OSTEOGENESIS IMPERFECTA

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The following is a report of three cases.

CASE I (Figures 1-8)

This Chinese boy, L.C.S., aged 3 years was admitted with a history (given by the father) of having sustained frequent fractures in all four limbs during the past two years. In all, he sustained more than 20 fractures, each of which resulted from often trivial injuries. The following are examples:

(a) A fall, when the patient tripped while walking.

(b) Dressing or undressing against the wishes of the patient.

(c) While playing with other brothers and sisters.

For the first few fractures the patient was treated by Chinese bone setters, but the father having learnt the art from them, treated the patient himself subsequently.

All the fractures occurred in either the femurs or the humerus and they healed rapidly. The child did not appear to have suffered unduly from the fractures as they did not cause pain.

Milestones: A normal delivery, the child weighed 5½ lbs. was able to sit up and talk at about the age of 12 months and walk at the age of 18 months.

Fig. 1. Case I. Age 3 with deformity of left upper arm. Patient was treated with "gallows" traction for fracture of femoral shaft.

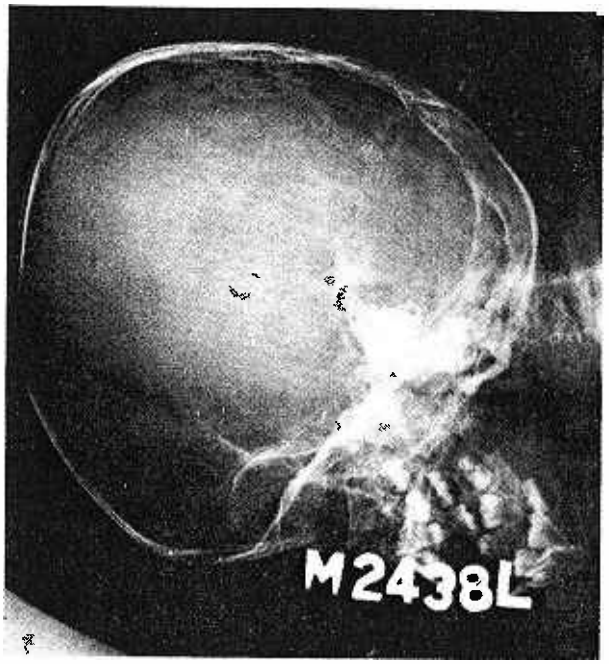


Fig. 2.

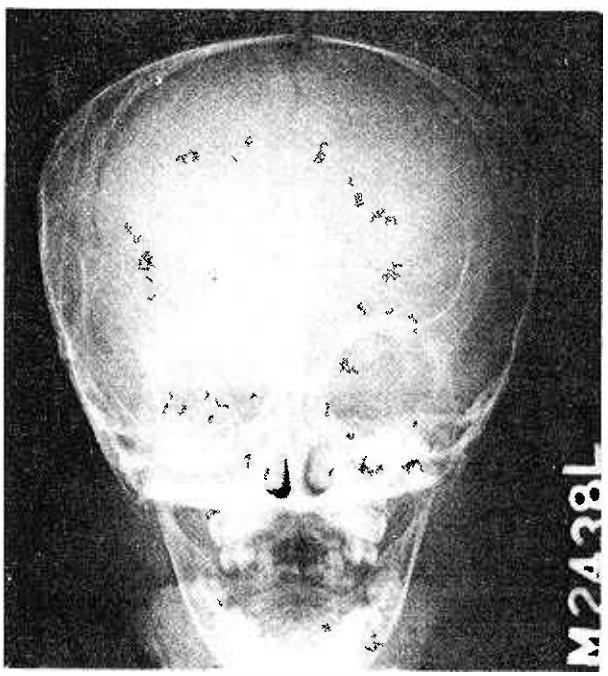
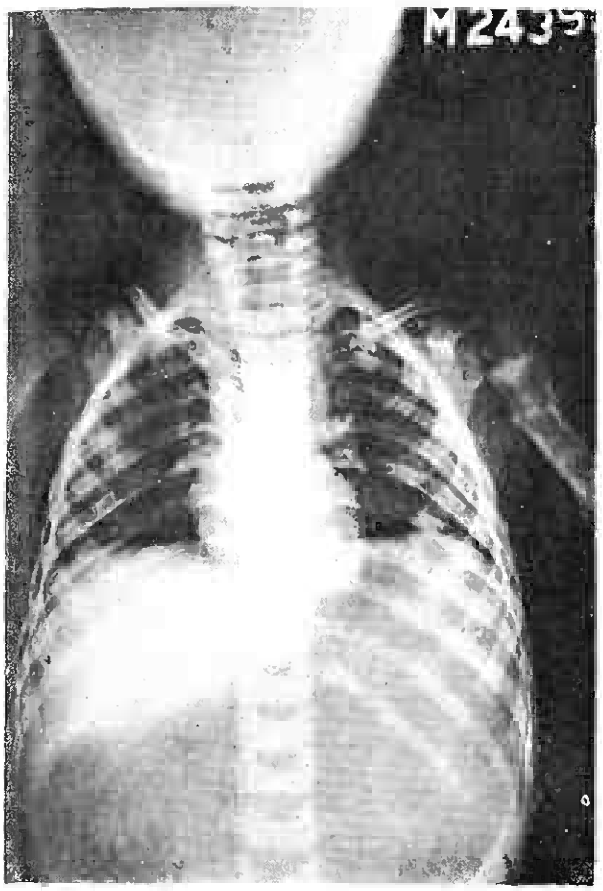


Fig. 3.

Figs. 2 and 3. Case I. Age 3 with skull showing prominence of parietal and occipital bobes.

Family History: The patient was the 5th in the family of 7 children and they were all well except that the 6th child also had occassional fractures from trivial injuries (Case II).



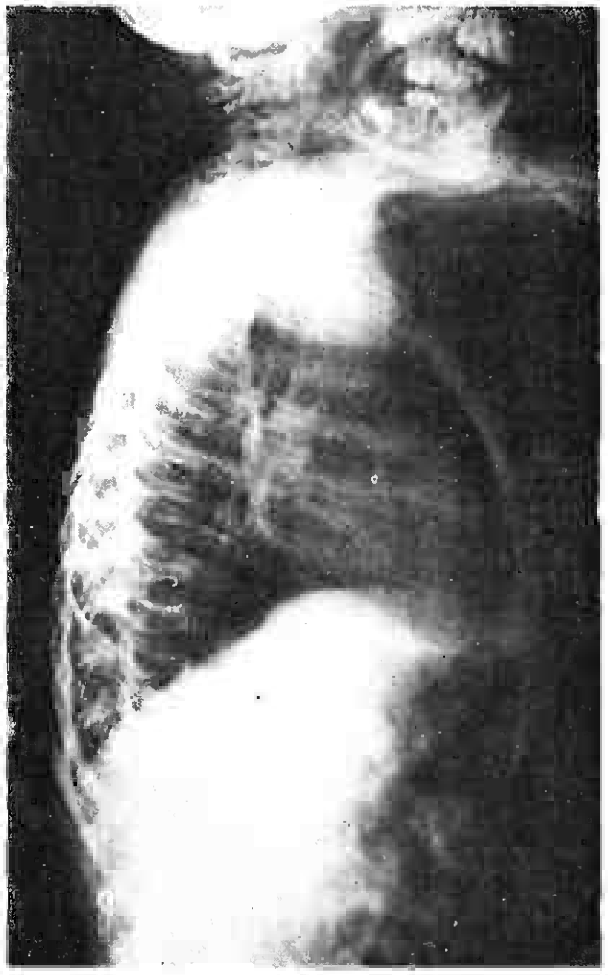
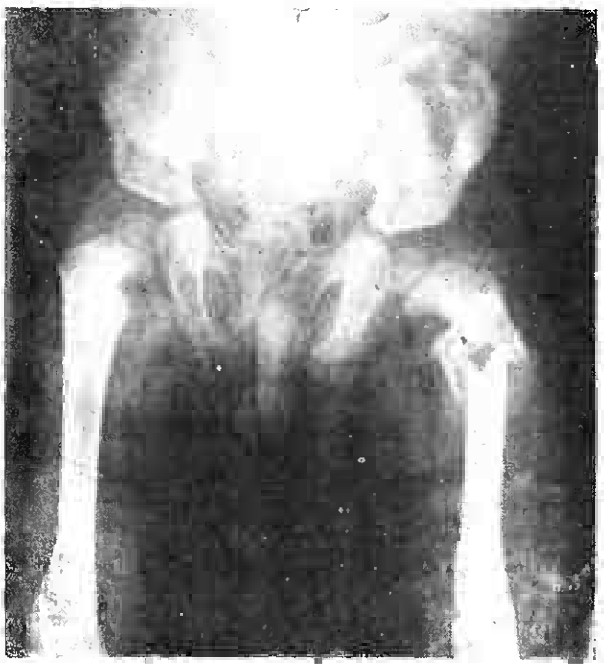


Fig. 4. Fig. 5.

Figs. 4 and 5. Case I. Age 3 with vertebral bodies showing osteoporosis and biconcavity and ribs showing multiple fractures.



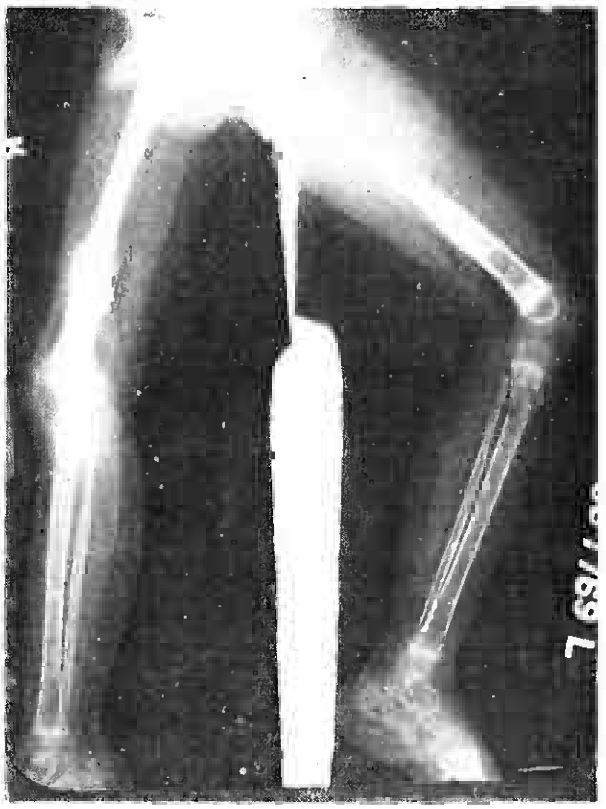


Fig. 6. Fig. 7.

Figs. 6 and 7. Case I. Age 3 with slender lower limb long bones showing general osteoporosis, thin cortex and multiple fractures.

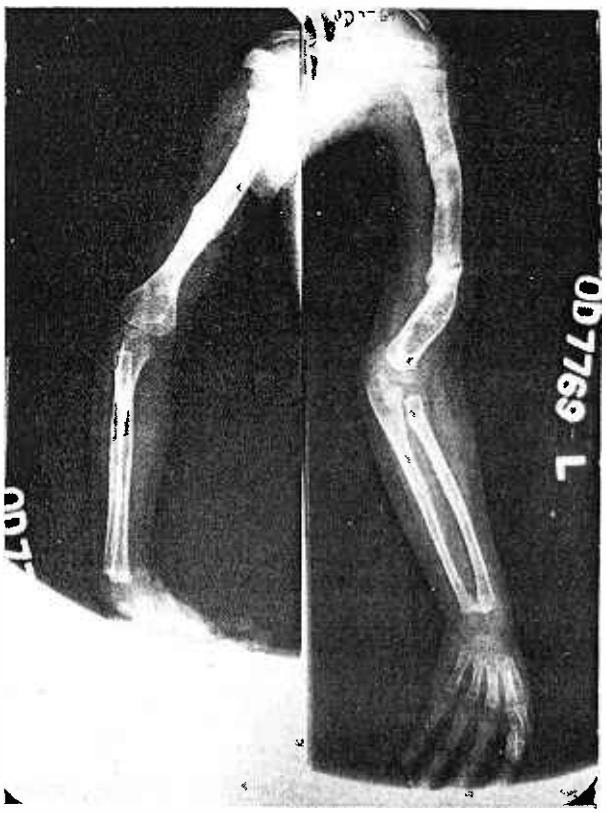


Fig. 8. Case I. Age 3 with upper limb long bone showing general osteoporosis, slender shafts, thin coxtex and multiple fractures of both humerus.

On Examination: Child was small for his age (Fig. I) was pale and irritable. He resisted examination. There was bossing of both parietal regions and there was a bluish tinge of the sclera. He had bad teeth. There was no clinical abnormality of the spine. There were fusiform swellings and deformities of both upper arms, but neither of these were tender; the forearms were normal. There was also a swelling over the left thigh. which was tender on applying pressure.

The lungs and heart were clinically normal except for some rhonchi over the lungs. The Central Nervous System was normal. There was no undue laxity of his joints.

There was bossing of the parietal bones bilaterally and biconcavity of many of the vertebral bodies. The long bones were slender with general osteoporosis and thin characteristic cortex. There were fractures of several ribs, and of both humeral and femural shafts.

Other Investigations:

Blood counts:

Total red count 4.17 million per cu. mm.
Total white count 11,200 per cu. mm.
Differential count: Polymorphs 65%, Lymphocytes 28%, Monocytes 3%, Eosinophils 4%.

Blood chemistry:

Serum calcium	11.8 mgm%
Serum phosphorus	2.4 mgm%
Blood alkaline phosphatase	14 units

CASE II

This Chinese girl, L.C.H. aged 2 years the younger sister of Case I, sustained a fracture of her left femur while the mother was dressing her against her will. Her mother said that she did not use any undue force.

The patient had 3 previous fractures, all of which resulted from trivial injuries. None of the fractures caused severe pain.

Milestones: A normal delivery, she weighed 5½ lbs, was able to sit up at 12 months and speak at 18 months.

Family History: Younger sister of Case I. The 6th child in the family.

On Examination: There was no clinical abnormality detected except the evidence of "callus" in the left upper arm and swelling, deformity and tenderness in the left thigh. The child was normal in size for her age and there was no bluish tinge in her sclera. The only other significant feature was that she had bad teeth.

X-rays: There was thinning of the cortex of the long bones generally, and some biconcave spinal vertebral bodies. There were fractures of both the left femoral and humeral shafts.

Other Investigations:

Blood count:

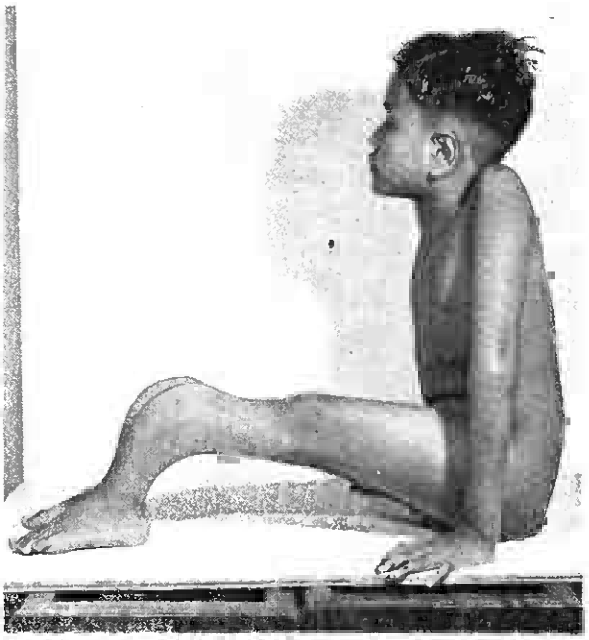
Total red count 3.82 million per cu. mm. Hb. 78%.
Total white count 7,800 per cu. mm.
Differential count: Polymorphs 65%, lymphocytes 32%, monocytes 1%, eosinophils 2%.

Blood chemistry:

Serum calcium	12.4 mgm%
Serum alkaline phosphatase	20 units.

CASE III (Figures 9-12)

This Malay boy B.B.A., aged 12 years was admitted with a history of having blue sclerotics since birth and deformity of his legs. At the age of 14 months, when he began to stand, it was noticed that his legs began to bend. At the same time he began to sustain fractures from



Figs. 9 and 10. Case III. Age 12 with marked anteriorly curved deformity of both tibias. The feet were everted due to medial curvature of the tibias.

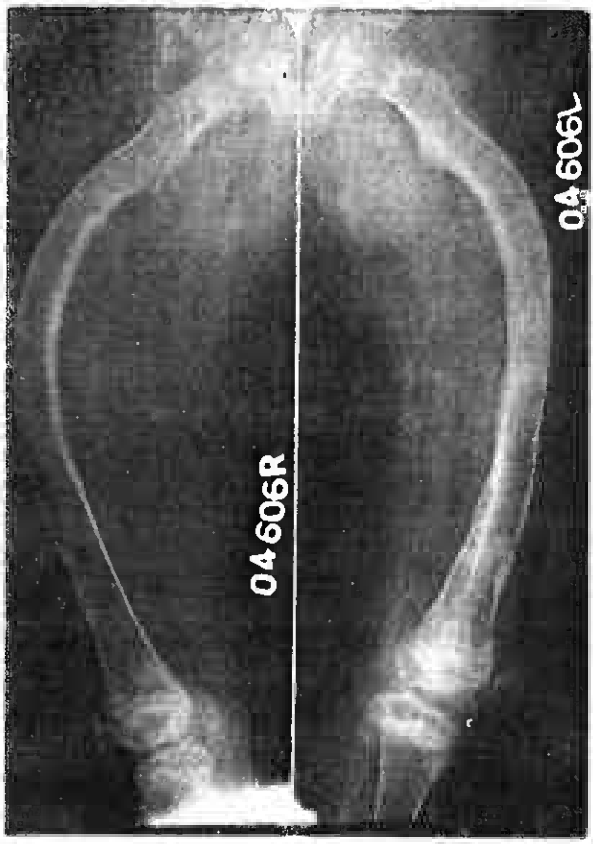


Fig. 11. Case III. Age 12 with femurs showing marked bowing of the upper one-third of the shaft, characteristic osteoporosis and thin cortex.

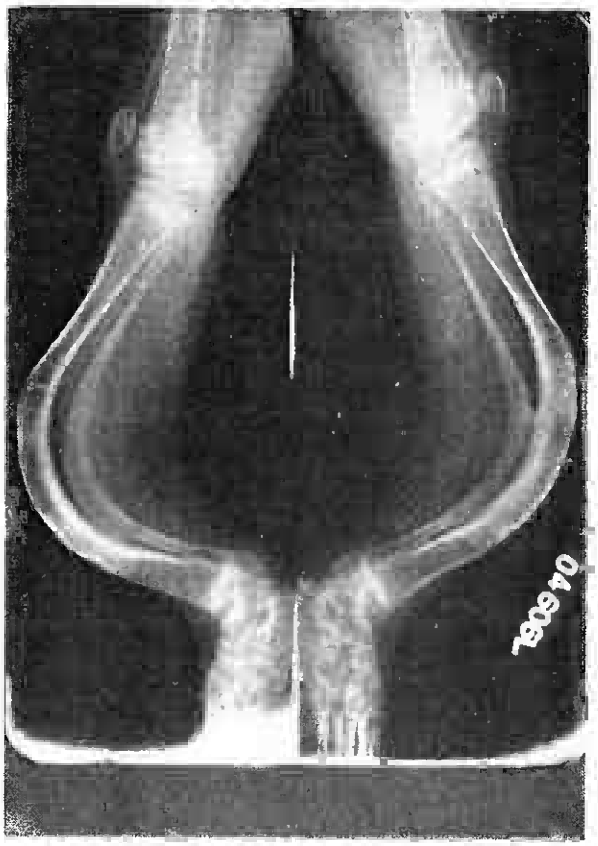


Fig. 12. Case III. Age 12 with tibia and fibula markedly deformed. Both bones were osteoporotic with thin cortex.

trivial injuries such as while crying in a fit of temper or when one of his elder brothers carried him to the bathroom (as he had never been able to walk). Often the fractures occurred without any apparent cause. All the fractures healed rapidly. The last fracture occurred about 10 weeks ago.

The brothers observed that during the first few days after sustaining a fracture the colour of his sclera deepened.

Milestones: A normal delivery, his growth was described as normal. He began to stand and speak at about 14 months.

Family History: There was no family history of similar illness. The patient was the 6th in a family of six sons. Other siblings were normal.

On Examination: The patient was well developed. The shape of his head was normal and the sclera of his eyes was deep blue in colour. He had good teeth.

He had scoliosis of his spine and marked bowing of the shafts of both his femur and tibia: the tibia was bowed anteriorly 90°. There was no undue laxity of his joints.

His respiratory central nervous and cardiovascular systems were normal. There was no abnormality of his abdomen.

X-rays (Figures 11-12): The skull was normal. There was osteoporosis of his bones generally. His spinal vertebral bodies were binconcave. His long bones were not only osteoporotic but the cortex were generally thin, and they were slender. There was evidence of healed fractures of the left radial and ulna shafts and both the femur and tibia shafts.

Other Investigations:

Blood count:

Total red count 4.09 million per cu. mm. Hb. 83%.
Total white count 5,700 per cu. mm.
Differential count: Polymorphs 62%, Lymphocytes 34%, Monocytes 2%, Eosinophils 2%.

Blood chemistry:

Serum calcium	12.5 mgm%
Blood alkaline phosphatase	16 units.

COMMENTS

Osteogenesis imperfecta (synonyms: fragilitas ossium, idiopathic osteopsathyrosis, periosteal dysplasia, brittle bones or Lobstein's disease) in a disorder of the skeleton characterised by its marked liability to pathological fractures. Two types are recognised: osteogenesis imperfecta congenita (prenatal) and osteogenesis imperfecta tarda (post-natal); the former is congenital, more severe and often fatal at or soon after birth, the latter is relatively mild.

The etiology is unknown, some observers, however, associate the disease with endocrine disturbances particularly to hyperthyroidism. It is hereditary only in a minority of cases. The prenatal type is believed to be transmitted through a Mendelian recessive and the prenatal type a variable dominant. It is a rare disease and both sexes are equally affected.

The clinical features are variable. They include blue sclerotics, otosclerosis and deafness, laxity of joints, osteomalacia, fractures, dwarfing, skull with prominent parietal and occipital bones (crane a'retord) defective milk teeth and often poor muscular development due largely to disuse. The blood chemistry usually normal, except for the occasional raised alkaline phosphatase and is of no diagnostic value. There is a tendency to improve and severe cases are known to have survived many years, although grossly disabled.

Fairbank considers that the essential pathology is imperfect formation and imperfect calcification of bone trabeculae. The bones are slender with thin cortex. Islets of cartilage, thickened periosteum, pathological fractures and a deficiency in the number of osteoblasts are features described.

Radiological appearances: Fairbank describes three types:

Type I—Thick bone type (Fairbank, 1930). These occur in severe prenatal cases whose major long bones are short, thick and osteoporotic with multiple fractures.

Type II—Slender fragile type. These are seen in the less severe prenatal and all postnatal cases. The major long bones are slender and osteoporotic with characteristically thin cortex.

Type III—Osteogenesis imperfecta cystica. This is extremely rare. It is progressive and is characterised by cystic appearance of the bones.

Case I is an example of a case of osteogenesis imperfecta tarda presenting with classical clinical and radiological features. The history that he sustained over 20 fractures, all of which

occurred in the shafts of the femur and the humerus is not a new observation. Fairbanks described a case with 23 fractures in 3 bones.

Case II is the sister of Case I and has the same disease. It is interesting to note that their parent's ascribed their disease to unsuccessful attempts to abort them, as no such attempts were made on the other apparently normal children.

Case III showed marked anteriorly curved deformity of the tibia, which is not uncommonly associated with the disease and is probably due to repeated fractures and softening of the bones, both of which are features of the disease. Wiles (1955) described that the bones may be soft and bend causing striking deformities and Howott (1952) wrote that the tibia is apt to be curved anteriorly. The surprising finding was the observation by his three brothers (all of whom were intelligent adults) that the sclera became more blue soon after a fracture. Such a feature has not been previously described. It is generally accepted that the blue sclerotic is not due to alteration in the thickness of the sclera but to its translucency, thus the choroidal pigment is more visible through it.

Lastly, the serum alkaline phosphatase which has been described as often raised was found to be above normal limits in Cases II and III, being respectively 20 and 16 units (normal 3 - 13 King-Armstrong Unit).

SUMMARY

Three cases of Osteogenesis imperfecta are described, two of which are from the same family.

ACKNOWLEDGEMENT

I am indebted to Mr. D. W. C. Gawne, M.A., M.D., F.R.C.S. and Dr. Khoo Oon Teik, M.D., M.R.C.P.E., F.R.F.P.S.G., for their valuable advice in the writing of this article.

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