

# BLUE SCLEROTICS ASSOCIATED WITH BRITTLE BONES

REPORT OF A CASE IN A CHILD TWO YEARS OLD \*

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Blue-tinged sclerotics are not infrequently seen in infants and young children. In tuberculous patients also such a blue tinge may occasionally be noted. Congenital heart disease with marked cyanosis may be associated with blue sclerotics, and is largely due to venous congestion. In negro infants and children I have sometimes noted patches of pigmentation in the sclerotics. The blue sclerotics which I am about to describe are entirely distinct from any of these. The condition is congenital and the sclerotics are of a uniform blue color. The first to note the peculiarity was von Ammon;<sup>1</sup> his description is as follows:

Congenital diseases of the sclerotic are rare. . . . Of importance is a peculiar whitish blue coloration of this membrane occasionally met with when the whole development of the eye is retarded. The sclerotic in such cases appears thin and almost transparent. I have seen it also in congenital hydrophthalmus. . . . A similar thinness occurs in patients suffering from congenital heart disease. In that case the sclerotic is dark blue, this being due partly to the thin condition of the membrane and partly to an accumulation of venous blood and a large mass of pigment in the eye.

In 1903 Buchanan<sup>2</sup> made the first microscopic examination and found:

The cornea and sclerotic were unusually thin, the cornea being three-fifths and the sclerotic one-third of its normal thickness. Histological examination showed that the fibers of the cornea and sclerotic were of about normal size, but unusually few in number. The anterior elastic lamina was entirely absent.

The hereditary transmission of blue sclerotics was first pointed out by Peters<sup>3</sup> in 1908. He reported cases occurring in four generations. Four of his patients also showed a typical embryontoxon.<sup>4</sup>

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1. Von Ammon: *Darstellungen der Krankheiten und Bildungsfehler des menschlichen Auges*, 1841, iii, 73.

2. Buchanan: *Tr. Ophthal. Soc. U. Kingdom*, 1903, xxiii, 267.

3. Peters: *Klin. Monatsbl. f. Augenh.*, 1908, p. 130; *Die angeborenen Fehler und Erkrankungen des Auges*, 1909, p. 80.

4. An extension of the upper layers of the sclerotic into the cornea. Pressure of the lids is said to cause accentuation at the upper and lower corneo-sclerotic margin. •

Peters also regarded the condition as due to an abnormally thin or abnormally transparent sclerotic. In 1910 Stephenson<sup>5</sup> described a series of twenty-one cases occurring in four generations of one family. In two cases the presence of an arcus senilis or an embryontoxon was noted. Subsequent investigation enabled Harman<sup>6</sup> to add another generation to this family, so that of a total of fifty-five members, thirty-one showed the same congenital peculiarity with some slight individual differences. By referring to Figure 1, it will be seen that the condition is transmitted only through affected members; it never skips a generation, and no members with white sclerotics transmit blue sclerotics to their offspring. About 50 per cent. of the children born to parents with blue sclerotics show the anomaly.

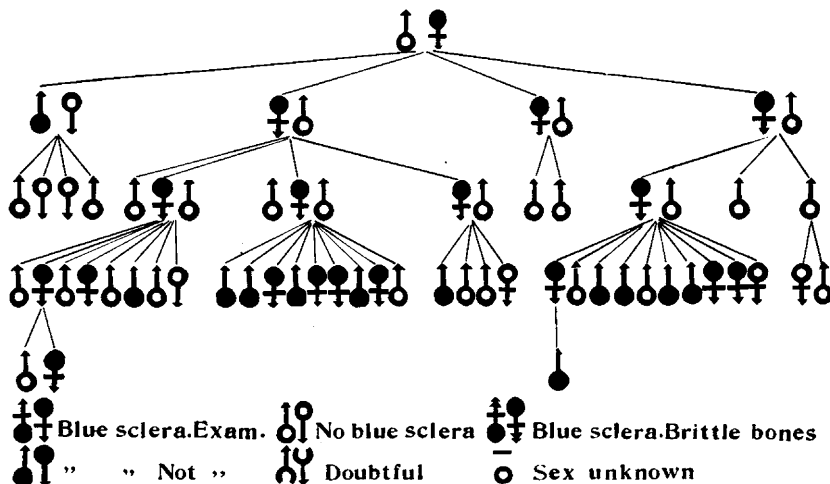


Fig. 1.—Chart of Stephenson and Harman's series of cases of blue sclerotics.

The first author to mention the association of blue sclerotics with brittle bones was Eddowes.<sup>7</sup> In 1900 he published a brief report on "Dark Sclerotics and Fragilitas Ossium," in which he described a case in a young woman whose father also had a similar condition. He mentions having seen some years previously a boy with blue sclerotics who in the course of two years during which he was under observation had nine fractures.

In 1911 Burrows<sup>8</sup> reported a series of cases occurring in four generations of one family, in which, of twenty-nine individuals,

5. Stephenson: *Ophthalmoscope*, 1910, viii, 330.

6. Harman: *Ophthalmoscope*, 1910, viii, 559.

7. Eddowes: *Brit. Med. Jour.*, 1900, ii, 202.

8. Burrows: *Brit. Med. Jour.*, 1911, ii, 16.

thirteen had blue sclerotics and nine of those fractures also (Fig. 2). Of these nine, seven had multiple fractures.

In 1912 Adair-Dighton<sup>9</sup> published a report of four generations of blue sclerotics; of thirty-two individuals, twenty-one had blue sclerotics (Fig. 3), and several of these brittle bones. During the last two years cases have also been reported by Conlon,<sup>10</sup> Cockayne<sup>11</sup> and Ostheimer.<sup>12</sup> Rolleston's<sup>13</sup> case, which was associated with congenital syphilis, probably belongs to this group.

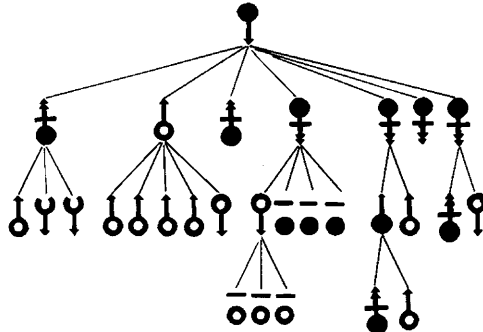


Fig. 2.—Chart of Burrows' series of cases of blue sclerotics associated with fragility of bones.

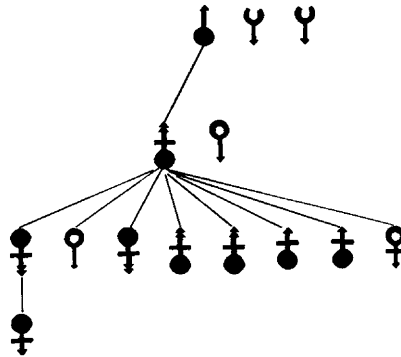


Fig. 3.—Chart of Adair-Dighton's series of cases of blue sclerotics associated with fragility of bones.

#### REPORT OF CASE

The following is a brief account of a boy of 2 years whom I have recently observed:

The patient was first seen in February, 1914, at the age of 20 months. The parents are apparently healthy, but the father is rather small and thin, weighing only 120 pounds; the mother is of average size and robust. As far as the

9. Adair-Dighton: *Ophthalmoscope*, 1912, x, 188.

10. Conlon: *Boston Med. and Surg. Jour.*, 1913, clxix, 16.

11. Cockayne: *Ophthalmoscope*, 1914, xii, 271.

12. Ostheimer, Maurice: *Fragilitas Ossium*, *Jour. Am. Med. Assn.*, Dec. 5, 1914, p. 1996.

13. Rolleston: *Brit. Jour. Child. Dis.*, 1911, p. 202.

parents know, no other members of the family have had blue sclerotics or brittle bones. There were no miscarriages; no evidence of syphilis or other constitutional disease. The parents have been married eleven years and have three children besides the patient. Two of these children I have examined and found perfectly normal. They began to walk and talk at the proper age. The patient was born normally and nursed for ten months. The first teeth appeared at 8 months, and the child began to sit up at 9 months. At 20 months (February, 1914) he was not yet able to stand or to talk distinctly. In December, 1913, he fell from an ordinary chair and fractured his right tibia, and six weeks later while in bed with the cast still applied, he sustained a fracture of the right femur. He began to walk at 23 months and now at 2½ years can say



Fig. 4.—E. G., aged 30 months; blue sclerotics associated with brittle bones.

short sentences. Physical examination on Dec. 16, 1914, aged 30 months, shows (Fig. 4) a small, thin, fair-haired boy distinctly below the normal, both physically and mentally. Height 32 inches (81 cm.), weight 22 pounds.

| Patient                         |        | Normal   |
|---------------------------------|--------|----------|
| Head—Biparietal, diameter ..... | 14 cm. | 13.5 cm. |
| Above the ears, diameter.....   | 13 cm. | 12 cm.   |
| Occipitofrontal, diameter ..... | 17 cm. | 17 cm.   |
| Circumference of head .....     | 51 cm. | 48.5 cm. |
| Circumference of chest.....     | 48 cm. | 49.7 cm. |
| Circumference of abdomen.....   | 47 cm. |          |

The head is large and brow prominent, the fontanel closed. The sclerotics are of a distinct peculiar blue color throughout. The patient has sixteen teeth. There are no signs of rickets, no rosary, no enlargement of the epiphyses. The examination of the chest is negative. There is no congenital heart dis-

ease. Radiographic examinations of the legs and arms were made. In the patient I employed a method which I have used for a number of years. The extremity of the patient was taken with that of a normal child of the same age at the same time on the same plate. In this way even those not accustomed



Fig. 5.—Right femur and tibia (*A*) of E. G., aged 18 months; taken on same plate at same time of femur and tibia (*B*) of normal infant of 18 months.

to interpret plates will have no difficulty in detecting differences in structure. It will be seen (Fig. 5) that all trace of the fracture has disappeared. Comparing the two bones, it will be noted that in length and thickness they are about the same, but one is far more transparent and lacks density. It is deli-

cate in structure especially towards the proximal end of the tibia; hardly any cortical portion is seen; the epiphyseal ends are broadened and show a distinct lack of cortical substance. Not only the bones but also the soft parts, the muscles and ligaments show the same lack of density, which indicates a diminished amount of fibrous tissue. The roentgenogram of the arm, in striking contrast to that of the leg, shows very little change from the normal (Fig. 6).

The patients affected with this combination of blue sclerotics and brittle bones are usually small and delicate, frequently fair-haired. The large size of the head is mentioned in several cases. Whether the intelligence is slightly retarded in many cases I cannot say. In my own patient his inability to run and play with other children may have had something to do with the matter. In another patient whom I saw through the courtesy of Dr. Reitzfeld, a boy of fifteen years, the intelligence was apparently normal.

The color of the sclerotic varies in different individuals from a pale azure or porcelain blue to a leaden hue. The color is uniform throughout and involves the entire sclerotic from the cornea as far as the eye

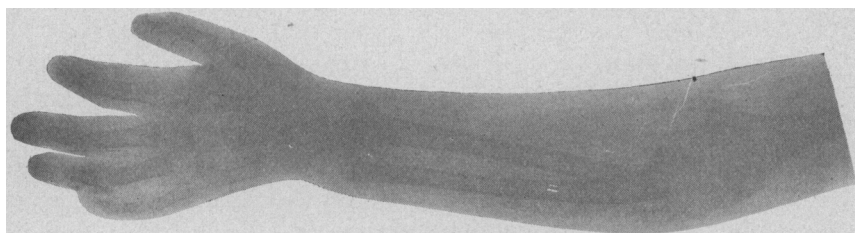


Fig. 6.—Hand and arm of E. G., aged 30 months.

ball is visible. According to Rolleston, the color is more distinct on some days. Usually there is no accentuation in the ciliary zone, but Harman mentions one case in which the color was deeper in the ciliary region and masked toward the equator on account of the increased thickness of the overlying conjunctiva, and also masked in those regions in which the muscle tendons overlay the globe. As was pointed out by Buchanan and others, the fibrous tissue is abnormally thin; the individual fibers are of normal thickness, but are deficient in number. The cornea is thin and the anterior elastic lamina is absent, which is said to account for the astigmatism frequently present in these patients. The choroid is also abnormally thin, which causes Fuchs' coloboma and the oval appearance of the optic disks (Stephenson, Harman). As to the bones, there seems to be a defect in the quality and quantity of fibrous tissue forming the framework which causes a lack of elasticity and the tendency to fracture. This change is well shown in the radiographs, and, as has been pointed out, not only the bones, but also the soft parts are also affected so that sprains are not infrequent. I

have no doubt that a careful examination will reveal changes in structure and a lack of fibrous tissue in other parts of the body. The change in the eye is so apparent that it would naturally be the first part noticed. According to Ostheimer,<sup>12</sup> of thirty-two patients with "fragilitas ossium" who were able to go about, only six were noted to have blue sclerotics, but I have no doubt that in a few other cases the peculiar color of the eye may have been overlooked. Not all patients who have blue sclerotics have brittle bones, but in the families in which blue sclerotics was an inherited peculiarity, all those members who had brittle bones had blue sclerotics also. The blue sclerotics like the fragility of the bones, may, however, occur independently of hereditary transmission. Ostheimer<sup>12</sup> states that in only eight of 117 cases of "fragilitas ossium" was the condition known to exist in either parent and in only twenty-four was it present in brothers or sisters.

## COMMENT BY DR. FRIDENBERG

Ophthalmologic examination of the little boy with blue sclerotics shows no abnormality of the eyes beyond just this peculiar condition. The color of the sclerae is uniform, as far back as it can be seen. I should call it a Delft or china blue, as it has a decidedly grayish cast. I also notice a peculiar leaden-gray color of the iris, like that seen in the newly-born, and think this may be of significance. I am not aware that it has been noted by previous observers in these cases, and will refer to its causation later on. The vision seems acute, and there is but slight ametropia. The child was very restless, and I could, at best, make out a general hyperopic refraction. The fundus was not deeply pigmented, in contrast to the case examined by Stephenson. In fact, I should call it very blond if not suggesting, at least, albinism. The disks were perfectly normal in appearance and of the usual size and shape. I might mention here that the congenital crescent downward or Fuchs' coloboma was seen in but one of these cases, and is occasionally noted in otherwise perfectly normal eyes. I do not think it has any significance for us.

As to these blue sclerotics, it was early recognized that they are due, not to any actual pigmentation, but to the unusually translucent sclera allowing the brown-black choroid to shine through, as it were, producing a color interference. That a thinning is the principal cause of this optical condition is, I think, disproved by the fact that there was in all these cases, say more than fifty, no mention of sclerectasia or of a distention of the globe in the form of infantile glaucoma or hydrophthalmos, and no ophthalmoscopic evidence in any case, of a giving way at the posterior pole (coloboma, posterior staphyloma). The condition quoted from Ammon is, as indicated in the title, some-

thing quite different, namely, a case of true infantile glaucoma or hydrophthalmus with marked thinning of the sclera, and consequent bluish translucency.

To come back to the peculiar lead-gray iris in this case, the normal color of this membrane ordinarily depends on color interference, the brown-black of the posterior or pigment layer shining through more or less as the pigment in the stroma is absent, scanty or marked. Where the pigment is entirely absent we get a light gray-blue iris which is translucent, the pink or rabbit eye of albinism. The marked transparency of both sclera and iris, in spite of an apparently normal or but slightly reduced thickness, is, I believe, due merely to absence of lime salts in the connective tissue elements of these structures. Lime salts are the deposits which as we know produce opacities in the ocular media which appear white to reflected light. Transillumination did not show any remarkable difference from the normal eye, and this again, I think, indicates that there is at least no excessive thinning of the sclera. I doubt, too, very much, whether there is in most or in any of these cases an actual absence of the anterior elastic lamina or of any other layer of the cornea, as such a condition occurring in early life would, I am sure, give rise not only to astigmatism, but to marked keratoglobus or conical cornea if not to even more decided developmental anomalies.<sup>14</sup>

The following passage in Dr. Herrman's paper is significant: "Not only the bones but the soft parts, the muscles and ligaments show the same lack of density, which indicates a deficient amount of fibrous tissue." If we lay stress on the defective quality, that is, lack of lime salts, of actual calcification and ossification, rather than on the amount of fibrous tissue, I think we shall be more nearly right. This is indicated by the roentgenograms of the fragile bones, which were found to be of about normal length and thickness. It is probable, as this passage suggests, that all connective tissue throughout the body is lacking in density, and it would be interesting to examine, among other structures, the nails, teeth and hair for further evidence of this anomaly.

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14. The correctness, or rather, the applicability of Buchanan's findings have been questioned (see Conlon).