
von Recklinghausen's Disease vs. Proteus Syndrome

A Comparative Analysis of "Elephant Man's Disease"

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Abstract

Joseph Carey Merrick, better known as the "Elephant Man," suffered from an extreme condition initially diagnosed as Neurofibromatosis I. Recent research, however, has challenged that view and has attributed his massive disfigurement to the effects of Proteus syndrome, one of the most debilitating and ravaging diseases known to man. An analysis of the data and pathology from Merrick's records, compared with the data and pathology from instances of Proteus, supports this view.

Biography

Joseph Merrick lived in England in the latter half of the nineteenth century. Accounts differ as to the status of his parents during his lifetime. His father left when he was young and either died or worked in London until after his son's death. According to Frederick Treves, Merrick's physician, his mother "basely deserted him when he was very small, so small that his earliest clear memories are of the workhouse to which he had been taken" (Treves, 1923). Montagu, on the other hand, states that Merrick kept a small portrait of his mother with him at all times, and that she came to visit him in the workhouse (Montagu, 1971). Whichever is true, we do know that Merrick had a very isolated, unhappy childhood, especially as his disorder was becoming more and more pronounced.

After the workhouses, Merrick joined a number of traveling circuses, trying to scrape together enough money to live. Here, the taunts and jeers of his childhood continued, until his "sole idea of pleasure was to creep into the dark and hide" (Treves, 1923). Finally, Merrick ended up in London. By a twist of fate, the Mile End freakshow he was a part of was directly opposite the London Hospital, and that was how he met Sir Frederick Treves. When Treves heard word of the presence of the Elephant Man right across the street, he arranged for a private exhibition and recorded Merrick's condition:

There stood revealed the most disgusting specimen of humanity that I have ever seen...[he was] a little man below the average height and made to look shorter by the bowing of his back...From the brow there projected a huge bony mass like a loaf, while from the back of the head hung a bag of spongy, fungous-looking skin...The osseous growth on the forehead almost occluded one eye. The circumference of the head was no less than a man's waist [36 inches].

(Treves, 1923)

He continues by describing the massive exostoses (bone protrusions) on Merrick's upper and lower jaws that so deformed his mouth that he was barely able to speak. Both the front and back

of his body carried the same sort of soft tissue growths as his head. His left arm and hand were normal, but his right limb was massively distorted, where the hand was "more like a fin or paddle than a hand" (Treves, 1923). The right and left legs were as deformed as the right arm had been: "unwieldy, dropsical looking, and grossly misshapen" (Treves, 1923).

Treves gave Merrick a hospital card which was to save his life two years later. The show that Merrick was in was evicted from the store on Mile End, and after a couple of years of trying to obtain permits to show Merrick, the owner of the show decided that his Elephant Man was more of a burden than a money-

maker and sent him back to London with no money and no means of providing for himself. When he arrived, he created such a stir that the police had to be summoned to prevent the crowd from becoming too excited. They discovered his hospital card on him, and Treves was summoned. He came and took Merrick back to the hospital with him, and allowed him to live there, away from his tormentors.

While Merrick was there, Treves was able to further observe him, and he found that the condition did not reduce Merrick's intelligence at all. Rather, "Merrick was highly intelligent... he possessed an acute sensibility" (Treves, 1923). Joseph Merrick died in 1890, apparently resulting from an attempt to sleep in a reclining position. Merrick's head was so massive and unwieldy that he had to sleep with it resting on his knees, and when he laid his head down, "the massive skull was inclined to drop backwards" (Treves, 1923),

causing him to feel as though he were suffocating. It appeared that he attempted this one more time, but was unable to lift his head again, and the weight of his skull dislocated his neck (Montagu, 1971).

Comparative Pathologies

Joseph Merrick's condition was postulated in 1909 to have been caused by von Recklinghausen's disease, or Neurofibromatosis I. Von Recklinghausen's is a disorder that involves "the wild proliferation of the cells derived from these neural tissues [e.g. Schwann cells] with many nerve fibers embedded in their midst together with large numbers of connective tis-



Joseph Merrick

sue cells...that produces the typical disorder of neurofibromatosis” (Montagu, 1971). Lesions (growths or pitting) appear on the skin and in the bone, causing the individual to have a lumpy, deformed appearance. The nerves often reach as far as the osteoblastic cells, which are responsible for laying down new bone. In many cases, this causes exostoses and hyperostoses (overgrowths of bone) to form. The fact that there are consistent growths both in the integument (the dermal layer) and in the bone below lends some weight to the argument that there were growths that affected both the nerve plexi innervating the integument and those in the underlying bone.

One of the unfortunate associations this caused was the labeling of people with von Recklinghausen’s disease as having the “Elephant Man’s Disorder.” It created a stigma and often made it quite difficult for people with neurofibromatosis to live with their disorder for fear that they would one day look the way Merrick did. This diagnosis of Merrick was thought to be true for well over half a century. Not only had it become the premiere case of von Recklinghausen’s, but it had also slipped into everyday language.

In 1986, a new theory was proposed by Cohen and Tibbles. They believed that Merrick’s symptoms were much more consistent with a disorder that had recently been distinguished from von Recklinghausen’s. The new disorder had aptly been named after a Greek god who could transform himself in order to escape his enemies. Proteus syndrome covers a large number of different pathologies. It begins prenatally as a disorder of the mesodermal layer, which eventually forms most of the human muscular and connective tissue. This disruption causes symptoms that include “partial gigantism of the hands and/or feet, hemihypertrophy, macrocephaly,... lymphangiomas, lipomas, and hemangiomas, as well as a number of other anomalies” (Ng, *et al.*, 1997). All of these disorders result in disfiguration and deformation, normally on a massive scale. The disorder was originally considered an extreme form of neurofibromatosis until 1979, when it was discovered to be a new disorder by Cohen and Hayden. In the 1980’s, a group of researchers led by Weidemann created seven diagnostic criteria for Proteus: gigantism of the hands and/or feet; pigmented nevi (large darkened spots on the skin); partial or complete hemihypertrophy (excessive growth on one side of the body); subcutaneous tumors-lipomas, lymphangiomas, and hemangiomas; skull anomalies and exostoses; accelerated growth; and visceral abnormalities (Ng, *et al.*, 1997). It was only a matter of time before researchers began to debate over whether or not Merrick had had von Recklinghausen’s or Proteus syndrome.

Diagnostic Assessments

As is the standard for any diagnosis, one must compare the case at hand to other similar cases, or cases of a specific condition in order to make an accurate judgment of the nature of a disorder. Proteus cases are not very common, with about a dozen or so reported every five or six years (Clark, *et al.*, 1987). Each case, however, shows a massive deformation much like what Joseph Merrick experienced.

In their article in the British Medical Journal, Cohen and Tibbles compared Merrick’s case to that of a young boy afflicted with Proteus syndrome. The boy showed, among other things, the same form of distortion of the integument of the plantar surface,

the extremely enlarged skull, and “soft tissue masses diagnosed by palpation as lipomas... noted on the chest and abdomen” (Tibbles & Cohen, 1986) similar to those Merrick exhibited. There was also “severe craniofacial asymmetry, redundant skin around the nose, and a pointed chin” exhibited in the case by five and a half years of age. The right hand was grossly distorted, much the same way as Merrick’s was. This led them to believe Merrick may not have von Recklinghausen’s, but may indeed have been afflicted with Proteus syndrome. Several other points about Merrick’s pathology did not coincide with that of von Recklinghausen’s. “‘Café-au-lait’ spots [pigmented areas on the abdomen and chest] are present in over 99% of patients with neurofibromatosis,” (Tibbles & Cohen, 1986) yet Merrick exhibited none. Also, physicians have yet to see any deformations due to von Recklinghausen’s that remotely resemble the totality of Merrick’s. “Merrick had many features of the Proteus syndrome --namely, macrocephaly; thickened skin and subcutaneous tissues, particularly of the hands and feet, including plantar hyperplasia [see included photos], lipomas, and unspecified subcutaneous masses; hypertrophy of long bones; and overgrowth of the skull” (Tibbles & Cohen, 1986).

Later cases also show these conditions. The case of a Malay baby born in 1996 with Proteus corroborates the theory Cohen and Tibbles presented. The child had “right facial hemihypertrophy involving the pinna [ear], cheek, upper and lower lips, right half of the tongue and alveolar margin, clearly demarcated at the midline...she had a smaller fluctuant swelling over the left side of her neck with excess nuchal folds” (Ng, *et al.*, 1997). Upon excision of the swellings, it was determined that they were lymphangiomas. The child also had macrodactyly of the feet—her first and second toes were separated by a very wide cleft. Merrick exhibited all of these characteristics.

Another case was that of an 18-month-old Greek boy who exhibited “macrocephaly, partial gigantism of hands and feet, pigmented nevi, and other anomalies compatible with Proteus syndrome” (Malamitsi-Puchner, *et al.*, 1987). They found that the child had hypertrophies of the head and the right leg, lymphangiomas on the trunk, and excessively long and thick fingers with clinodactyly (crossing) of the third and fourth fingers on each hand (Malamitsi-Puchner, *et al.*, 1987). He had all of the common signs of Proteus syndrome, many of the less common, and few of the rare. He also exhibited a pathology much like that of Merrick, only much earlier in life.

Joseph Merrick’s pathology *resembles* each of the individuals who have been diagnosed with Proteus syndrome. However, because he managed to survive until the age of 29 without treatment beyond some simple excisions, it is difficult to compare him to some of the recent cases. His condition was able to progress to such a state as to render him nearly unidentifiable as a human. He could speak, but with great difficulty. He could not be seen in public for several years after his entry into the hospital, and he could not chew or swallow without much trouble. He never experienced the mentally debilitating aspects of Proteus syndrome, however, and Treves acknowledges that he did have an intellect and an imagination that was perfectly normal. In fact, he was an avid theatergoer, and enjoyed having intelligent discussions.



Right Elbow

Merrick's right elbow, viewed from the lateral position (towards the midline of the body). Here, one can see the distortion of the phalanges, especially the third and fourth middle, which caused the irreparable crossing of Merrick's fingers. One may also see the right-angle twisting of the ulnar-humeral articulation at the joint of the elbow, which caused Merrick to be unable to use the arm.



Skeleton

Merrick's skeleton, viewed from the anterior-ventral position (from the front). One may see the totality of the distortion from this view. Merrick's skull is seen in proportion to the rest of his body and the discrepancies between his right (useless) and left (mildly underdeveloped) arms are evident, as is the massive hyperostosis of the right femur. The feet are seen to resemble the hands with respect to distortion.



Face (right)

The right side of the skull is by far the most disfigured of any part of the skeleton. The ex- and hyperostoses are immense, the partial occlusion of the right eye is obvious, and one notes that the articulation of the mandible occurs in the wrong place (posterior to the zygomatic arch).

Face (left)

The left side of the skull serves primarily to show the hemihypertrophic (tending to one side) nature of the disorder. There is some deformation of the mandible and some bony masses exist on the superiormost (uppermost) portion of the cranium, but not in the excess that one sees on the right.

Conclusions

There have been many other occurrences of Proteus syndrome that are similar to the disorder that affected Joseph Merrick. Since Cohen and Tibbles' article, the theory that Proteus syndrome is "Elephant Man disease" has become very widely accepted, but since there are no remaining tissue samples with which to disprove the von Recklinghausen's theory, it remains an unanswerable debate. Even without a definite diagnosis, Merrick's case illustrates the value and the necessity of continual re-examination and questioning of accepted information.

Detailed Osteological Discussion

Merrick suffered not only from huge growths in his cutaneous and subcutaneous regions, but also to the bone beneath. His skull had so many exostoses that it only retained its basic form on the left side, his spine suffered from terrible curvature, and his upper and lower extremities experienced much distortion. By and large, the distortion is on the right side of the body. Merrick's skeleton reveals the terrible course his condition took in ravaging his body.

Joseph Merrick's skull is one of the most extraordinary in the world. In a photograph from the left, we can see the basic form of a human skull, but from the right, it becomes virtually unidentifiable. There are enormous exostoses on the occipital, temporal, parietal, and frontal bones, and slightly smaller ones on all of the external bones of the skull. His mandible has a large, pointed exostosis on the right side pointing inferiorly, and a smaller, rounded one directly above it pointing superiorly. The articulation of his jaw to the skull is normal on the left, while on the right, the articulation appears to be beneath the zygomatic arch, well anterior of the glenoid fossa. The foramen magnum is partially occluded: there is an exostosis on the left side projecting towards the center of the foramen.

Merrick's humerus was a good deal thicker than a normal specimen's, and the articulation of the humerus to the radius and ulna appears to be twisted so that the radius articulates superiorly to the ulna, rather than medially. The coronoid and olecranon processes of the ulna arise from the medial and lateral portions of the ulna, respectively, and articulate superiorly and inferiorly with the humerus. As one can see, this is almost a 180° twist of the head of the ulna. The hand is in its correct position and orientation, but the massive deformation of the metacarpals and phalanges, as well as the growths that existed in life, rendered his hand useless for everything except maintaining balance.

Merrick's spine and rib cage experienced little of the extraordinary exostotic growth seen elsewhere, although there was a curvature to the left due to scoliosis. His scapulae are normal, but the ribs on his right are somewhat stunted and misshapen. His right clavicle is thickened, and his sternum is bent to the right.

Merrick's legs, feet, and pelvic girdle also suffer from this deformative process, and again, we see the effects much more clearly on the right than the left. Merrick had had a disease of the hip when he was a child that essentially obliterated his left acetabulum and femoral head and forced him to rely on a cane for the rest of his life. His right femur shows a slight thickening of the anatomical neck of the femur, but the portion of greatest interest is the lower 2/3 of the bone. There is a major hyperostosis of the

diaphysis that increases as it reaches the epiphysis. The distal end of the femur is larger than normal, and is composed of what appears to be a random growth of bone, rather than an ordered structure. The left femur is only remarkable in that it is twisted inwards about 45 degrees. In both legs, the tibiae and fibulae appear to be unaffected, except for the fact that the right tibia and fibula are markedly shorter (as the right femur is larger). Merrick's feet show the same hyperostotic condition as his hands.

Merrick could not sleep in a normal position due to the convolutions of his body, as Treves accounts:

The attitude that he was compelled to assume [due to the heaviness of his head] was very strange. He sat up in bed with his back supported by pillows, his knees were drawn up, and his arms clasped round his legs, while his head rested on the points of his bent knees.

(Treves, 1923)

The overall tendency for the skeleton to be deformed towards the right is supported by postmortem measurements of bone mass. The bones of Merrick's right side are almost invariably heavier than those of the left (the right fibula is an exception, but it is a good deal shorter than the left). The growths that Merrick experienced during his lifetime also tended to be far more pronounced on the right side.

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