Weill-Marchesani Syndrome:

A clinical and ultrasonographic study of two siblings

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Weill described in 1932 (1) two patients with ectopia lentis and nanism out of a group of eight patients with skeletal malformations and lens subluxation. In 1939 Marchesani (2), when studying four patients out of two families, related brachydactyly to congenital spherophakia. Nowadays those are recognised signs of Weill-Marchesani Syndrome (3).

This paper consists of two case reports, a brother and a sister, with a marked bilateral lens luxation, nasally in case 1 and temporally in case 2, its degree being infrequent at their age. A Kretz ecograph set 7200 and a 10 megahertz probe were used to measure the intra-ocular structures better, which could help us to theorise on the sequence of events that resulted in the anterior segment changes present in case 2, an uncommon finding (4). Some technical difficulties, owing to the intra-ocular changes, were met with to keep the probe exactly perpendicular to the ocular tissues. After three photographs of each eye had been taken, the echograms and the biometric measurements were analysed.

CASE REPORTS

Case 1: (Right, Picture 1) A. R. S., an 11-year-old black boy, born at Auriflama (Brazil) was referred to the Hospital by the school teacher who noticed deficient visual acuity. At a physical examination he proved to be 108 cm high, with a cephalic perimeter measuring 51.5 cm, normal muscular development, abundant subcutaneous tissue and symmetrical reduced growth. Hands and feet were short. There were marked limitations of the articular movements of the fingers, toes and wrists. The patient couldn't close his hands (Pictures 2a and 2b). X-ray examinations showed scaphocephaly, somewhat shallow orbital cavities, small zygomatic arches, a small maxilla and also a delay in the one maturation of carpus, metacarpus, tarsus and metatarsus in hands and feet. Bone age was about 5 1/2 years.

Ocular findings — The best aided visual acuity was: RE + 12.50/-1.50 x 180° 20/80 and LE + 11/-1.00 x 180° 20/40. Owing to bilateral marked lens dislocation the normal undilated pupil was almost aphakic. Bilateral microspherophakia and iridophacodonesis were present and both lenses were luxated nasally (Pictures 3a and 3b). Temporally the zonular fibres were torn and the anterior hyaloid membrane was pushed forward. In both eyes a very slight persistence of mesodermal tissue in the angle could be seen at a gonioscopic examination, with a nasally prominent and anteriorly dislocated Schwalbe's line. Corneal diameter was 11 mm and ophthalmoscopic findings were normal OU. IOP was 14 mmHg in the RE and in the LE, 16 mmHg.

Table 1 shows the ocular biometric results (See also Picture 4).

Case 2: (Left. Picture 1) E. R. S., a 10-year-old black girl, born at Auriflama (Brazil) was referred to the Hospital for the same reason as her brother. At a physical examination she proved to be 101 cm tall with a cephalic perimeter measuring 53.5 cm. Other findings were very similar to case 1, including X-ray examinations (Pictures 5a and 5b), and bone age was about 6 1/2 years.

Ocular findings — The best aided visual acuity in the RE was 20/40 (+ 11.50 DS) and in the LE 20/60 (+ 12.00 DS). Bilateral microspherophakia was present, associated with a marked temporal superior lens luxation and iridophacodonesis. Zonular fibres were torn nasally and the anterior hyaloid membrane was pushed forward. There was a microbullous oedema in the regions of the right cornea where extensive peripheral anterior synechiae were present, namely from 6 to 12 hours, from 0.30 to 2.30 hours and from 4 to 5 hours (Picture 6). The same ocurred in the left cornea in the peripheral region between 12 and 2 hours. No pigmentary dispersion or atrophy of the iris were seen in etither eye. Gonioscopically the iris in both eyes appeared adhered to the posterior surface of the cornea in the regions where it was oedematous; a very slight amount of residual mesodermal tissue was present, with no posterior embryotoxon. The traction in the anteriorly synechiae iris produced a localised iridoschisis. The corneal diameter in both eyes was 11.5 mm, the fundus normal, and the IOP was 14 mmHg in the RE and 16 mm Hg in the LE.

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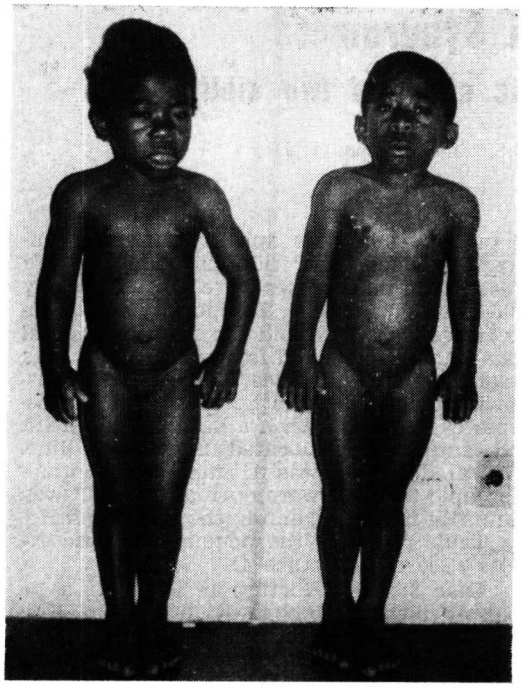


Picture 1 — Photo of the naked siblings. The numbers in the ruler cannot be seen clearly, but E. R. S. (Case 2), on the left side, is 101 cm tall and her brother A. R. S. (Case 1), on the right side, is 108 cm tall.

Table 1 shows the ocular biometric results (See also Picture 7).

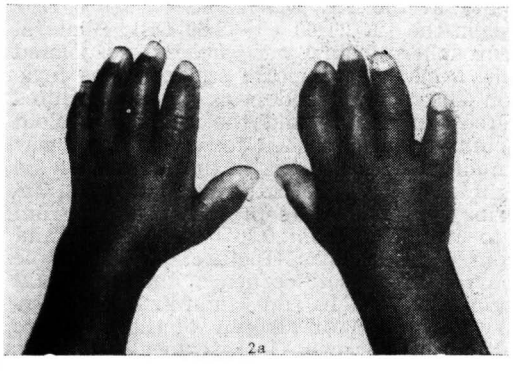
TABLE 1

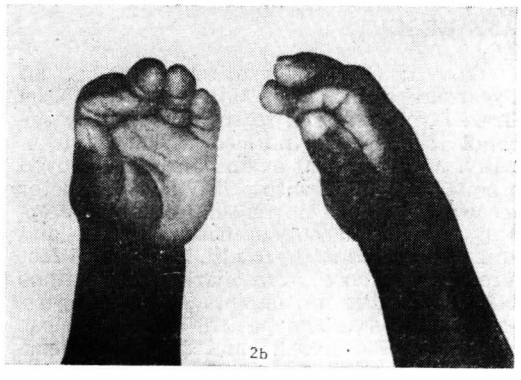
Biometric measurements in both eyes taken n both patients (cases 1 and 2) with a Kretz ecographic set 7.200 and a 10 megahertz probe. The antero-posterior diameter of the eye is the result of adding up the anterior chamber depth, the lens thickness and the vitreous space diameter measurements. The values are in millimetres.

Case	Biometric Measures	Anterior Chamber	Lens	Vitreous	Antero-posterior diameter of the eye
1	RE	3.22	5.49	13.71	22.88
	LE	3.98	5.49	12.95	22.81
2	RE	2.45	5.41	15.47	23.79
	LE	1.92	5.41	16.09	23.87

Family History

The parents come from different parts of the country and have no blood relation. The father's paternal grandmother is reported to have had severe visual impairment from childhood but no reduced growth was noticed. The father, examined by us, has no ocular or skeletal changes and is 148 cm tall with a cephalic perimeter of 58 cm. One of his adult sisters is 130 cm tall and another, who has dental malformations, is 140 cm tall and cannot close her hands. The children's mother is 160 cm tall and no skeletal or ocular changes are reported in her family.

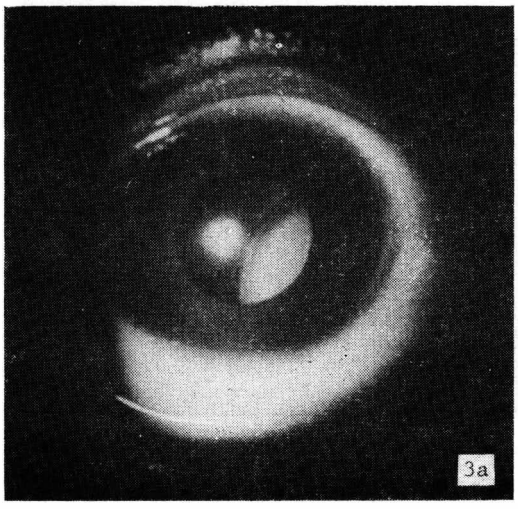


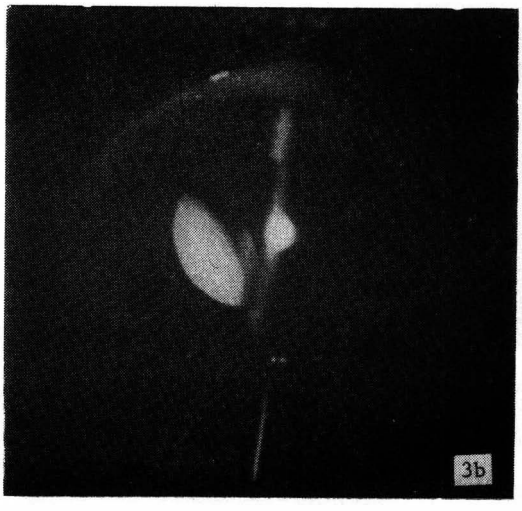


Picture 2 — (a) A. R. S. (Case 1) Hands showing brachdactylia and somewhat fusiform fingers. (b) This photo shows A R. S. (Case 1) trying unsuccessfully to close his hand.

DISCUSSION

In the Weill-Marchesani Syndrome skeletal changes are typically brachymorphic (5, 6, 7, 8, 9, 10, 11) and the affected patients have a reduced growth, brachycephalia and brachydactylia with limitation of the articular movements of the fingers, toes and wrists. The musculature is generally well developed with abundant subcutaneous tissue. Ocular changes (5, 6, 7, 8, 9, 10, 11) include microspherophakia, ectopia lentis, iridophacodonesis, lenticular myopia and, sometimes, secondary glaucoma, normally of very diffi-





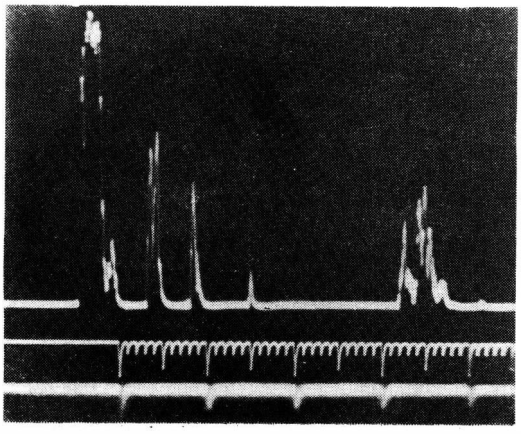
Picture 3 — (a) RE, A. R. S. (Case 1). A nasal lens displacement and microspherophakia are visible under cycloplegia. (b) LE, A. R. S. (Case 1). Same as in picture 3a.

cult management (6, 12). This syndrome is relatively rare (6).

The patients reported here show a typical symmetrical shortening and widening of the metacarpus, metatarsus and phalanges in both hand and feet. The delay of carpal ossification differs from the one normally found in mucopolysaccharidosis in that it does not show any narrowing of the distal one-third of the metacarpal bones.

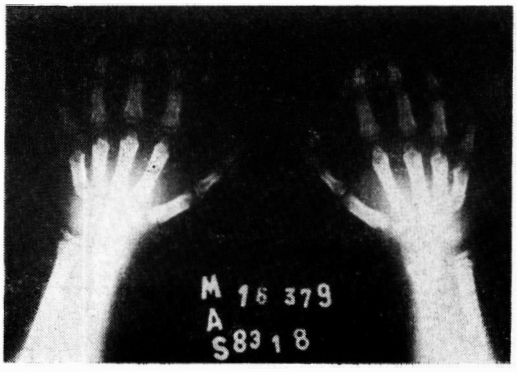
As the anomalies seen in this syndrome may occur in others, and as there is no known metabolic defect present, the diagnosis has to be made when those changes are typical and occur together (13, 14). Microspherophakia associated with ectopia lentis can be present in Marfan Syndrome and even in homocystinuria.

Rosenthal and Kloepfer (15) demonstrated that a variable expression of the basic diagnostic components may occur in this syndrome. Jenson and Cross (6) believe that it is definitely familial and MacKusick (16) lists it as autossomal recessive. In our patients the study of the heredogram was not very useful, as only the probands and their father could be examined.

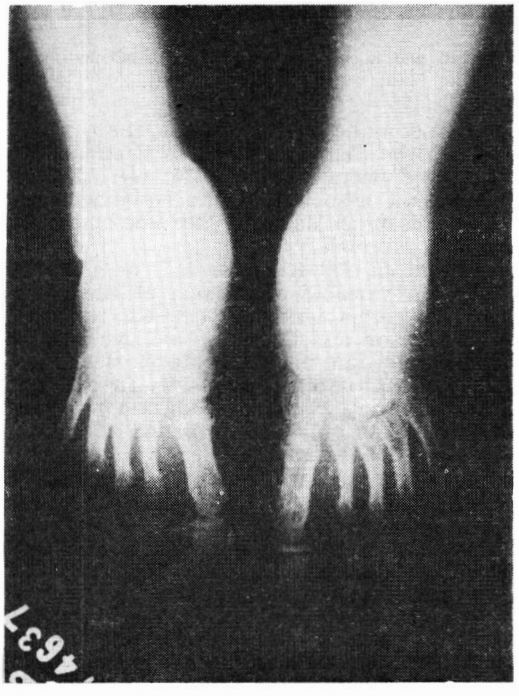


Picture 4 — Echogram of the RE, patient A. R. S. (Case 1), taken under cycloplegia at 60 decibels, with a Kretz 7.200 ecographic set and a 10 megahertz probe. Picture 5 (a) — X-ray photo of both hands, patient E. R. S. (Case 2). A symmetrical shortening and widening of the metacarpal bones and phalanges is present as well as a delay of the carpal ossification.

In both patients the ultrasonographic studies revealed a longer than normal lens antero-posterior diameter (17, 18) owing to the lens sphericity and a normal antero-posterior diameter of the globes. Bilaterally the depth of the anterior chamber in case 2 was shorter than normal (19, 20, 21), probably owing to the lens anterior displacement. as the vitreous space diameters of her eyes were longer than the corresponding diameters found in her brother's eyes. This displacement has probably made possible a contact between the iris and the corneal posterior surface, which led to a tractional iridoschisis and a corneal decompensation, manifested here as a microbullous oedema. The hypothesis of an inflammatory process to explain those adherences has no biomicroscopic support. There are reports (4, 22) on chamber angle anomalies in this syndrome. Therefore those findings could represent an uncaractheristic mesodermal dysgenesis, but no change in the iris structure was seen, ex-



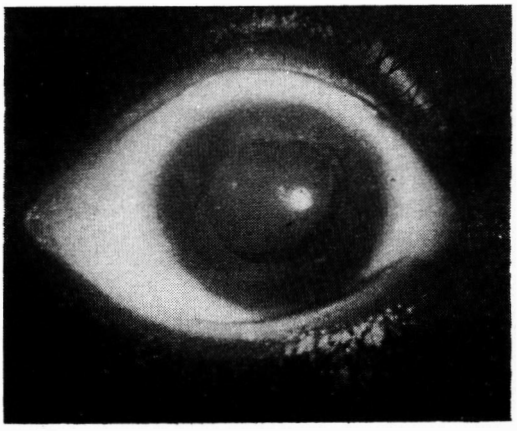
Picture 5 (a) — X-ray photo of both hands, patient E. R. S. (Case 2). A symmetrical shortening and widening of the metacarpal bones and phalanges is present as well as a delay of the carpal ossification.



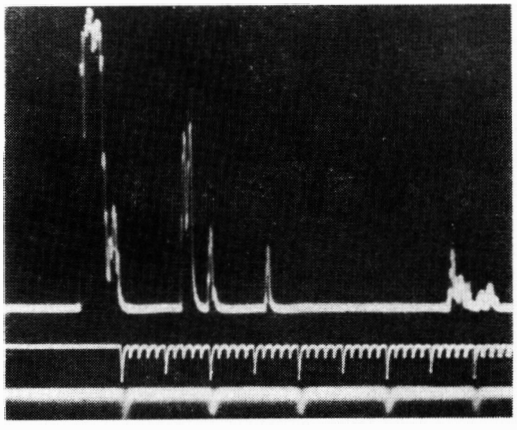
Picture 5 (b) — X-ray photo of feet, patient E. R. S. (Case 2), showing a delay in the metatarsus and tarsus bone maturation.

cept for tractional iridoschisis. Here there are neither bundles of iris tissue adhered to the cornea as in Axenfield and in Rieger Syndromes nor the changes in the posterior surface of the cornea seen in Peter's Syndrome.

Either an anterior dislocation of the lens or an uncaractheristic mesodermal dysgenesis could be the answer to the findings in case 2. Further studies are required to establish the link between mesodermal disorders and intraocular changes seen in Weill-Marchesani Syndrome.



Picture 6 — RE, E. R. S. (Case 2). Under cycloplegia a temporal lens displacement and microspherophakia are visible. Anterior synechiaes are also present (see text).



Picture 7 — Echogram of the LE, patient E. R. S. (Case 2), taken under cycloplegia at 52 decibels, with a Kretz 7.200 echographic set and a 10 megahertz probe.

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SUMMARY

A clinical and ultrasonographic study of two siblings of different sex with Weill-Marchesani Syndrome is presented. The sister shows a change in the anterior segment of either eye, probably owing to either a confirmed ultrasonographic anterior displacement of the lens, with an increase of the antero-posterior vitreous space diameter and shortening of the anterior chamber depth, or an uncharacteristic mesodermal dysgenesis of the anterior chamber.

RESUMO

Os autores apresentam um estudo clínico e ecográfico de um irmão e sua irmã ambos portadores da

síndrome de Weill-Marchesani. A irmã apresenta uma alteração do segmento anterior em ambos os olhos, provavelmente devida seja a uma anteriorização-confirmada ecograficamente — do cristalino com consequente aumento do diâmetro ântero-posterior do espaço vítreo e diminuição da profundidade da câmara anterior, seja a uma disgenesia mesodermal não característica do segmento anterior.

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