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A Historical Vignette

"Be proud of yourself: you have a History!"

History of Osteogenesis Imperfecta or Brittle Bone Disease: a few stops on a road 3000 years long

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Abstract. *History of Osteogenesis Imperfecta or Brittle Bone Disease: a few stops on a road 3000 years long*. The attested history of osteogenesis imperfecta began three thousand years ago and it continues down to the present day. Through the centuries, we find a dislocated mummy of a child of Ancient Egypt, a young Arab named Satib, a Viking prince known as "Boneless", a subject of Louis XIV with a broken skeleton and finally, in the 20th century, a young deaf mother with blue sclerotics and a jazz pianist unable to walk except on crutches… Without being exhaustive, this review of contrasting cases attests to the universality and to the permanency of this disease.

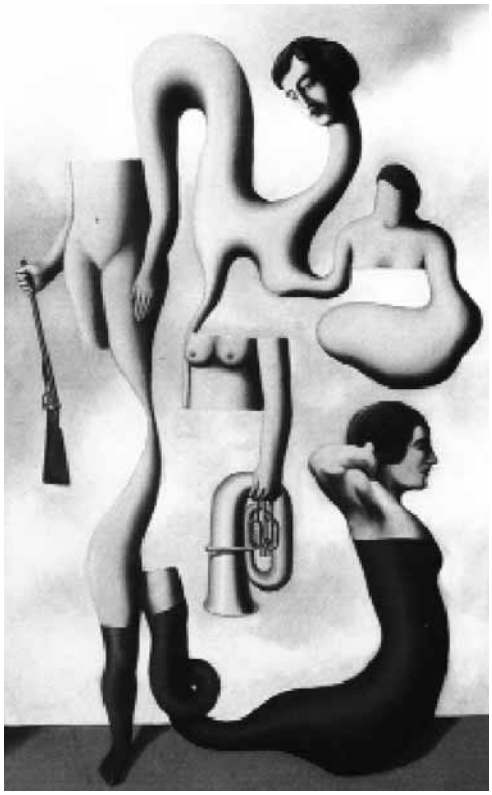


Figure 1
"The Acrobat's Exercises" of René Magritte, 1928. One of the characteristics of O.I. is the laxity of the joints and the deformation of the limbs.

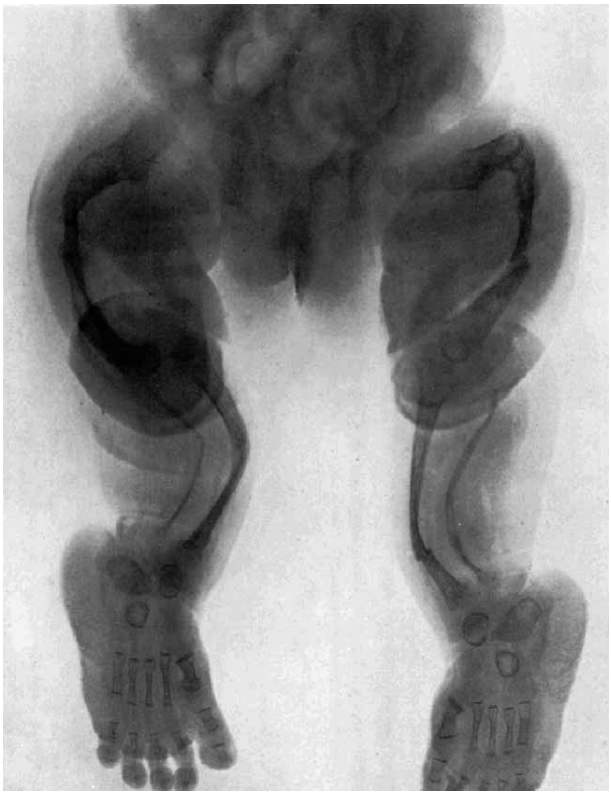


Figure 2
A severe case of prenatal O.I. (9 months) with several fractures in the ribs (not shown here) and the four limbs. There are fractures and major deformations of the bones in the lower limbs.[1]

Figure 3
Liverpool harbour in 1924. In the background, we see the customs building. By a curious coincidence, the city of Liverpool was involved in the history of O.I. At the beginning of the 20th century, two of its citizens played a role in the discovery of the disease: Adair-Dighton was a physician (see below) and Garstang was a Professor of Archaeology at the University. The University's archaeology museum now bears Garstang's name. It is associated with the SACE (School of Archaeology, Classics and Egyptology).

[1980 AD] Introduction: Soldier Blue*

One day, thirty years ago, I saw a patient at the Military Hospital in Brussels. I was struck by the similarity of the blue-grey colour of his Air Force uniform and the colour of the sclerotics of his eyes. He came to me for his deafness. He was a career non-commissioned officer of about thirty with no noticeable history of fractures. His deafness was conductive. I operated on him, and found a crumbly but stiff stapes. I conducted a stapedectomy and used a Teflon piston. The result was good. The biopsy of the stapes attested to a disease I never encountered before, and it was the only time during my military career that I came across osteogenesis imperfecta (O.I.) I should point out that the Military Recruitment and Selection Centre usually did its job well...

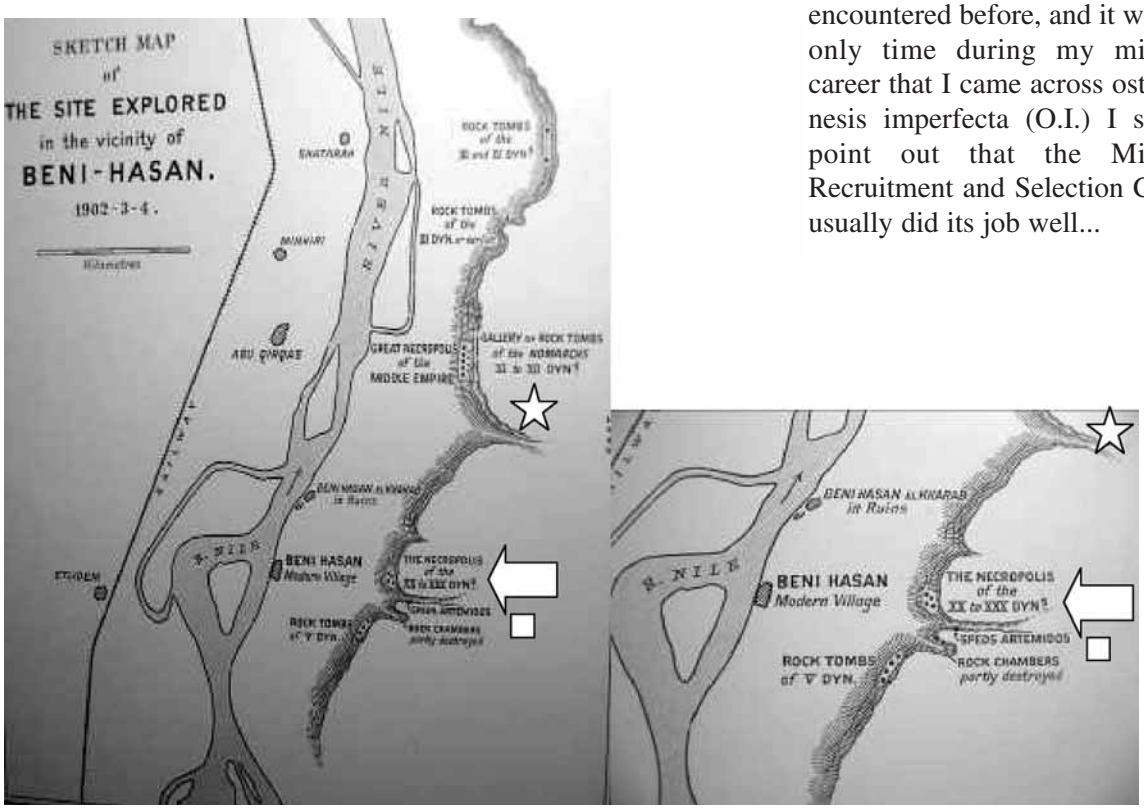


Figure 4
A map showing the site of Beni Hassan from Garstang's book "The burial customs of Ancient Egypt", published in 1907. The star indicates the tombs usually visited today by tourists. After disembarking on the banks of the Nile, they must climb up a rather long slope before arriving at a cliff containing the graves. The arrow indicates the place where the famous mummy of "a monkey" was found. The square designates the temple of Artemis, "speos Artemidos", as it was named erroneously by the Greeks.[2]

* This is a reference to the blue of the uniforms of the Northern Armies in the War of Secession. It is also the name of a contentious Western movie of the 1970s, which was filmed against the background of the My Lay massacre during the Vietnam War. The movie told of the slaughter of Cheyenne Indians at Sand Creek, Colorado, by the "Soldiers Blue" in 1864.

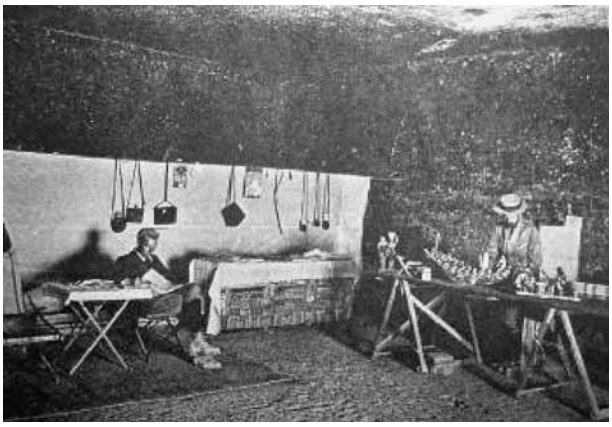


Figure 5A
A view of one of the graves of Beni Hassan that was fit out for use as a living room for Garstang (we see him standing, engaged in sorting archaeological materials). A painter friend (seated) accompanied him during his expedition. An underground room allowed an Egyptian collaborator to develop photographs (more than 1000), 300 of which were reproduced in Garstang's book "The burial customs of Ancient Egypt".[2]

Figure 5B
John Garstang later in life

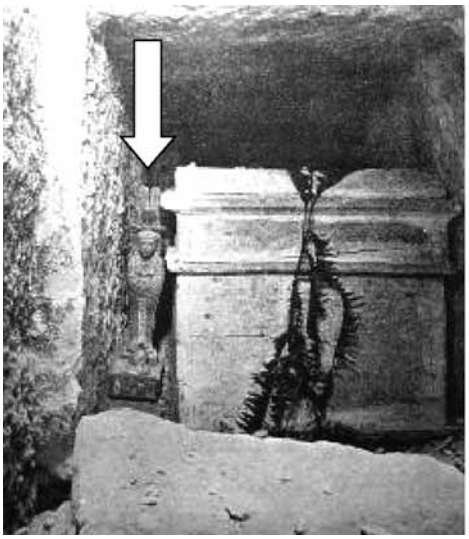


Figure 6
Example of one of the burial chambers of Beni Hassan. Garstang discovered, beside the principal tomb, a smaller sarcophagus standing up (arrow). The latter is of the same type as the one in which Garstang found what he thought was the mummy of a monkey (Figure 10). On the principal tomb shown here, we see some garlands in their original position, and, in front, the stone that closed off the entrance to the tomb.

Figure 7
Garstang had good reason to hesitate about the true nature of the occupant of the small sarcophagus: it was not unusual to find mummies of animals in smaller sarcophagi alongside the principal tomb. We see here a baboon, after removal of the wrapping bandages, which was found in the grave of the Theban priestess Makare. Usually, the animals were pets of the deceased, or votive offerings.[3]

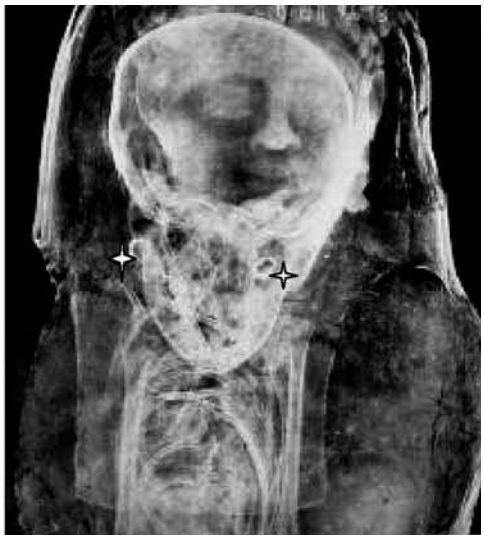
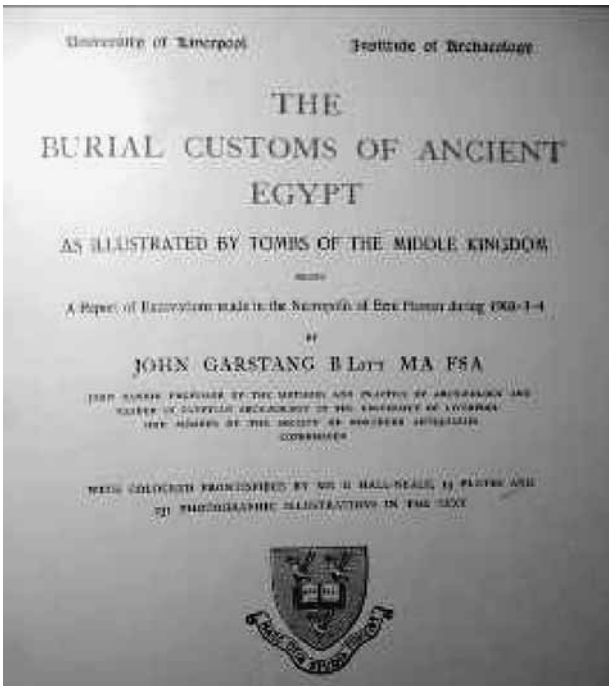


Figure 8
Garstang was also justified in wondering whether the fractures of the occupant of the small sarcophagus in Beni Hassan were post-mortem lesions (Figure 10 and 11). This child's sarcophagus, dating from the Roman period and X-rayed by Gray, shows the superposition of the mask of the deceased on the skeleton, which was roughly handled because of the narrowness of the coffin. The head has been forced in flexion, the thorax has been squeezed so much that the clavicles and the ribs both overlap. We can make out two earrings (stars). In the case of the mummy discovered by Garstang with O.I., the coffin was actually too large for its occupant. That rendered unlikely the possibility of *post-mortem* fractures due to the narrowness of the container. Logically, the archaeologist had to suspect *pre-mortem* fractures. Nevertheless, two hypotheses remained open: accident or brittle bones.[4]


University of Liverpool Institute of Archaeology

THE
BURIAL CUSTOMS OF ANCIENT EGYPT

AS ILLUSTRATED BY TOMBS OF THE MIDDLE KINGDOM

JOHN GARSTANG B Litt MA FSA

Figure 9
Frontispiece of John Garstang's book reporting on the results of his excavations in Beni Hassan (Middle Egypt) during the winters of 1902-1903 and 1903-1904.[2]

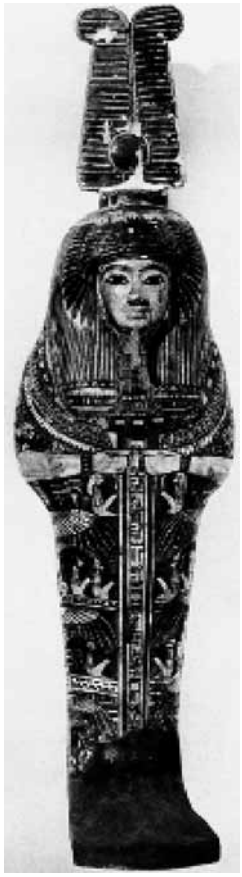


Figure 10
The sarcophagus containing the remains that were to be found later to be the first known case of osteogenesis imperfecta. It was transferred to the British Museum. In 1968, Dawson and Grey would list it in their catalogue of the Egyptian Antiques of the British Museum: "Infant in cartonnage case; date 22nd dynasty, from Beni Hassan (Speos Artemidos); date of acquisition: 1905. The bones being those of a child, might suggest a Roman date, but nothing in the archaeological evidence would support such a dating. Undoubtedly it must have been the peculiar appearance of these bones which led Garstang to describe the contents of the case as *bones of monkey (?)*. It is evident that the cartonnage itself could have accommodated a larger child. The case is remarkable because it is surmounted by the double plumes. It resembles closely a normal figure of Ptah-Seker-Osiris. The foot end of the case is now missing."
The sarcophagus was 73 cm. long.[5]

Thereafter, however, I was haunted by this pathology, and from time to time I studied it in more detail because I wanted to link this minor personal history to the broader history of the disease.

I. [1000 BC] The coffin of "a monkey" dating from 1000 BC is discovered in Egypt by John Garstang, an archaeologist from Liverpool[2-11]

In a certain sense, a medical ailment is the logical meeting point for an ill person and a physician. However, on occasion, the meeting will be fortuitous; the "physician" is not always a real physician, the ill person may be a child who is taken for a monkey (!) and, finally, the diagnosis of the illness may wait for sixty years before being made, inevitably, by somebody else!

John Garstang (1876-1956) was professor of archaeology at Liverpool University for more than thirty years (Figures 3,5B). He was involved in several digs in the Near East and in Egypt, where he was first assistant to Petrie, another British archaeologist to whom we owe the discovery of some remains of the Egyptian Labyrinth, not far from the Fayoum Lake in Lower Egypt. Later, Garstang returned to the site of Beni Hassan[2] in Middle Egypt, where he carried out two series of excavations during the winters of 1902-1903 and 1903-1904 (Figure 5B). By coincidence, he was financed by the discoverer of the Cretan Labyrinth, another British archaeologist, Sir Arthur Evans, director of the Ashmolean Museum of Oxford.

In the southern part of the Beni Hassan site, not far from a temple which was named by the Greeks "speos Artemidos" or the temple of Artemis (Figure 4), Garstang

discovered a series of graves, in which the tomb-like rooms sometimes contained a smaller sarcophagus (Figure 6). It was in this type of coffin that Garstang thought that he found the mummy of a monkey (Figure 7). The skeleton had multiple fractures and its fragments were completely disorganised.

• The monkey hypothesis

It is not rare to find mummies of various animals, sometimes in large numbers, accompanying the deceased, gathered all together in the same covering. In the Guimet museum of natural history in Lyon, there is a mummy of a baboon found in the grave of a Theban priestess of the 21st dynasty (Figure 7). Elsewhere, thanks to radiography, it was found that a mummy thought to be that of a child was in reality that of two cats brought together for the occasion! On the site of Beni Hassan itself, there was a cat cemetery dedicated to the goddess Pakhet, the protector of the region.

Garstang had this in mind when he wrote his book "The burial customs of Ancient Egypt" in 1907 (Figure 9).[2] Talking about the tombs of the 20th and 21st dynasties, he wrote: "the tendency to animal worship was rapidly growing, or throwing off its disguise, and is illustrated here and there by objects found within these graves. The mummied monkey encased in a painted cartonnage in the form of an Osiris, shown in Figure 219, is an illustration [corresponding precisely to the mummy that concerns us] (Figure 10). At a later time, such cults took definite and local forms, illustrated at Beni Hassan by the cemetery of cats lying only half a mile distant..."

It should, however, be noted that, in spite of the self-confidence displayed in the principal part of his text, Garstang's description of his discoveries in brief in the appendix to his book reveals a doubt about whether the mummy is actually that of a monkey. Indeed, he adds a question mark to his inventory: "Cartonage, brightly painted, containing bones of monkey **(?)**".

According to the radiologist Gray, Garstang opened the sarcophagus, and took out its contents with the wrapping. Its strange appearance may have suggested to him the idea that it was the mummy of a monkey.

Be that as it may, the coffin and its mummy were presented to the British Museum, which bought them in 1905, that is to say one year after the end of the excavations. The item remained in that condition there for just over sixty years...

• Radiologist P. H. K. Gray scrutinises the sarcophagus sixty-five years later (Figure 11).

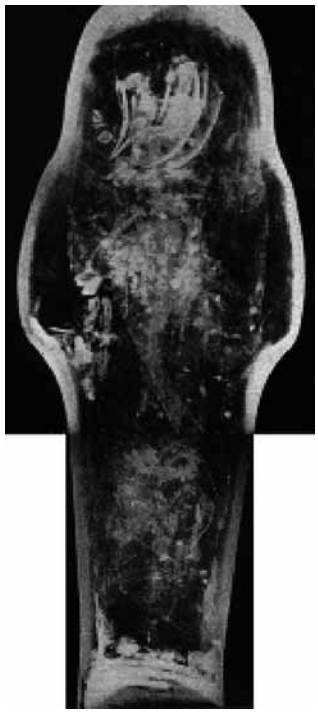


Figure 11
The X-ray of the Beni Hassan mummy made by Gray in 1967. It showed a jumble of strange bones. Removal of the wrappings made it possible to see that the mummy was that of a child and, moreover, the oldest known case of O.I.[10]

As early as March 1896, three months after the discovery of X-rays by Roentgen in 1895, W. Köning published the first X-ray of a mummy.[6]

Very soon after, in 1897, Petrie published some excellent plates of the lower limbs of his mummies.[7]

In 1931, Moodie published a brief description of his radiological studies of 17 mummies.[8]

In 1964, Gray X-rayed 78 mummies of the British Museum. It was at this time that he X-rayed the mummy of Beni Hassan, a detailed study of which he published in 1969.

In 1966, Gray published the result of his X-ray studies of the mummies of Ancient Egypt from the Royal Antiquities Museum in Leiden.[9]

In 1968, Dawson and Grey published a catalogue of the Egyptian antiques of the British Museum that included the mummy of Beni Hassan.[5]

In 1969, Gray published his fundamental paper about this mummy, securing the collaboration of a member of the Natural History Museum who reconstructed the skull of the subject, a professor of oral pathology who examined the dentition, another pathologist who examined the rest of the skeleton, and finally a member of the Research Laboratory of the British Museum, who X-rayed the long bones (Figure 11).[10]

Figure 12A
After painstaking efforts, the skull of the mummy of Beni Hassan was more or less completely reconstructed. It had many Wormian bones. It was characterised by the "tam-o'-shanter" deformity typical of the disease.

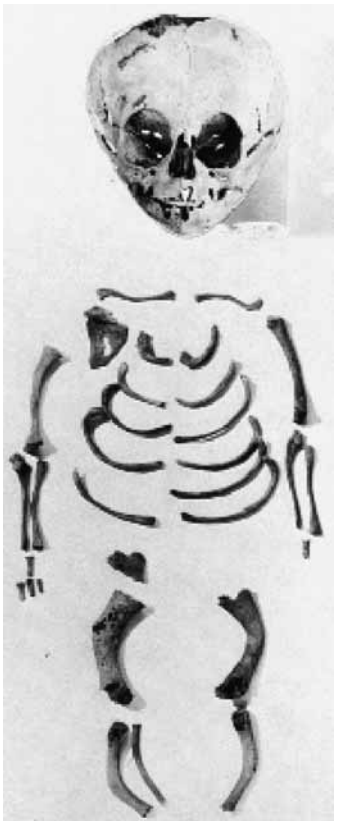


Figure 12B
The reconstructed skeleton of the mummy of Beni Hassan is characterised by abnormal curvatures of the limb bones, particularly those of the lower limbs, and a pronounced thickening of the femoral bones.[10]

Figure 13
A case of infantile O.I. in a contemporary Egyptian girl aged 12 years. This form is less serious than the congenital one. This subject was 105 cm tall and weighed 17 kg. She learned to walk at the age of four with assistance only. In practical terms, she had to remain seated in her bed. She was monitored until the age of 16: she remained a dwarf and failed to attain puberty. Her skull and her teeth were normal but her thorax was severely distorted, the position of her shoulder blades was asymmetrical, there was a kyphosis and a scoliosis of the vertebral column, the limbs were thin and their muscles were atrophic, the left forearm was distorted, both hips were ankylosed and there was an abnormal curvature of the left femoral bone.[11]

During the 1970s, Gray succeeded in collecting X-rays of 133 mummies preserved in several European museums, in particular in Great Britain.

• The results of the radiological studies, particularly of the Beni Hassan mummy (Figure 11)

The mummy contained a disorganised skeleton, not of a monkey but of a child of indeterminate sex. The child had suffered from a rare disease, osteogenesis imperfecta. The foot end of the case, which was 73 cm. long, was missing. It was obviously made for a larger child (Figure 10).

The mummy dated from the 21st / 22nd dynasty (third Intermediate Period) between 1000 and 850 BC. It had numerous fractures, some of which may have been caused by the manipulation associated with mummification, given the fragility of the bones. Otherwise, O.I. was shown by the curvatures of the lower limbs, the abnormal thickening of the femoral bones compared with the bones of the upper limbs (Figure 12B), the appearance of the cortical bone, with thin and undulating lines, and the spongioid bone, which had been reduced to scattered strands of amorphous material. Turning to the dentition, the roots of the molars were abnormally small compared with the crowns, and the tubular structure of the dentine was malformed. The reconstructed skull had a large number of Wormian bones and there was a Scottish beret deformity (a "tam-o'-shanter" deformity); this explains the triangular appearance described as "*à rebords*" (with projecting edges) in the French literature (Figure 12A,12B).[10]

The study of the DNA, which was not conducted at that time, could have confirmed the diagnosis and made it more precise. Indeed, we know today that the genes involved are gene COL 1, situated on chromosome 17 and commanding two of the three chains of collagen (alpha 1 chains), and gene COL 2, situated on chromosome 7, commanding the third chain of collagen (alpha 2 chain).

Figure 14
Picture of Vikings dating from the 9th or 10th century.

Figure 15
Ivar the "Boneless" transported on a shield. Extract from a BBC TV programme from 2003. The actor playing Ivar's role was himself suffering from the same disease, O.I. He was Nabil Shaban, a British actor of Jordanian origin.

II. [872 AD] Ivar the Boneless, a Viking chieftain, dies in Dublin[12]

An historical figure of legendary stature, Ivar the Dane lived in the 9th century AD (794-872). He could claim to be the first famous European to suffer from the disease.

His destiny was indeed remarkable. The story has been passed down to us in the Scandinavian saga of Ragnar Lodbrok. In 865, to avenge his father Ragnar's murder by Aella, the king of Northumberland, Ivar invaded East Anglia and Northumbria in the East of England (Figure 14). From 869 onwards, this invasion led to a series of murders and conquests. The king of East Anglia was canonised later under the name of St. Edmund because he refused to become the vassal of a pagan (Figure 16). The Danes conquered the city of York, naming it Jorvik.

The story ends with the ritual murder of Aella, the principal culprit. Ivar suggested that "the

Figure 16
St. Edmund's martyrdom. He was shot dead by the Viking archers. This mediaeval painting is in the church of Bishopbourne, near Canterbury, Kent.

Figure 17
A section of an engraved stone in the city of Lärbro on the island of Gotland (Sweden). It represents the torture known as "the Blood Eagle". The victim lies on his belly; the sacrificer is ready to open up his back. The figure at the top probably represents an eagle. The complete flat stone from which this stylised fragment comes is 3 metres high. It is located in the Statens Historika Museet of Navavagen, Sweden. It dates from the Early Middle Ages (400-1000 AD).

blood eagle" should be carved on his back. Aella's back was opened, his ribs were disconnected from his vertebral column, and then turned outside to transform them in a sort of bloody wings; finally Aella's lungs were taken from his thorax and brought as an offering to Odin (Figure 17).

Ivar died in peace in Dublin, but he had expressed his desire to be returned to England and be buried in a tumulus near the coast, at Hastings facing Normandy. He said that, as long as his body mounted guard on that section of the English coast, no enemy would be able to invade it. The saga reports that his prophecy proved true until William the Conqueror landed there, went to the site of the tumulus and noticed that Ivar's corpse had not decomposed. William then had a great funeral pyre erected on which he had Ivar's corpse burnt. Having warded off the prophecy, William succeeded in invading the country and secured a definitive victory.

• The origin of the name "Boneless"

In 1949, well before the radiographic study of the mummy of Beni Hassan by Gray, one of Ivar's distant Danish compatriots, Knud Seedorf, published a paper entitled "Osteogenesis imperfecta: a study of clinical features and heredity based on 55 Danish families". He wrote: "*Of historical personages the author knows of only one of whom we have a vague suspicion that he suffered from osteogenesis imperfecta, namely Ivar Benlos, eldest son of the Danish legendary king Regnar Lodbrog. He is reported to have had legs as soft as cartilage ("he lacked bones"), so that he was unable to walk and had to be carried about on a shield*".[12]
It is probable that, with the exception of his inability to walk, Ivar was quite normal because he was afflicted with a less severe form of the disease. The actor Nabil Shaban, who was affected by the same disease and played the role of Ivar in 2003 (Figure 15), demonstrated that it was possible for a patient like him to use a longbow and to participate actively in battle, something that the Viking warriors naturally expected from a chief.

• The legendary origin of Ivar's disease

According to the saga, Ivar's malformation was caused by a curse. His mother was the second wife of his father and she had magic powers and the gift of prophecy. She warned her husband to wait for three nights before consuming their marriage:

"Three nights together, but yet apart,
Shall we bide, nor worship the gods as yet;
My son will by this be saved from lasting harm,
For boneless is he thou wouldst now beget".

But Regnar refused to believe in the curse and made love to his new wife without delay. The result was the birth of Ivar the "Boneless". In effect, he had bones resembling tendons. He grew up unable to walk, and had to be carried everywhere on poles or on the back of a shield.

• Other interpretations of the nickname

However, the epithet "boneless" may also be scabrous, suggesting sexual impotence. This interpretation is supported by another passage of the saga, in which it is stated that "*neither love nor lust played any part in his life*". Indeed, Ivar had no descendants. This interpretation inspired another British film in 1989 in which Ivar was a rather weedy-looking and cowardly man who spoke with a falsetto voice!

Other explanations are also possible. Given well-attested Viking humour, one of their own who was taller than the others with strong and impressive bones may have been given the ironic name "boneless", just as we might call a corpulent person "Twiggy"...

Finally, the epithet "boneless" may also be attributed to a lanky, gangling appearance due to the obvious flexibility of Ivar's joints. Let us not forget that, according to the saga, the tendons had replaced the bones.

Be that as it may, the cremation of Ivar's corpse on the orders of William the Conqueror has deprived us forever of any objective way of establishing his actual physical condition, opening the way to legend rather than history.

III. [Ancient Times] Arabia too? The history of Satib[13-16]

In indeterminate "ancient times", an Arabic writer named Gschuzio or Gschaubarius [quoted by Ekman[13,14] in his thesis for doctor of medicine written in Latin in 1788, who was himself quoting from a quotation by Reiske[15] dating from 1776], "a certain Satib or Satibum had himself carried where he liked on a net made of palm-tree branches, because he had no bones except in his head, his neck and his hands. The rest of his body from the feet to the bones

of the neck could be folded like simple clothing. Except for his fingers, he was unable to move anything else. He owed his name Satibum or 'soothsayer' to the fact that Dzeib's sons told that except for his head, he had no bones".
Unfortunately, it is unclear when this character lived. Furthermore, in his paper about O.I., Weil, a professor of orthopaedics at Yale University, acknowledges that he was unable to confirm Ekman's writings.[16]

IV. [1675] The Philosopher Nicolas de Malebranche evokes the torture on the wheel in relation to an osseous disease generally thought to be osteogenesis imperfecta[17-20]

Nicolas de Malebranche was a precise contemporary of Louis XIV. He was born and died in the same years as the Sun King: 1638-1715 (Figure 18).

His physical condition was, however, fragile. Fontenelle, who praised him at the Academy of Sciences (which de Malebranche joined in 1699), said that "he had a twisted spine and a deeply sunk sternum". Nevertheless, it should not be thought that he suffered from osteogenesis imperfecta...

In any case, his general condition meant that he was taught by a tutor at home until the age of sixteen. He then attended theology lectures at the Sorbonne, receiving ordination at the age of 26. He joined an Augustine order named "the Oratory". He immersed himself in the study of St. Augustine, who had a major influence on him for the rest of his life.

However, another serendipitous event was also to have an influence on him. An edition of Descartes' "Treatise on man" came into his possession and from that moment onwards he was fascinated by the Cartesian method, which he studied for a decade. It finally inspired him to produce his most famous work in 1688: *De la recherche de la vérité. Où l'on traite de la nature de l'Esprit de l'homme, & de l'usage qu'il en doit faire pour éviter l'erreur dans les Sciences* ["Concerning the Search after Truth. In which is treated the nature of the human mind and the use that must be made of it to avoid error in the sciences"] (Figure 20).[17] That work contains an allusion to medicine, and in particular what was to be known later as "osteogenesis imperfecta". However, the pathological reference remained subordinate to the philosophical theme, touching only in a distant way on the genetic reality of the disease.

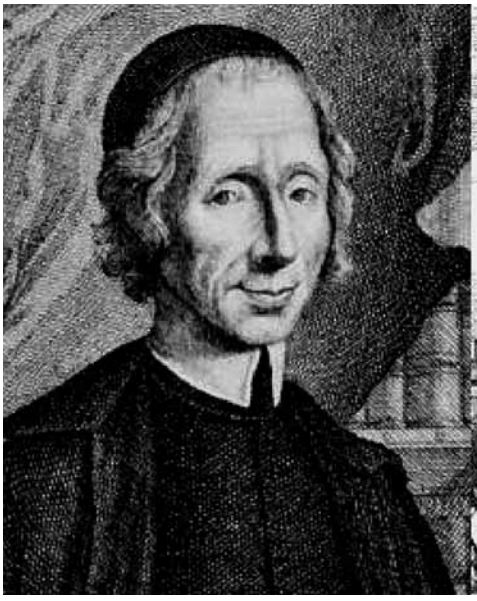


Figure 18
Engraving by de Rochefort showing de Malebranche (1707). De Malebranche was the first to describe a patient suffering from O.I., but he was totally unaware of the nature of that disease. He attributed it to mysterious connexions between the sight and brain of the mother and the physical condition of the child she was carrying.

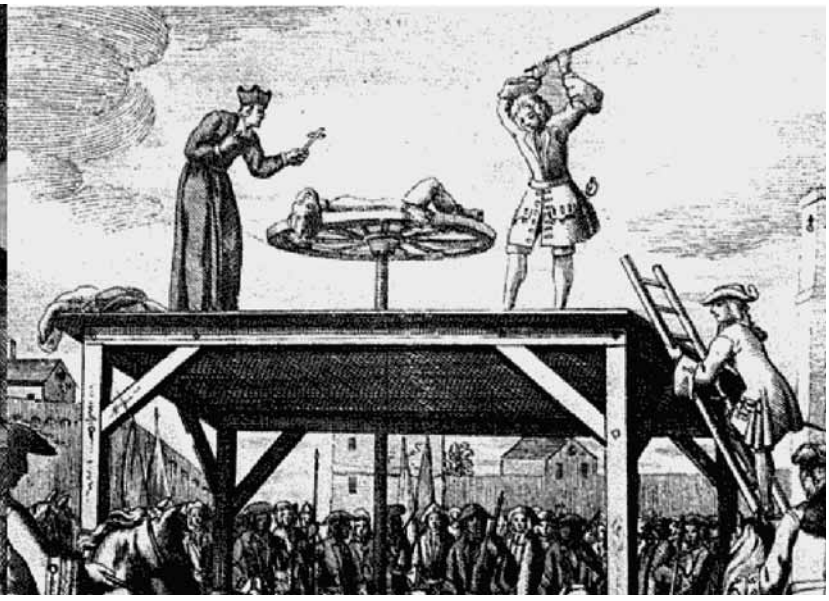
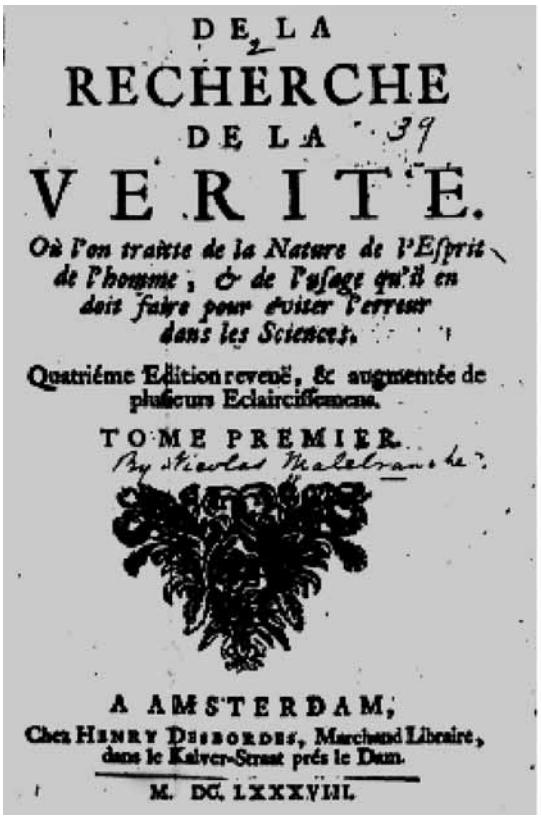


Figure 19
The torture on the wheel intended to break the limbs of the victim with heavy strokes of a square iron bar with a handle. Eight blows were first given to break each limb twice. Two further blows were then given: one on the chest and the other on the stomach. The victim was then left to die on the wheel. Sometimes he would die strangled. The famous Mandrin was killed in this way in 1755.
[Engraving published with the kind permission of Thierry Tillet, the webmaster of the site blisetborn.free.fr.].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VERITE.
Où l'on traitte de la Nature de l'Esprit de l'homme, & de l'usage qu'il en doit faire pour éviter l'erreur dans les Sciences.
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Figure 20
Frontispiece of de Malebranche's book published in 1688. Curiously his name was added by hand in English and without the "de".[17]

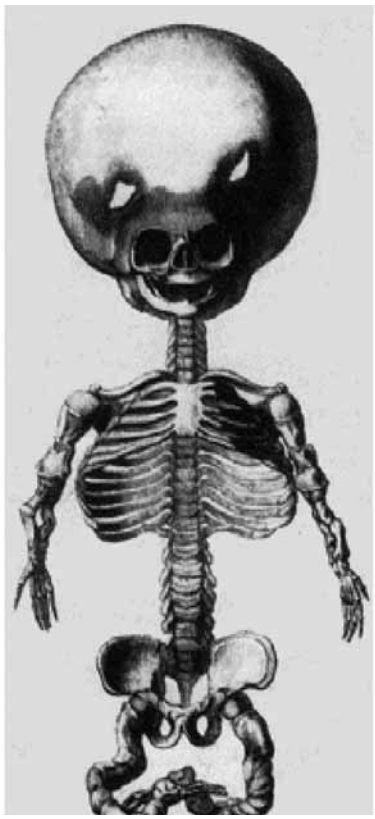


Figure 21
This is probably one of the first deliberate representations of O.I. (1758). It is a plate from a work by Toussaint Bordenave (1728-1782), probably his *Recherches sur l'ostéogénie ou la formation des os* [Researches on osteogenesis or the formation of bones] (1758-1763). He also published *Recherches sur la façon dont se fait la reunion des os fracturés* [Researches on the way that fractured bones are reunited].[18]
The skull represented here corresponds to one of the most severe forms of O.I., with a skull that is still membranous, with enlarged fontanels and sutures. We see several fractures of the long bones with deformations and telescoping of the fragments. In the field of ENT, Bordenave is mainly known for his work on the maxillary sinus and the treatment of lachrymal fistulas.

Figure 22
Willem Vrolik (1801-1863) was the first to introduce the term "osteogenesis imperfecta" in 1845 and to make of it a disease in the proper sense. This plate from 1849 precedes the one above (Figure 21). It shows, in addition to the deformation of the limbs, the scattered ossification of the upper skull, in islands. These are named Wormian bones after the Dane Olaus Worms. They are particularly numerous here.[19,20]
There is a museum of anatomy and teratology in Amsterdam named after Vrolik. It was founded by Willem Vrolik and his father Gerard, who was also a professor of anatomy. A minor historical fact worthy of mention is that Willem Vrolik participated on the Dutch side in the war against the young Belgium in 1830...

• De Malebranche's succinct description of the disease[17]

After writing a first book devoted to the *Erreurs des Sens* [Errors of the Senses], it was in the first part of book II dealing with the errors due to the imagination that de Malebranche included chapter VII entitled *I De la communication qui est entre le cerveau d'une mere & celui de son enfant. II De la communication qui est entre notre cerveau & les autres parties de notre corps, laquelle nous porte à l'imitation & à la compassion* ***III Explication des enfants monstrueux & de la propagation des espèces*** *IV Explication de quelques dérèglements de l'esprit & de quelques inclinations de la volonté. V De la concupiscence et du pêché originel. VI Objections & réponses*. [I. About the communication between the brain of a mother and that of her child II. About the communication that exists between our brain and the

other parts of our body, that brings us to imitation and compassion **III. Explanations relating to monstrous children and the propagation of the species** IV. Explanation of some disturbed spirits and of some inclinations of the will V. About concupiscence and original sin VI. Objections and responses].

III. *Il y a environ sept ou huit ans, que l'on voyoit aux Incurables un jeune homme, qui était né foû, & dont le corps étoit rompu dans les mêmes endroits, dans lesquels on rompt les criminels* (Figure 19). *Il a vêcu près de vingt ans en cet état ; plusieurs personnes l'ont veu, & la feuë Reine mere étant allée visiter cet Hospital eut la curiosité de le voir, & même de toucher les bras, & les jambes de ce jeune homme aux endroits où ils étaient rompus*.

[III. About seven or eight years ago, a young man could be seen at the Incurables hospital who was born mad and whose body was broken in the same places as murderers' bones are broken. He lived for about twenty years in this condition; several people saw him and the late Queen Mother, when paying a visit to this hospital, was curious to see him and even to touch the arms and legs of that young man where his limbs were broken (Figure 19).]

This text is the reason why de Malebranche was justly credited for being the first author to notice and describe a case of osteogenesis imperfecta long before the term existed.

• De Malebranche's account of the pathogenesis of the disease[17]

As we can see, the priest-philosopher interpreted the pathogenesis of the disease in his own way:

"*...la cause de ce funeste accident fut, que la mere ayant sçeu qu'on alloit rompre un criminel, l'alla voir exécuter. Tous les coups que l'on donna à ce misérable, fraperent avec force l'imagination de cette mere, & par une espèce de contrecoup le cerveau tendre & délicat de son enfant. Les fibres du cerveau de cete femme furent étrangement ébranlées, & peut-être rompuës en quelques endroits par le cours violent des esprits produit à la veuë d'une action si terrible, mais elles eurent assez de consistence pour empêcher leur bouleversement entier. Les fibres au contraire du cerveau de l'enfant ne pouvant résister au torrent de ces esprits furent entièrement dissipées, & le ravage fut assez grand pour lui faire perdre l'esprit pour toûjours. C'est là la raison pour laquelle il vint au monde privé de sens.*

Voici celle pour laquelle il étoit rompu aux mêmes parties du corps que le criminel, que sa mere avoit veu mettre à mort. A la veuë de cette exécution si capable d'effraïer une femme, le cours violent des esprits animaux de la mere alla avec force de son cerveau vers tous les endroits de son corps, qui répondoient à ceux du criminel, & la même chose se passa dans l'enfant. Mais parce que les os de la mere étoient capables de résister à la violence de ces esprits, ils n'en furent point blessez. Peut-être même qu'elle ne ressentit pas la moindre douleur, ni le moindre frémissement dans les bras ni dans les jambes, lorsqu'on les rompoit au criminel. Mais ce cours rapide des esprits fut capable d'entraîner les parties molles & tendres des os de l'enfant. Car les os sont les dernières parties du corps qui se forment, & ils ont très- peu de consistence dans les enfans qui sont encore dans le sein de leur mère. Et il faut remarquer, que si cette mere eût déterminé le mouvement de ces esprits vers quelqu'autre partie de son corps en se chatoüillant avec force, son enfant n'auroit point eu les os rompus, mais la partie, qui eût répondu à celle vers laquelle la mere auroit déterminé ces esprits, eût été fort blessée, selon ce que j'ai déjà dit".

[" ... the cause of this terrible accident was the fact that the mother, having heard that a criminal was to be put on the wheel, went to see the execution. All the blows that were given to that wretch struck the imagination of that mother forcefully and, as an indirect consequence, the tender and delicate brain of her child. The fibres of the brain of that woman were strangely shaken, and perhaps broken in some places by the violent course of the spirits produced by the view of such a terrible action, but these fibres were sufficiently resistant to prevent their complete disruption. By contrast, the fibres of the brain of the child could not resist the torrent of these spirits and were entirely dissipated, and the devastation was so great that it made him lose his mind for ever.

This is why he came into the world deprived of sense.

This is why he was broken in the same parts of the body as those of the criminal whose execution had been watched by the mother. At the sight of that execution, so apt to frighten a woman, the violent course of the animal spirits of the mother moved forcefully from her brain to the other places of her body that corresponded to those of the murderer, and the same thing happened in the child. But because the bones of the mother were able to resist the violence of

these spirits, they were not injured by them. It is even possible that she felt not the slightest pain or trembling in her arms or legs, when those of the murderer were broken. But this rapid course of the spirits did involve the soft and tender parts of the child's bones. For these are the last parts of the body to be formed, and there is little consistency in them when children are still in their mothers' womb. And it should be noted that, if that mother had directed the movement of the spirits to another part of her body, by tickling herself forcefully for instance, the bones of her child would not have been broken, but the part of the child responding to the part of the mother where these spirits were directed would have been seriously injured, as I stated above].

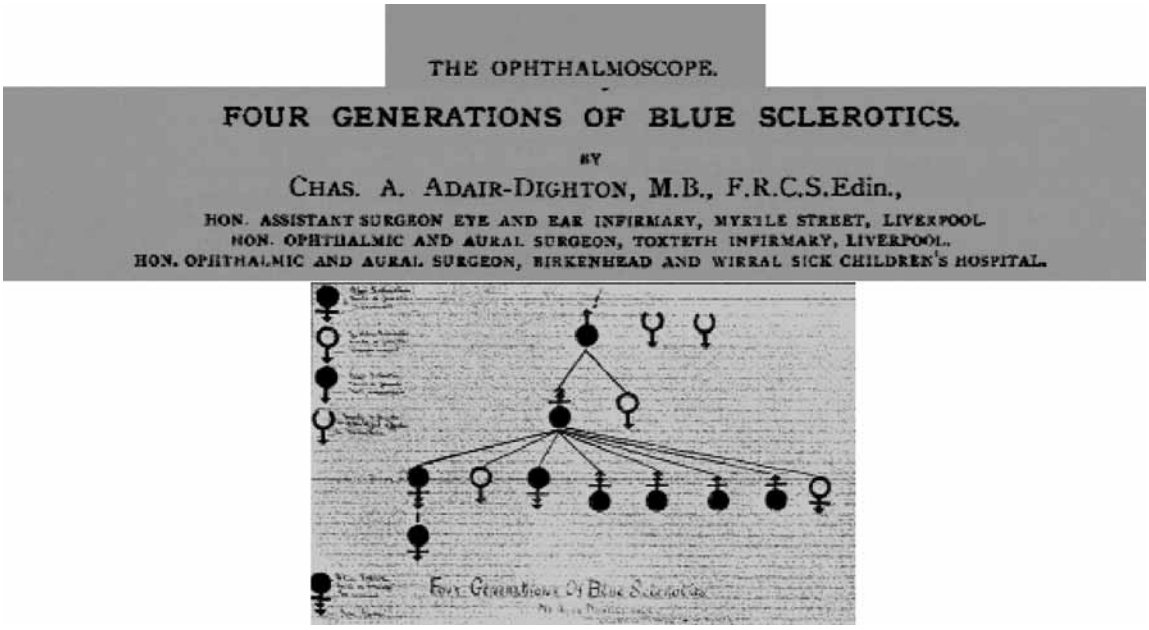
According to de Malebranche, the sight of a horrible spectacle acts by a sort of sympathy on the corresponding organs of the witness (here, the mother), but it cannot cause identical lesions on them because her organs are resistant, whether they are the brain or the bones. By contrast, because of a similar sympathetic action between the mother and child, there are effects on the new-born in so far as his brain or his bones are less resistant.

Popular imagination today still preserves the notion that pregnant women must avoid gruesome sights in early pregnancy to prevent damage to the unborn child. Gynaecologists do not believe that this idea has a scientific basis, except in so far as stress could provoke a miscarriage.

V. [1912] Charles Allen Adair-Dighton describes for the first time a case of deafness in four generations of blue sclerotics[21-23]

In 1912, the association of deafness with osteogenesis imperfecta was mentioned for the first time in a modest paper of two pages published by Charles Adair-Dighton in the English review "The Ophthalmoscope" and entitled "Four generations of blue sclerotics" (Figure 23).[21]

At the beginning of his presentation, the author affirms with typical British humour: "The shortness of my reports is due to the fact that the patients realised, or over realised, the value of their histories, and rendered further investigations impossible for any but a millionaire. Otherwise, I was


THE OPHTHALMOSCOPE.

FOUR GENERATIONS OF BLUE SCLEROTICS.

BY

CHAS. A. ADAIR-DIGHTON, M.B., F.R.C.S.Edin.,

HON. ASSISTANT SURGEON EYE AND EAR INFIRMARY, MYRTLE STREET, LIVERPOOL.
HON. OPHTHALMIC AND AURAL SURGEON, TOXTETH INFIRMARY, LIVERPOOL.
HON. OPHTHALMIC AND AURAL SURGEON, BIRKENHEAD AND WIRRAL SICK CHILDREN'S HOSPITAL.

Figure 23

The only paper, dating from 1912, published by Adair-Dighton concerning the association between blue sclerotics, osseous fragility and a case of deafness. The deaf patient is located at first left at the level of the third generation. At the time of the paper, her child (4th generation) was too young to be considered as a possible case of deafness.[21]

We have two other papers from the same author about a quite different subject: sarcoma of the nose. They date from 1913 and 1914 and were published in the British Journal of Surgery. Adair-Dighton was later forced to abandon his medical career (see below).

hoping to obtain the co-operation of an expert radiographer (*X-rays had only discovered 17 years before)* and write a complete paper from both the ophthalmic and orthopaedic side of the question (*no mention of deafness)*, and thus possibly offer conclusive proof of the dependence of blue sclerotics and osteoporosis one upon the other or some factor common to both".

He continues in the same manner, with praise too excessive to be honest of the Welsh origin of his patients: "The family, I may add, is of Welsh extraction, so that I am only one more of the medical world who owes something to Wales".

After this preamble, which is copious given the brevity of his paper, Adair-Dighton continues with the description of his cases. Among the four generations of patients composed of nine subjects presenting blue sclerotics, deafness is reported in one patient only, a young mother aged 26 years with blue sclerotics and a history of fractures of both legs caused by insignificant traumas. Furthermore "nerve deafness" occurred in this young woman three months after the birth of her child.

No other details are given, but this anonymous woman could claim to be the first to join the list of people suffering from deafness in conjunction with osteogenesis imperfecta.

Some remarks may be made about this "historical" deafness given current knowledge.

The age at which this type of deafness appears is quite surprising since the perceptive forms of deafness generally occur later in life.

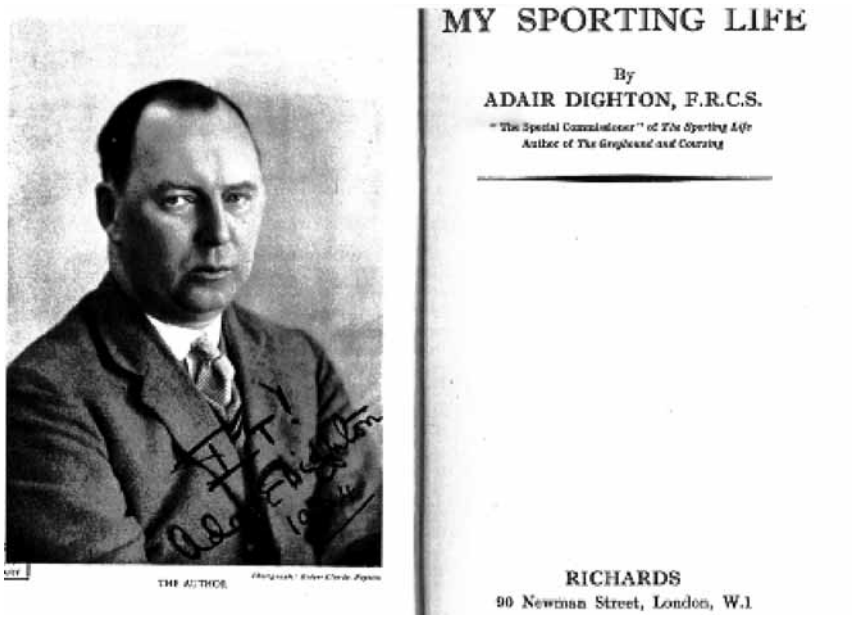


Figure 24
Here we see Charles Allen Adair-Dighton in 1934 at the age of 49 (disregard the initials F.R.C.S.). This is the frontispiece of his book written as a sports journalist, in which he gives a brief account of his life and some anecdotes about his medical career.
"*My Sporting Life* is very jocular in tone, full of anecdotes and humour. There are also some passages which, I'm sorry to say, display a less agreeable aspect of his character. He refers to black people in quite racist terms for example. There is little in the book about his medical career and I certainly would not have guessed from what he had written that his name had been given to a surgical procedure!"
[From a personal communication with Mr. Paul Webster, Librarian of the Central Library of the City of Liverpool].

Only one case of deafness in nine blue sclerotics associated with O.I. would seem to be a very small number, but we have to consider the low number of patients examined and also the fact that deafness does not usually appear in O.I. before the ages of 20, 30 or even 40 years. Of Adair-Dighton's nine patients, five were under the age of 20.

• The two sides of Charles Allen Adair-Dighton: between Medicine and Sports Journalism (Figures 24,25A,25B,25C)

1. The Ophthalmic and Aural surgeon

Adair-Dighton was a specialist who practised ophthalmology and ENT. He was born in Liverpool in 1885. When his paper was published in 1912, he was Hon*. Assistant surgeon at the Eye and Ear Infirmary, Myrtle Street, Liverpool; Hon. Ophthalmic and Aural Surgeon, Toxteth Infirmary, Liverpool and Hon. Ophthalmic and Aural Surgeon, Birkenhead and Wirral** sick children's Hospital.

• The Eye and Ear Infirmary, a joint entity independent of the General Hospitals in the 19th century.

At this time, even ophthalmology had to work hard to gain a place in general hospitals despite its supe-

* Honourable, and not honorary. It means that he was appointed.
** Two towns near Liverpool

Figure 25A
The Eye and Ear Infirmary in Myrtle Street, Liverpool, in 1906. The building was built in the 1870s, functioned as the Eye and Ear Infirmary from 1881 onwards, and closed in 1978! In 1948, the Ophthalmologic department was relocated and only the ENT Service remained there until the closure. Today, the building has been converted into a block of flats called "The Symphony" but its red-brick façade in typical Victorian style has been preserved and its appearance has scarcely changed.

Figure 25B
Toxteth Infirmary, Liverpool, in 1900

Figure 25C
Birkenhead and Wirral children's hospital in 1900. The special Ophthalmic and Aural Department was opened here in 1909, and Adair-Dighton acted as surgeon from 1911 onwards. In 1914 or 1917 (there is a gap in the annual reports of the hospital at this period), Adair-Dighton was called up for military service and it was found impossible to maintain the department, that was temporarily closed down. In 1918, Adair-Dighton resumed his duties (see below for the reasons). A new Hon. Ophthalmic and Aural surgeon was then appointed.
[From Emma Challinor, Archivist, Wirral Archives Service].

rior status to otology, particularly in the United States.

- For example, an "Eye Infirmary" was founded in New York in 1820. Diseases of the ear were also treated there but the ear was not mentioned in the title of the institution! The title was officially modified in 1864 only, to the "Eye and Ear Infirmary". Even later, in 1873, a department of throat diseases was added without the name being changed. People had to wait until 1896, in other words more than 70 years, to have two distinct departments of ophthalmology and otology.

- In Liverpool, one of the hospitals where Adair-Dighton practised, "the Eye and Ear Infirmary", was first founded in 1820, under the name of "Ophthalmic Infirmary" by the same practitioner who founded "the Ear Institution" elsewhere in Liverpool in 1839. Shortly thereafter, both health care institutions merged administratively and this was all the easier because they were directed by the same physician. The new institution became "the Eye and Ear Infirmary". In 1870, a shared new building was built in Myrtle Street (Figure 25A) and it operated until 1978. However, ophthalmology and otology were split up after 1948, when the ophthalmology department was relocated.

So during the 19th century, people thought that lack of knowledge about the eye and the ear on the one hand, and perhaps their non-vital nature on the other, meant that they did not merit consideration as separate entities or a place in a general hospital.

However, in the early 20th century, things changed. The Medical Inspection of School Children, for instance, decided to form a special Ophthalmic and Aural Department in the Birkenhead and Wirral children's hospital to deal with the numerous cases of this

kind among children (Figure 25C). The special department was opened in 1909 inside the hospital.

From another point of view, the integration between two services such as Ophthalmology and Otology certainly favoured the discovery of Adair-Dighton because, as he practised both disciplines, he was ideally positioned to notice the association between blue sclerotics and deafness.

• An anecdote concerning the hospital told by Adair-Dighton himself

"One of theses amusing stories concerned a patient of his, a Mrs Tripp, who had been rescued from 'the Great Abyss' (Metaphor used for Death), by a rather wonderful operation in the brain".

["Post and Mercury, June 7, 1934; Memories of Liverpool"]

"Admittedly rather proud of her recovery, I had a crowd of students from the University down one evening and after telling them her story and the details of her case, was in the middle of dressing her ear and, incidentally, the healing brain cavity, when, presumably, I hurt her a little more than usual, and before the whole assembly, that included a swarm of nurses, she ejaculated: 'Now then, chuck it, Charlie, you're tickling me too much'. Ever afterwards, if I swore at a nurse or cursed the house surgeon, a sort of echo of 'Chuck it, Charlie' reverberated from the background".[22]

2. Medical examination for the Army: the prelude to a personal disaster

When the Great War was declared in 1914, Adair-Dighton was called up and sent to a major base hospital in France. But a purely formal medical examination as part of the recruitment procedure revealed that he was suffering from tuberculosis. He was assigned to home service, forced to abandon his lucrative practice and, after a period as head of the Army ophthalmic department at Blackpool, he was discharged from the Army as unfit for service and given three months to live.

However, contrary to all expectations, he survived! He recovered and, two years later, was "covering" the Waterloo Cup for the *Sporting Chronicle*! It should be pointed out that, since his childhood, he had been a horse-racing fan and that his father frequented these circles.

From then on, he began a new life. The most striking consequence was the publication of his book, "My Sporting Life"[23] (Figure 24) when he was 49 years old. Unfortunately, I have lost track of him from that point onwards, despite the enquiries of Mr Paul Webster, librarian of the City of Liverpool, to whom I owe the information presented here on Adair-Dighton's life and whom I thank warmly.

VI. [1963-1999] Michel Petrucciani, a jazz pianist: the courage of a patient suffering from osteogenesis imperfecta[24,25] (Figures 26A,26B,26C)

On the sixth of January 1999, Michel Petrucciani died in Manhattan, New York. He was 36 years old. He died from an infection of the lungs exacerbated by his thoracic deformity. Five years earlier, President Chirac made him a *chevalier de la Légion d'Honneur* in Paris. He was buried in the *Père Lachaise* cemetery. He was born in Orange in 1963. His parents were French but his father was of Italian origin. He was born suffering from the disease and was raised in Montelimar (France). As an adult his height was less than one metre (three feet). His weight was between 25 and 40 kg. (which he felt to be too heavy).

• Childhood

In a brief account in an interview, he said: "I did not really have a very happy childhood: moving from hospital to hospital, from broken legs to broken arms; from orthopaedic plaques to plasters; from dreams to forgotten things, I was shut up at home: my parents were so afraid to let me go outside..."

• Percussion first

He discovered the piano at the age of four, but his father made him play percussion first on a set of instruments made to size, "which allowed me to develop the muscle of my legs as well".

• Then the piano

His hands were of normal size and the muscles of his upper limbs were adequate (probably like Ivar the Viking) to permit Petrucciani to become a famous pianist. Again like Ivar, he first had to be carried on stage to the instrument. However, he later mastered crutches. The pedals of the piano were equipped with extension pieces.

• A failed suicide

He emigrated to the United States, where he was successful. However, he returned to France regularly. He was married but then divorced. He was acquainted with several women and he had several children. One of them suffered from his disease. His busy life, alcohol, some timid experiments

Figure 26A
Michel Petrucciani playing in concert. The pedals of the piano have been adjusted.

Figure 26B
Petrucciani standing up with his crutches, in conversation with violinist Stephane Grapelli with whom he sometimes played.

Figure 26C
Close up, Petrucciani's hands were medium-sized and allowed him to play normally, in spite of his swollen fingers.

Figure 27
The mayor of Paris, Bertrand Delanoë, inaugurates a square bearing the name of "Michel Petrucciani, French composer and jazz pianist", in the 18th *arrondissement*. The son of the pianist, who suffers from the same disease, is present sitting in a wheelchair. This is a strange image showing a child forced to sit because of fate, and an adult squatting by compassion.[25]

with drugs and maybe his emotional life led him to attempt suicide. He tells: "really, I wanted to die. I threw myself down the stairs. I flung my crutches forwards... and I lost consciousness. None of my ribs were broken, even though my bones are fragile. I woke up. I had no injuries. There are small miracles sometimes".[24]

• Deaf?

Like every musician, Petrucciani was probably hard to please when it came to his hearing. The allusions he made to his hearing should therefore not be taken entirely at face value. However, I will quote the declaration he made in an interview when he was 34 years old, two years before his death: "I have some audio cassettes dating from that time (not specified). It is clearly audible; **even with my ears as they are today, that gets through.**"[24]

• An optimistic conclusion

During the same interview, he affirmed: "I look back at the path I have covered. In 1980, I couldn't walk; I weighed 25 kg... In 1997, I am stronger, I walk with crutches, I manage on my own, my health is better, I have learnt things... and even if I have lost some hair, I am more handsome than I was"...[24]

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