BLUE SCLERA, BRITTLE BONES
AND RETINAL DETACHMENT.

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Blue sclera is the most constant
manifestation of a congenital multiple
mesodermal syndrome that may also ex-
hibit increased fragility of bones, deaf-
ness due to otosclerosis as well as more
unusually lax joints, arachnodactyly, syn-
dactyly, cleft palate, spina bifida, congeni-
tal heart disease, macular atrophy of the
skin and anomalies of teeth. In the eye
anterior embryotoxon, cataract, color
blindness and lid deformities have been
reported as part of this syndrome in addi-
tion to blue sclera.\(^1\) The abnormal blue
coloration of the sclera is due to an
increased transparency and the term “clear
sclera” has, therefore, been suggested in
stead of blue sclera.\(^1\) The anatomical cause
of the increased transparency has been a
subject of dispute. Most observers be-
lieve, however, that general scleral thin-
ing is the cause.\(^2\) This thinning is said to
go along with immaturity and sparcity of
the collagen fibers. One would expect
retinal detachment to be a common com-
plication of this syndrome—since retinal
detachment commonly occurs in associa-
tion with localized scleral thinning (sta-
phyllomas).\(^6\) A review of the literature,
however, revealed no reported occurrence
of retinal detachment in association with
blue sclera and osteogenesis imperfecta.

Retinal detachment in a well developed
case of osteogenesis imperfecta congenita
with blue scleras is reported in this paper.

Case history:

This 26 year-old white female is the
only child of normal parents. She was a
full term baby weighing 6 pounds and
1\(\frac{1}{4}\) ounces. During the delivery the right
femur was fractured; one month later the
left hip was accidentally broken when a
physician attempted to straighten the
child’s legs. After several more fractures
including fractures of the spine the diag-
nosis of osteogenesis imperfecta congenita
was made. Many more fractures during
the first years of life caused extensive
crippling. Multiple orthopedic surgery
procedures resulted in useful arms and
hands. However, the patient cannot walk.
She measures 3 feet 2 inches in length
(Fig. 1).

Blue scleras had been noted many
years ago. Glasses to correct slight my-
opia were prescribed about 10 years ago.
However, the patient is not deaf and has
no other obvious anomalies.
On 2-15-67 the patient suddenly noted a shower of floaters in her left visual field. Along with this a shadow came into her left visual field from the nasal side. This progressed quickly and caused loss of left central vision within about a day. Retinal detachment was diagnosed and the patient was admitted to the University Medical Center on 2-19-67.

The eye examination revealed her vision to be OD: 20/25 and OS: counts fingers in 2 feet eccentrically. Her glasses measured OD: \(-4.50\) sph \(+1.00\) cyl \(x\) 180 and OS: \(-3.00\) sph \(+1.25\) cyl \(x\) 180. Except for very definitely blue scleras the eyes were externally normal. Fundus examination showed left retinal detachment due to a round retinal hole of the atrophic type in the 2 o'clock periphery (Fig. 2). Areas of advanced retinal degeneration were seen in the down and out periphery of the left fundus.

An operation for retinal detachment was done in local anesthesia on 2-20-67. The conjunctiva of the left eye was opened from 12 to 6 o'clock and the lateral rectus muscle was temporarily detached. The hole was localized and a scleral trough was created over it extending from 12 to 3 o'clock and from 12 to 17 mm behind the limbus (Fig. 3). The bluish sclera was found to be very thin and of poor strength and elasticity. Diathermy was applied all over the trough. Penetration of the choroid at 3 o'clock released extensive retroretinal fluid. A roll of Teflon mesh was placed in the trough and the trough was closed over this with single silk sutures. The closure of the trough was possible in spite of the thin sclera since the eye had become soft after release of the retroretinal fluid. Surface diathermy was applied to the inferior sclera in the region of the extensive degeneration until rows of reactions were seen on the retina surrounding these areas. The muscle was reattached and the conjunctiva was closed.

The postoperative course was entirely without complications. The hole remained on the surgical buckle and the retina in
place. Vision improved. The patient was discharged from the Hospital on 2-25-67.

A follow-up examination on 5-9-67 revealed good external healing (Fig. 4). Vision was unchanged in OD and OS: —3.50 sph + 1.50 cyl x 180 = 20/40. The left retina was in place and the region of the old retinal hole was well scarred. The scleral buckle, however, had flattened considerably.

Comments:

Patients with the fully developed syndrome of blue sclera and osteogenesis imperfecta used to die early. This may explain the fact that retinal detachment has not been known as a complication in patients with this syndrome. Modern medical treatment and care have resulted in a longer life of these patients. The retinal detachment in the present case may, thus, be considered a late complication due to relatively long survival.

The clinically evident blue sclera in the present patient was during surgery found to be due to extensive general thinning and translucency of the scleral tissue. This made it difficult to create a scleral trough. Poor strength and elasticity of the sclera was observed when the scleral trough was closed over a Teflon mesh implant. The original plan to use an encircling band was abandoned since it was thought that such an encircling band of any material could easily cut through and migrate into the eye. It was realized, at the same time, that a segmental implant also was not ideal since it could easily be extruded from the trough. This latter complication, however, appeared less dangerous. The follow-up showed that the segmental Teflon implant did not result in a permanent scleral buckle, but its flattening caused no complications.

The choroid was also found to be thin and of little pigmentation in this eye. The vitreous was fortunately in good condition and showed virtually no strands. The retinal hole was atrophic in type and it layed flat on the buckle created by the

Fig. 2: Schematic drawing to show the arrangement of the retinal detachment, hole and degenerations in the left eye.

Fig. 3: Schematic drawing to illustrate the surgical procedure. A rolled-up piece of Teflon mesh was used as a segmental implant in the scleral trough.

Fig. 4: External view of well-healed surgery site one month after the operation. Photograph.
scleral implant when the retroretinal fluid was withdrawn.

It seems remarkable that the eye muscles as well as Tenon's capsule were thick and well developed in contrast to the thin and clear sclera. This may explain in part the easy and uncomplicated postoperative period with very good superficial healing.

It is hoped that the present case with its favorable operative course will serve as a warning. It may be wise to do a careful fundus examination in all cases of blue sclera as part of their regular medical observation to find retinal breaks before they cause detachment.

Summary:

Retinal detachment occurred in a well developed case of osteogenesis imperfecta congenita with blue scleras. The detachment was successfully treated with retinal diathermy and a segmental scleral implant of Teflon mesh.

References:


