IDIOPATHIC INFANTILE SCOLIOSIS AND HYPOTROPHY

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Between 1950 and 1974, 155 cases of infantile scoliosis were examined and treated at the Centre des Massues. The first investigations concerned:

(a) children in the evolutive period of their scoliosis, before puberty or before 'osseous' maturity, and

(b) adults after the end of the evolutive period, but whose deformity slowly increased with the passing years.

Amongst the 155 cases noted (consisting of children, adolescents and adults), the abnormal leanness of several patients was noted with surprise.

The selection consisted of infantile scoliotic patients whose weights showed a large variation from an average sample - that is, more than two standard deviations from the mean. In this way, there were 19 cases out of the 155 studied, which is statistically significant i.e. 12% instead of the 2.5% expected from a normal sample. Two examples are given:

A.C. Female. Scoliosis first noted when 3 years old. First seen when 10½ years old.
The site of the scoliosis was between T2 and T6, and T6 and L2 = 44° and 62° of lateral curvature.

Height = 1.25 m (predicted height 1.60 m)
Weight = 38 kg (predicted weight 55 kg)
Female. Scoliosis first noted when 2 years old. First seen when 9 years old. The site of the scoliosis was between T7 and L1 = 90° Height = 1.25 m (predicted height 1.27 m) Weight = 18 kg (predicted weight 27 kg)

Hypotrophic Infantile Scoliosis first seen in Adulthood

It would be helpful to know if hypotrophy is related to the occurrence of scoliosis or kyphoscoliosis. In some cases, there does seem to be a link.

Female. Scoliosis first noted when 2 years old. Discontinuous treatment with plaster casts, braces etc. First examination - 21 years The site of the kyphoscoliosis was between T4 and T12 = 137° Height = 1.35 m Weight 32.5 kg (predicted weight 55 kg) Several reductions with plaster casts, Harrington procedure and fusion were done. The lateral curvature was between T4 and T12 = 99° The height benefit was 8 cm The weight benefit was 9 kg

In this case it appears that hypotrophy is related to the severity of kyphoscoliosis and the partial reduction gives the patient a better physical condition. On the other hand, we have also found that in more severe kyphoscoliosis, hypotrophy does not occur.

Female. Scoliosis first noted when 1 year old. Treatment - spinal bracing until 12 years First examination - when spinal curvature measured 45° The site of the kyphoscoliosis was between T6 and L1 = 144°, convexity to the right Height = 1.63 m Weight = 85 kg (when 35 years old, weight 70 kg)
Another case shows that significant reduction of a severe spinal curve was not always followed by an increase in body weight.

R.C. Female. Scoliosis first noted at 2 years old.
The patient had always been thin. In spite of orthopaedic treatment, scoliosis worsened. When aged 19 years, the lateral measurement of the right scoliosis was between T4 and T10 = 162°

Height = 1.32 m
Weight = 25.5 kg
Partial reduction and fusion was carried out with Harrington device; the curvature between T4 and T10 of 120° was stabilised at 125°.

16 years later, when 35 years old -
Height = 1.35 m
Weight = 33 kg (predicted weight 55 kg)

In some patients, therefore, it appears that there is no relationship between hypotrophy and infantile scoliosis.

Another study concerns 23 cases of infantile scoliosis which were seen and treated for scoliosis or kyphoscoliosis when adult. It appears that two patterns of trophicity can be clearly defined. From the histogram of weight distribution of patients, there are two findings (Fig. 1):

Fig. 1 23 patients with infantile scoliosis above 100° are seen when adults; 10 are hypotrophic.
One concerns patients with a subnormal trophicity and
a weight between 43 and 60 kg (predicted weight 55 kg),
with a mean spinal curvature of 138°, while the other
concerns rather hypotrophic patients with a weight be­
tween 30 and 35 kg, and mean spinal curve of 141°.

It seems that there are two different patterns of
infantile scoliosis: severe hypotrophy or quite normal
 trophicity. The difference in the spinal curve between
the two groups is not significant, about 9° only.

Evolution of Infantile Scoliosis and
Hypotrophy in Infancy

In the second group, there seems to be a parallel
between the evolution of scoliosis and evolution of hypo­
rophy.

J.R. (figs. 2a,b,c,d,e,f)
Female. Scoliosis first noted at 4 years old.
The site of the scoliosis was between
T 3 and T11 = 39°, convexity to the right.
Photographs show normal trophicity.
The curvature remained untreated from 4 to 10 years
old.
Scoliosis between T3 and T11 increased to 82°
Height=1.18 m (predicted height 1.35 m)
Weight=19 kg (predicted weight 30 kg)

This small height is partly the consequence of the spinal
curve. However, the similarity mentioned at first does
not hold. After that, the spinal curve was reduced to
59° by distraction (Harrington procedure and fusion).
The patient's progress was followed until she was 25
years old. The weight is still poor at 36 kg (predicted
weight 55 kg).

Diagnosis

It appears that there is a factor of leanness in
some infantile scoliosis. This 'leanness factor'
suggests a dysplasia. During the evolution of some
hypotrophic infantile scoliosis, it proved possible to
Fig. 2a. Evolution of an infantile idiopathic scoliosis. Back view age 4 years.

Fig. 2b. Front X-ray age 4 years. T3 to T11 = 390
Fig. 2e Back view (after surgery) age 25 years.
Fig. 2f Front X-ray (after surgery) age 25 years. T3 to T11 = 5º.
include them in well-known syndromes rather than to retain the label 'idiopathic'. Here are two examples—one concerns an example of Marfan's disease, the other of Ehlers-Danlos syndrome.

In the first years of life, these syndromes are not always obvious. Usually, such a diagnosis is easier to confirm in adolescence.

**S.P. Marfan's syndrome.**
Male. Scoliosis first noted at 1 year old.
The site of the scoliosis was between T4 and T12 = 80°, lateral curvature at 5 yrs.
Treatment - Milwaukee brace.
At 12 yrs 7 mths the patient was very thin.
Harrington procedure at 13 yrs 2 months.
At 14 yrs 3 mths, the boy is still equally thin.
Weight 23.5 kg (predicted weight 50 kg).

It is possible also to confirm arachnodactyly, hyperlaxity, pigeon breast, myopia, and mitral insufficiency.

(This case is not included among the 19 patients with idiopathic scoliosis that we have studied)

**M.C.** Male. First seen at 18 days old. Normal birth weight 3.34 kg. Deformation of hands and feet. Scoliosis first appeared at age 8 mths.
Contact with him was lost until age 12 yrs 10 mths.
He now had a very severe left kyphoscoliosis between T4 and L1 = 172° lateral curvature.
There was marked hypotrophy.
Weight = 22 kg (predicted weight 38 kg)

Joint hyperlaxity, skin laxity, furrowed aspect of palmar teguments, these enable us to confirm Ehlers-Danlos syndrome.

Thinness is often associated with Ehlers-Danlos syndrome. Therefore, in this case, there is an explanation for the hypotrophy occurring with infantile scoliosis.
Hypotrophic Infantile Scoliosis

For the 19 cases selected for this study, no well-recognised syndrome is involved. Before an assertion can be made that it is a dysplasia that is responsible for the hypotrophy, the 19 cases must be examined to be sure there are no other factors which could explain the phenomena, such as a long period of hospitalisation, many plaster casts, 'dysmaturation', cardiopathy and respiratory deficit.

Other Factors of Hypotrophy

Hypotrophy and Plaster Casts

P.T. Female. Scoliosis first noted at 8 mths old.
The baby walked normally at 10 mths, but the patient has undergone treatment in several clinics from the age of 1 to 17 yrs.
First personal examination at 17 yrs.
Left kyphoscoliosis between
T5 and T12 = 155° of lateral curvature.
Weight = 35 kg (predicted weight 54 kg)

(Long periods of hospitalisation and treatment with plaster casts or orthopaedic braces may cause hypotrophy in non-scoliotic patients).

Hypotrophy and Dysmaturation

R.C. Female. Scoliosis first noted at 2 yrs old.
She was hypotrophic during her childhood, adolescence and her adulthood.
At 37 yrs old she weighed 33 kg (predicted weight 55 kg)
She has a twin, her birth weight being 0.9 kg.

Fetal dysmaturation is a traumatic start to life and may account for hypotrophic dysplasia.
Hypotrophy and Cardiopathy

M.D. Female. Double-major scoliosis first noted at 2 yrs old.
Premature (7½ mths)
Weight = 1.35 kg. Cardiopathy (interauricular communication)
At 4 years of age, scoliosis between T11 and L3 = 56° of lateral curvature
Weight = 12 kg (predicted weight 15 kg)
Her cardiopathy did not make her orthopaedic treatment easy.
Her weight when 21 yrs old was 35 kg (predicted weight 55kg)
The spontaneous evolution of her scoliosis gave a kyphoscoliosis between T5 and T10 = 153° lateral curvature.

Surgical treatment was refused by the parents. This girl died at the age of 22 yrs 7 mths.
Cardiopathy and respiratory failure seem to predominate in this case. At 21 yrs her vital capacity was 400 ml However, photographs taken at the age of 21 yrs show us an unusual morphology of the upper and lower limbs for congenital cardiopathy.

Hypotrophy and Respiratory Deficit

Since scoliosis may be associated with severe respiratory failure, it is considered whether the respiratory deficit may be related to the amount of hypotrophy.

B.W. Female. Scoliosis first noted at 10 mths.
First personal examination when 26 yrs old.
Site of scoliosis between T4 and T12 = 179° of lateral curvature.
Height = 1.36 m
Weight = 32 kg
Vital capacity = 910 ml.

But none of these examples seem sufficient to explain a permanent respiratory deficit, such as those referred to:
For example -

**M.M.** Male. Scoliosis first noted at 2 yrs.
first examined at 37 yrs.
Site of the scoliosis between
T6 and L1 = 168° of lateral curvature.

Weight = 62 kg
Vital capacity = 1,000 ml.
This patient was not at all underweight.

**Hypotrophic Dysplasia**

Three cases are given as examples

**R.B.** (Figs. 3a,b,c,d)
Male. Scoliosis first noted at 7 mths.
Interrupted treatment. Milwaukee brace and
Harrington procedure with fusion at 12 yrs.
First examined at 15 yrs 3 mths.
Site of the kyphoscoliosis between
T4 and T12 = 160° of lateral curvature -
convexity to the right.
Evident hypotrophy = 21 kg.
Bone age - delayed by 5 yrs.
Arachnodactyly. Thumb and wrist signs. No
skin hyperlaxity but abnormality of the elbow
skin and webbed fingers. Myopia, but the
lens is not Marfan-like. Moreover, the height
is very short for a Marfan's syndrome: 1.30 m.

After a new Harrington procedure and fusion, the curve
was reduced but growth did not continue beyond 1.50 m.
The patient remains extremely hypotrophic - weight
25 kg at 18 yrs.

**V.B.** Female. Scoliosis first noted at 1 yr 6 mths.
No treatment until our first examination at
16 yrs 8 mths.
Site of the kyphoscoliosis between
T6 and L3 = 186° of lateral curvature.
Weight 21 kg.
Height 1.25 m.
Bone age - 5 yrs delayed.
Vital capacity = 610 ml.
Arachnodactyly: thumb sign 20 mm overcrossing wrist sign 20 mm
Joint hyperlaxity: left hip subluxation.
The illustrations show the extreme thinness of the body.
Halo distraction, Harrington procedure and fusion: T6 to L3 = 87° of lateral curvature.
Height increase 22 cm.
Even with this reduction, the height was only 1.47 m.

G.C. (Figs. 4a,b,c,d)
Male. Scoliosis noted at 3 mths.
First personal examination at 12 yrs.
Site of the scoliosis was between T5 and T12 = 144°, convexity to the left.
Very hypotrophic, weight = 19 kg (predicted weight 36 kg).
Short stature = 1.36 m.
Vital capacity = 310 ml.
Asymmetrical chest, depth: 22 mm, difference in level of the chondral part of the ribs.
Bone age - 2 yrs delayed.
Unstuck ears.
Teeth are set very irregularly.
Skin is fragile which gives trouble during treatment due to pressure sores.
Even with such problems the child underwent Harrington procedure and fusion without difficulty.
He is now 28 yrs old.
Spinal curve = 100° of lateral curvature.
Height now = 1.50 m.
Weight now = 29 kg.
Hypotrophic Dysplasia

One must look for several symptoms which may enable identification of this hypotrophic dysplasia.

A permanent weight deficit is the first obvious sign. This, to be typical, should be more than two standard deviations from the mean.

Arachnodactyly: wrist sign and Parker sign were evident in three cases.

Joint hyperlaxity is a usual finding. Velis (1) studied the incidence of this condition in idiopathic scoliotic patients and he found an increase in the percentage and the range of joint laxity in comparison with an average sample.

Wrist and elbow hyperextension and genus recurvatum are sometimes found. (It was evident in 4 of our cases). Fragility and thinness of the skin is often seen. It may also be translucent. The subcutaneous tissue has almost disappeared. The venous network is easy to see, especially on the chest.

The same features are found in Ehlers-Danlos syndrome.

In 4 cases the fragility of the skin caused numerous troubles during treatment.

Skin palmarures may be present, as in Marfan's disease (Fig. 3d). These changes may be so striking that one may think of arthrogryposis; one of the patients studied had a palmarure of the elbow. There are no cases in the study with Status Dysraphicus.

Thoracic deformities: pectus excavatus occurred in 4 cases (Fig. 4c), asymmetrical pectus carinatus in 1 case.

Facial deformities: triangular shape: teeth malposition (Fig. 4d) were both found.

Genetic: 4 cases have a family history of the deformity.

CONCLUSION

Our approach has been more clinical than scientific. However, it seems evident to us that some cases of scoliosis, mainly infantile, show a severe hypotrophy.
Fig. 3a R.B. Infantile idiopathic scoliosis with dysplasia. when 15 yrs 3 mths T4 to T12 = 145°. Back view.

Fig. 3b. Lateral view.
Fig. 4a  C.C. Infantile idiopathic scoliosis. When 12 yrs old T5 to T12 = 169°.
Fig. 4b  Oblique X-ray.
Fig. 4c  Asymmetrical chest.
Fig. 4d  Teeth malposition.
Among the 19 cases above, some have dysplastic signs, some have only permanent hypotrophy even after partial reduction.

This hypertrophy can remain even after a significant reduction of the spinal curve. Thinness, giving a special appearance, suggests a metabolic disease. Up to now, we have no evidence for this disturbance but biochemical research in this direction is not possible by us. The knowledge of hypotrophic patterns is useful. Their treatment is insecure. Plaster casts, braces (Lyonese or Milwaukee) are barely tolerated. Their structural kyphoscoliosis is rigid and poorly reducible. The bone fragility and the stiffness of the tissues make the operative procedure more difficult.

A better knowledge of these patients with hypotrophic infantile scoliosis suggests the existence of an hypotrophic dysplasia X, which may be both the cause and consequence of the spinal deformity. While the cause (dysplasia) still exists, hypotrophy remains even after partial reduction of the curve. When its consequence (kyphoscoliosis) increases, with a decrease in vital capacity, the hypotrophy gets worse.

It is hoped to have a better knowledge in the future of the metabolic failure leading to such an hypotrophic scoliosis. It would be helpful to be able to differentiate between idiopathic scoliosis and scoliosis linked to histodysplasia and chemodysplasia, such as Type VI of Ehlers-Danlos syndrome, described by Pinnel and Krane (2), related to an hydroxylysine deficit, and perhaps caused by a lack of lysylhydroxylase.

Marfanoid hypermobility (3) has some symptoms that may be found in Marfan's disease or Ehlers-Danlos drome. This condition could lead to another way of gaining a better knowledge of hypertrophic scoliosis.

In the same way, hereditary bone dysplasia with kyphoscoliosis has been described by Epstein and Graham (4).

Studying the fibroblasts of idiopathic scoliotic patients, Connen (5) pointed out metachromasia. Previously, Matalon and Dorfman (6) described the same anomaly, which is evident in Marfan's disease. This
metachromasia, although non-specific, suggests some histochemical relationship between the two diseases.

REFERENCES


