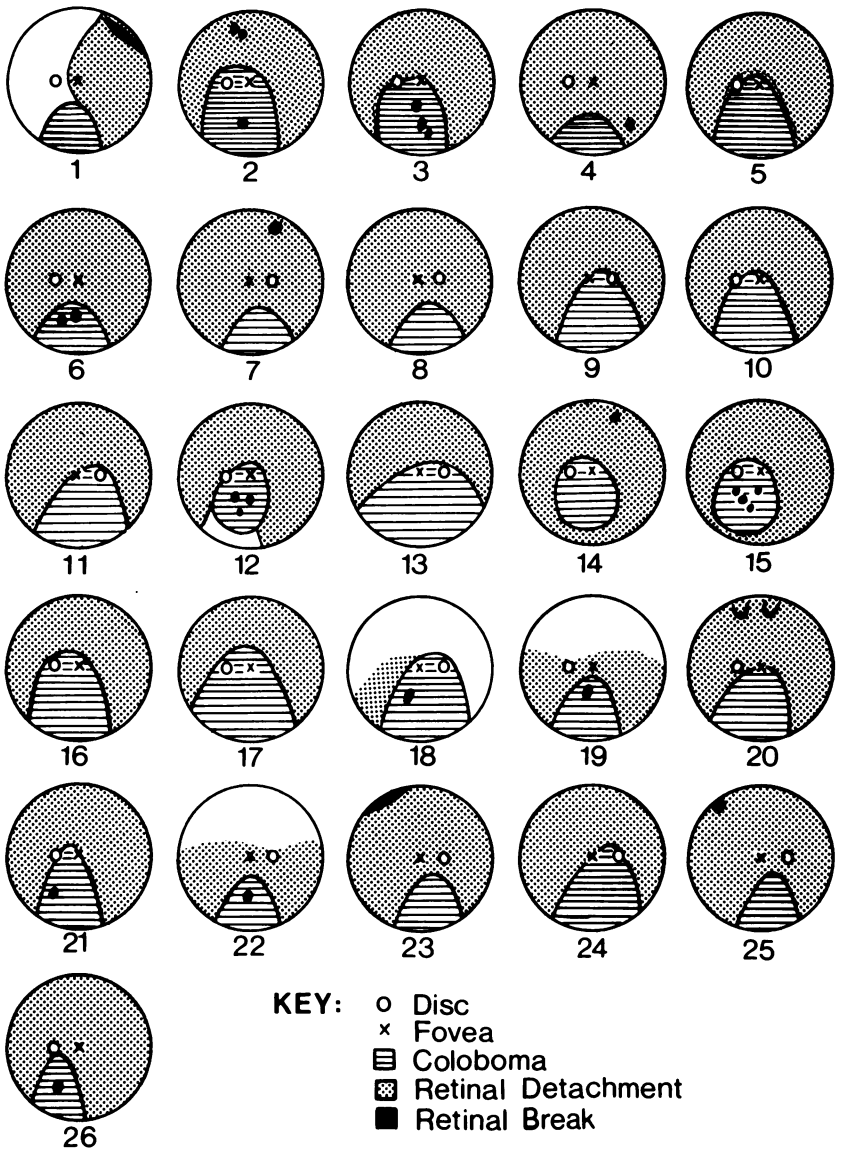

FIGURE 1
Twenty-six eyes with retinal detachment associated with choroidal coloboma.

TABLE III: ASSOCIATED OCULAR ANOMALIES

ANOMALIES	NO OF EYES (%)
Coloboma of iris	26 (100%)
Microphthalmos/ microcornea	16 (61.5%)
Cataract	20 (76.9%)
Optic nerve coloboma	3 (11.5%)
Nystagmus	16 (61.5%)
Strabismus	6 (23.1%)

seen to traverse the colobomatous defect. The sclera may be thin and, occasionally, staphylomatous. The retinal tissue is very thin and various authors have referred to this rudimentary retinal tissue with the following terms: abnormal, underdeveloped, undifferentiated, diaphanous, flimsy, anomalous, maldeveloped, and rudimentary.

JUVENILE RETINAL DETACHMENT

All of our patients were under the age of 30 and most other cases in the literature also involve young people. The differential diagnosis for juvenile retinal detachments includes: persistent hyperplastic primary vitreous, juvenile sex-linked retinoschisis, Marfan's syndrome, inferior temporal dialysis of the young, retinoblastoma, congenital retinal folds, retrolental fibroplasia, juvenile aphakic detachments, von Hippel's syndrome, Coats' disease, proliferative diabetic retinopathy, trauma, retinal dysplasia, Wagner's dystrophy, sickle cell retinopathy detachment, congenital nonattachment of the retina, optic pit with serous detachment, morning glory detachment, and giant retinal tear with coloboma of the lens.

PATHOGENESIS OF COLOBOMA DETACHMENT

Most eyes with choroidal coloboma do not develop retinal detachments. The reason for the occasional detachment is not clear, but some cases have been characterized by preretinal tissue in the area of the rudimen-

TABLE IV: EXTENT OF RETINAL DETACHMENT

NO OF QUADRANTS	NO OF EYES
One	1
Two	3
Three	1
Four	21
Total	26

TABLE V: RETINAL BREAKS

LOCATION	NO OF EYES	TYPES OF BREAKS		
		ROUND/OVAL	HORSESHOE	DIALYSIS
Inside of coloboma	9	9	0	0
Outside of coloboma	7	4	1	2
Inside and outside	1	1	0	0
No breaks found	9	—	—	—
Total	26	14	1	2

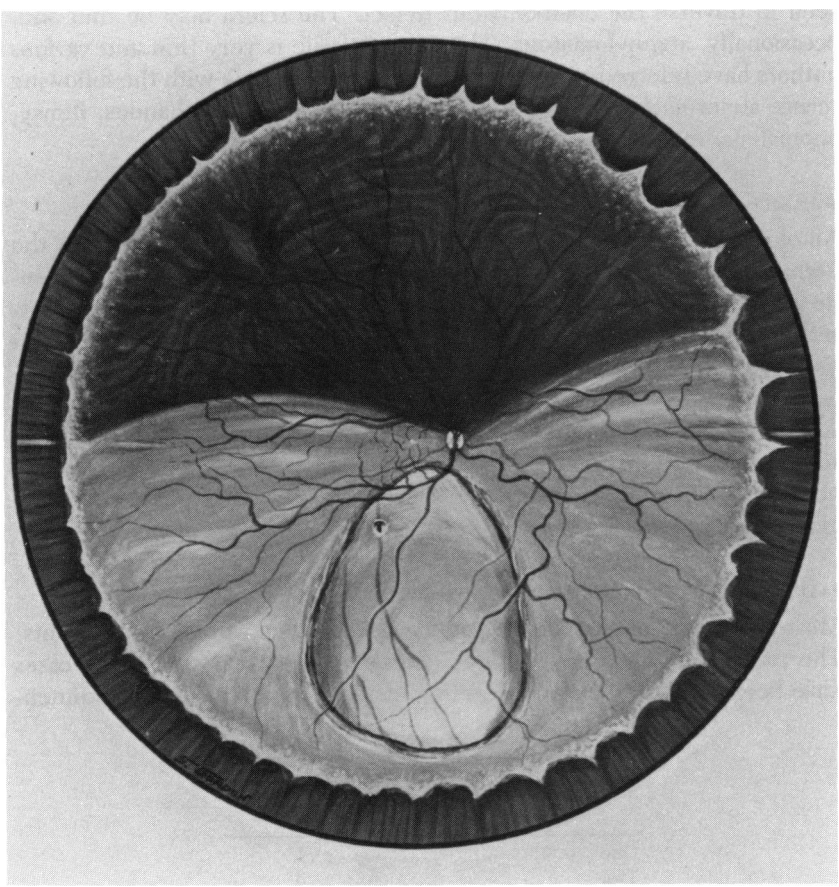

FIGURE 2
Case 22: Retinal detachment caused by an atrophic hole in rudimentary retina in choroidal coloboma.

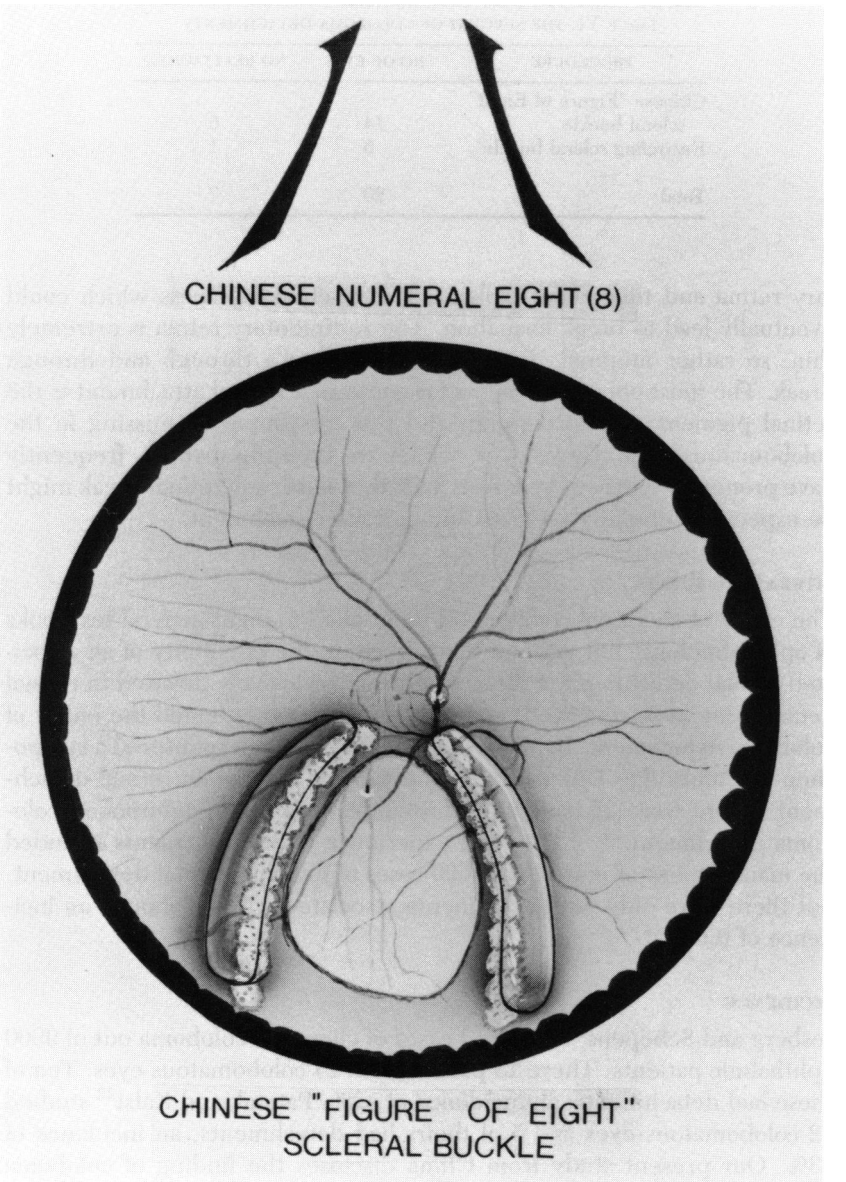

FIGURE 3
Case 22: Retinal reattachment following cryotherapy, drainage of subretinal fluid, and two radial scleral buckles (Chinese "Figure of Eight" scleral buckling operation).

TABLE VI: THE SURGERY OF COLOBOMA DETACHMENTS

PROCEDURE	NO OF EYES	NO REATTACHED
Chinese "Figure of Eight" scleral buckle	14	6
Encircling scleral buckle	6	1
Total	20	7

tary retina and this could explain some tractional process which could eventually lead to break formation. The rudimentary retina is extremely thin, so rather minimal changes could lead to a through and through break. The most effective process for continued retinal attachment is the retinal pigment epithelial pump and this, of course, is missing in the colobomatous area. These eyes, which are typically myopic, frequently have prominent vitreous syneresis and, therefore, any retinal break might be expected to rapidly progress into a frank detachment.

LITERATURE SURVEY

The entity of choroidal coloboma is mentioned in eight general textbooks of ophthalmology, but none of them mention the possibility of an associated retinal detachment. Fifteen textbooks exclusively devoted to retinal detachment were reviewed, and only 4 of them mentioned the entity of coloboma detachment. In 1968, the Club Jules Gonin sponsored a symposium at Cambridge University on the subject of juvenile retinal detachment. There were 22 papers on this subject but only 2 mentioned coloboma detachment.[17,18] The total experience of all participants included the management of more than 1600 cases of juvenile retinal detachment, but there were only ten detachments associated with coloboma: an incidence of 0.6%.[24]

INCIDENCE

Jesberg and Schepens[16] found 13 cases of choroidal coloboma out of 9900 ophthalmic patients. These 13 patients had 24 colobomatous eyes. Ten of these had detachments: an incidence of 42%. Patnaik and Kalsi[22] studied 22 colobomatous eyes and 5 of theirs had detachments, an incidence of 23%. Our present study from China discloses the finding of coloboma detachment in 1.7% of 1536 consecutive retinal detachments. This is considerably higher than the previously mentioned incidence of 0.6% found in Europe and North America.[17,18,24] There may be a definitely higher incidence in the Chinese race, but this may be an artifact of the

referral system utilized for patients to reach the medical center in Peking. If our series is combined with that of Jesberg and Schepens,[16] together with that of Patnaik and Kalsi,[22] we find a total of 37 males and 15 females with retinal detachment. This combined data seems to support the impression that the entity of coloboma detachment is definitely more common among male patients.

CLINICAL FEATURES

Our findings agree with several other reports in which we note that in some cases the coloboma is nothing more than an incidental finding in a rhegmatogenous retinal detachment definitely due to a break in full thickness retina elsewhere in the fundus. Of special interest is the group of patients where the detachment seems to be due to a break in the rudimentary retina found in the area of the coloboma. The elevation of the retina at the coloboma edge is typically very shallow, and sometimes it can only be appreciated with detailed biomicroscopy. There is usually a pigmented line along the edge of the coloboma and this might suggest a demarcation line with some adhesion, but this is not the case. This pigment line does not afford any barrier to the movement of subretinal fluid. The finding of breaks in the rudimentary retina is extremely difficult for several reasons: (1) the rudimentary retina is extremely thin, (2) the breaks are usually atrophic holes without the presence of flaps or operculae, (3) the white background of the sclera makes visualization of the breaks very difficult, (4) the tiny breaks may be hidden under the overhanging edge of the coloboma, and (5) tiny breaks may be hidden in areas of hemorrhage.

SURGERY

Several different types of scleral buckling procedures have been utilized in the attempted repair of these difficult detachments. Our limited success was generally associated with two radial scleral buckles, one on either side of the coloboma, typically placed from the ora serrato posteriorly to a point adjacent to the optic nerve. Treatment of the bare sclera of the coloboma would not produce an effective bond and, therefore, treatment is limited to the area of normal tissue along the edge of the coloboma. Either diathermy or cryotherapy may be utilized.

The two radial buckles that converge posteriorly are reminiscent of the figure in Chinese script that refers to the numeral 8. Therefore, in Peking, it is common to refer to this operation as the "Figure of Eight Buckle." The anglicized equivalent for this would be "two radial converg-

ing scleral buckles." Gonvers[23] has reported one successful case with the use of intraocular silicone tamponade. We now feel that the preferred method would be vitrectomy, endophotocoagulation, and fluid-gas exchange with postoperative positioning.

PROPHYLACTIC TREATMENT

Patnaik and Kalsi[22] have suggested the use of prophylactic treatment by laser application along the edge of the coloboma posteriorly and cryoapplication anteriorly. Jesberg and Schepens[16] have pointed out that any treatment along the coloboma edge creates nerve fiber bundle defects, so it would be of interest to note the nature of the visual field in the eyes that were treated prophylactically. The suggestion of Jesberg and Schepens[16] would imply that there would be a major loss of most of the visual field.

PROGNOSIS

Virtually all previous reports agree with Jesberg and Schepens,[16] who state that "colobomatous eyes do not tolerate surgery well." Surgical results are poor and operations are technically difficult due to the thin ectatic sclera, absence of choroid, and marked thinning of the rudimentary retina.

RESULTS

Fourteen eyes in our series were operated on with the Chinese "Figure of Eight" buckling procedure and 6 of these were reattached (43%). Six of our cases were managed with an encircling scleral buckle and only one was reattached. Our final anatomic cure was 7 of 20 (35%). This may be compared with the rather poor results generally noted in the literature (Table I). The visual result in all of our seven reattached cases was 20/200 or worse. This is partially explained by the fact that the coloboma completely surrounded the disc in the majority of our cases and the detachments were several weeks to months old before coming to surgery.

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DISCUSSION

DR ROBERT B. WELCH. The authors have presented a number of cases of an unusual type of retinal detachment that is associated with a coloboma of the choroid. Their series is unique in that it comes from Peking, China, and represents 1.7% of the total number of retinal detachments seen over a 9-year period. Indeed, their 26 eyes is the largest series ever reported in the literature which spans a period of over 60 years. To obtain a comparison, I reviewed my own retinal detachment experience and found an incidence of detachment and coloboma in 0.12%. Since coloboma of the choroid is often known to be of dominant inheritance, I would like to ask the authors if there are large pedigrees of genetic syndromes associated

with colobomas in this area of China and if indeed a family history was obtained in their patients?

Doctor Hilton and Doctor Wang have nicely reviewed the literature and clearly presented the clinical features, stressing the fact that there are detachments caused by peripheral breaks in eyes with colobomas as well as true colobomatous detachments secondary to breaks within or at the edge of the coloboma. Their surgical approach to these cases was the use of scleral buckling procedures and their success was highest with radial buckles along the edges of the coloboma, the so-called "Chinese Figure of Eight" procedure. Even so the success rate was low. Because of this and the technical difficulties in placing extensive radial buckles, I would propose that for those cases without peripheral breaks and with or without identifiable breaks within the coloboma a vitrectomy procedure with intraocular gas be considered, perhaps followed by laser treatment along the edge of the coloboma.

I would like to close with some provocative questions for Doctor Hilton and Doctor Wang. How do these cases differ from colobomas of the disc, optic nerve pits or the morning glory anomaly, all of which may be associated with retinal detachments? Could some of these detachments be associated with subarachnoid communication or are breaks always present? Is vitreous traction a major component?

I would like to congratulate Doctor Hilton and Doctor Wang on a very interesting and well prepared paper.

Dr Harold Spalter. Just a question on discussion. I had one case fail to reattach. But at a time antecedent to the appearance of the detachment, this individual, who was a 25-year-old woman—the age right, the sex incorrect according to your statistics, did develop a pharmacologically induced acute narrow angle glaucoma bilaterally. I wonder among your associated ocular anomalies whether the Chinese history has revealed any increased incidence, as an associated finding of narrow angles, perhaps again as an inherited problem and that is the question.

Dr Robert Drews. I would like to ask Doctor Hilton if he noted whether any of these patients had heterochromia of the iris as well as the coloboma.

Dr William G. Everett. Recently a small group of ophthalmologists toured through China visiting various eye departments and visited the same one Doctor Hilton refers to in Peking. I think what impressed us more than anything else was that if you analyze their number of cases, you find that this major clinic in Peking with a population in excess of 14 million was doing only 150 retinas a year. This was a large number of cases to them but to us was unbelievably small considering that they were one of the only three major retinal referral centers for this area. They were drawing from surrounding populations probably in the neighborhood of 30 million. Their data does not in any way compare to ours where a comparable metropolitan area probably would have produced 1000 retinal detachments at each of the three eye units. The high number of bilateral cases suggests to me that

they usually went to the eye clinic when the second eye was involved. All the data does not really relate and compare to the data from our country. I would like that having observed the level of care of retinal work in China, rather than suggest vitrectomy, fluid/air exchange, etc, they must learn the basic fundamentals of retinal detachment surgery as they are probably 20 years behind us in their level of treatment. I don't think they are ready for vitrectomy procedures as yet, where indirect ophthalmoscopy has achieved only rare usage.

Dr H. MacKenzie Freeman. My comments are only anectodal. They are only based on two cases but they relate to the suggestion that vitrectomy air/fluid exchange and endophotocoagulation could be worthwhile in these cases. I think it is very worthwhile. The two cases that I did do with endophotocoagulation, vitrectomy and air/fluid exchange were reattached. It is very difficult to place a radial buckle as far posteriorly as the disc. It is very difficult to place cryo and endodiathermy and not without complications but these two cases did work and I would suggest that additional cases be handled in that manner.

Dr Ronald G. Michels. We have had experience similar to that of Doctor Freeman, and we have also used vitrectomy and fluid/gas exchange, followed by photocoagulation around the margin of the coloboma, to treat three cases with retinal detachment due to breaks within the coloboma. These cases have been successful. However, our experience has shown that a firm chorioretinal adhesion is needed or there will be recurrent detachment. To achieve a firm chorioretinal adhesion, prolonged intraocular tamponade is useful. Air alone is not sufficient in most cases and tamponade lasting for about 1 week is preferable. We prefer sulfur hexafluoride or another long acting gas.

To answer one of the questions Doctor Welch raised, we have recently published our experience with a single case of the morning-glory type coloboma and rhegmatogenous detachment due to a break within the coloboma. In that eye we evacuated the subretinal fluid through the break by vitreous surgery means, thus demonstrating a communication between the vitreous cavity and the subretinal space.

I agree with Doctor Hilton that optimal management of these cases probably involves delimiting the area of the coloboma by photocoagulation because it seems impossible to flatten the retina within the area of the coloboma. If the coloboma surrounds the optic nerve, as in the case of the morning-glory syndrome, Krypton laser photocoagulation may be advantageous to minimize damage to the overlying nerve fiber layer. We used Krypton in our case of the morning-glory syndrome, and the patient recovered vision to the level that had been documented prior to the detachment.

Dr Sloan Wilson. I would like to simply make one additional comment related to your very nice paper, Doctor Hilton, and to agree with the two previous discussants. I suggest you consider a scleral reinforcement at the time of your original

surgery, such as the Miller-Thompson-Snyder procedure for high myopia and posterior staphyloma. This would give posterior support in addition to the vitrectomy.

Dr George F. Hilton. I am grateful for my colleagues who have added some constructive comments as well as raising a few interesting questions. There are a number of systemic syndromes that include coloboma as part of the picture and those have been listed in our paper that will be published. We did not identify any of these syndromes within this particular population of China. I am grateful that three of our commentors have agreed with the notion of vitrectomy and pneumatic retinal reaattachment with a long-acting gas tamponade and we hope to pursue that further. Regarding the possible relationship to the macular detachments seen with pits of the optic nerve head, we think this was quite different. These detachments are not rhegmatogenous and we feel that it is unlikely that we would cure them with either a buckle or air exchange. I think the mechanism of pit-related detachments still remains debatable and it has indeed been debated here many times. As to the possible role of vitreous traction, I will just say it was not identified on the biomicroscopic examination around these little breaks. They all had vitreous detachments. We did not see any instances of narrow angles, or angle closure glaucoma, nor was heterochromia recognized. Thank you for your comments and questions.