European Journal of Orthopaedic Surgery & Traumatology (2018) 28:1569–1572
https://doi.org/10.1007/s00590-018-2233-8

GENERAL REVIEW • GENERAL ORTHOPAEDICS - DYSOSTOSIS

Pycnodysostosis: the disease of Henri de Toulouse-Lautrec

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Received: 2 April 2018 / Accepted: 12 May 2018 / Published online: 24 May 2018

Abstract
Pycnodysostosis or Maroteaux–Lamy syndrome is a genotypic bone disorder, with autosomal recessive inheritance, individualized by Lamy and Maroteaux in 1962. It is characterized by diffuse condensation of the skeleton with thickening of the cortex and narrowing of the medullary cavity. This condensation is reminiscent of the one observed in Albers-Schönberg disease, which differs essentially in dysmorphism of the skull (no closure of fontanelles, gaping sutures, hypoplasia of the lower jaw with open mandibular angle) and extremities (hypoplasia or osteolysis of the phalanges). The patients have a short stature, short hands and feet, and malformed nails. The first scientifically correct diagnosis was made by Dr. G. Séjournet who, under the guidance of his teacher Professor J.-A. Lièvre, performed extensive research and diagnosed Henri de Toulouse-Lautrec with achondroplasia-related dwarfism. This article describes pycnodysostosis and reports the life of the painter Henri de Toulouse-Lautrec who died from the disease.

Keywords Pycnodysostosis · Henri de Toulouse-Lautrec · Maroteaux–Lamy syndrome

Introduction

A few years previously, the film Moulin Rouge familiarized the general public with the little bearded man named Toulouse-Lautrec, an important and eminent painter of the late nineteenth century. According to the history of medicine, physicians and surgeons from their specialist's point of view commented on the true cause of his small stature; according to some, short stature resulted from two fractures in adolescence that led to growth arrest [1, 2]. However, if the fracture of a long bone, poorly healed, is likely to cause a shortening of the affected limb, it is not considered as a determining factor of dwarfism. Therefore, medical historians discussed the possibility of preexisting osteopathy [1, 2].

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The life of Henri de Toulouse-Lautrec

The genealogy of Henri de Toulouse-Lautrec (Fig. 1) dates back to the time of the Counts of Toulouse, in the twelfth and thirteenth centuries. Henry took the name of the Comte de Chambord, pretender to the throne of France. Such families often married their children so as not to fragment their property—a disastrous solution for the offspring. Henri's father and mother were first cousins. On the maternal side, Henry had many cousins; one of them, Gabriel Tapie of Celeyran (Fig. 2), had been his inseparable friend and his scapegoat. He was studying medicine; he was very tall and the contrast with Henri was striking [3].

Henri was born in Albi on September 24, 1869. Born apparently healthy and normal, very quickly, he was brought to Lamalou-les-Bains for growth disorders. At the age of 14 years, he fell into his grandmother's living room and fractured a femur. The fracture did not heal; nevertheless, he was taken to Barèges, where, unfortunately, he broke his other femur as well. He certainly had congenital bone fragility because the falls that caused the fractures were not of high energy. The associated growth disorders were probably secondary to a hereditary disorder. His thorax grew, but his legs did not. He walked with great difficulty and suffered from recurrent pain. He had to move with a cane. He measured

Fig. 1 Portrait of Henri de Toulouse-Lautrec (1864–1901) as depicted by Paul Sescau in 1894 (Toulouse-Lautrec Museum, Albi, France)

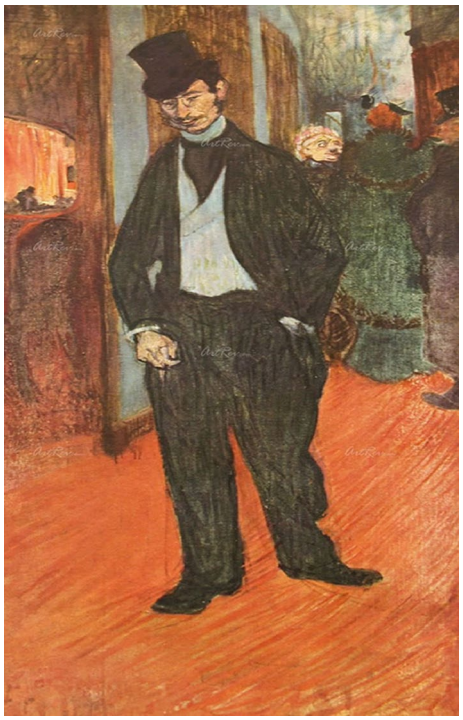

Fig. 2 Portrait of Gabriel Tapié of Celeyran as depicted by Henri de Toulouse-Lautrec in 1894 (Toulouse-Lautrec Museum, Albi, France)

152 cm in height, and he did not participate to athletic activities except for swimming [2, 4].

Handicapped for life, he lived on the edge of his environment keeping a certain distance from hunting, receptions in the castles of Albigensia and such activities of the people of his class. He opted for lower class habits and knew the life of cabarets and the circus of Moulin de la Galette (Moulin Rouge). From 1889 to 1895, he had a very productive and extraordinarily rich period in the choice of his subjects. He came to such an insightful observation in his paintings and so eloquent syntheses in his posters because he carefully sought the means of expression of a particular culture based on Montmartre [4, 5].

He was penciling and drinking for entire nights. In spite of the pain that kept him writhing and shut up, he had heroism and mockery. He was gourmet and expert in culinary art. He sought forgetting in painting and alcoholism: wormwood mixed with cognac [4, 5]. His delusions and visual hallucinations (zoopsies) were frequent. His drawings entitled Le Cirque, made after a detox at the clinic of Dr. Blanche, offer deformations perhaps inspired by a delirium tremens. At night, he went down to wake up his gatekeeper thinking he had heard burglars, and he hunted spiders with a revolver. Alcoholism and untreated syphilis did their work and lead to degradation. He was dirty, with his hands full of crusts. He insulted the employees at the post office when he was going to receive his correspondence. But there was not much time left [6].

Syphilis, a Morbus Gallicus, contracted by a prostitute and treated with mercury, wreaked havoc in Henri's alcoholic body. In the spring of 1899, on a final crisis of delirium tremens, he left the clandé and entered the Parisian clinic of Dr. Blanche to follow a treatment and a rehabilitation program. Throughout the year 1901, his condition worsened. He was weakening day after day. After a crisis of alcohol stupor and paralysis following an embolism, he found refuge with his mother at the Château de Malromé at Gironde. He died on September 9, 1901, at the age of 37 years. His last words were addressed to his mother, for whom he had a profoundly affection. He took her hands in his, looking at her intently, and said "Mom, no one but you, mom," and he died [4, 5].

The disease of Henri de Toulouse-Lautrec: pycnodysostosis or Maroteaux–Lamy syndrome

Thanks to meticulous research by Professor Maurice Lamy and his collaborator Pierre Maroteaux, we now have an accurate postmortem diagnosis of the disease of Henri de

Toulouse-Lautrec. His disease, extremely rare, was pycnodysostosis or Maroteaux–Lamy syndrome. Professor Maurice Lamy was a French pediatrician and geneticist (Amiens 1895–Bordeaux 1975). He studied medicine in Paris. He became a hospital doctor in 1934, head of service at the Children's Hospital in 1943, professor of medical genetics in 1950, and a member of the Academy of Medicine in 1966. He performed research on anemia and leucocytosis in children, in which he emphasized the importance of bone marrow puncture for diagnosis. In genetics, he was interested in twins, the congenital malformations of the skeleton, and the genetic origin of diseases such as diabetes and hemochromatosis [2].

Pycnodysostosis or Maroteaux–Lamy syndrome is a genotypic bone disorder, with autosomal recessive inheritance, individualized by Lamy and Maroteaux in 1962. It is characterized by diffuse condensation of the skeleton with thickening of the cortex and narrowing of the medullary cavity. This condensation is reminiscent of the one observed in Albers-Schönberg disease, which differs essentially in dysmorphism of the skull (no closure of fontanelles, gaping sutures, hypoplasia of the lower jaw with open mandibular angle) and extremities (hypoplasia or osteolysis of the phalanges). The patients have a short stature, short hands and feet, and malformed nails. The first scientifically correct diagnosis was made by Dr. G. Séjournet who, under the guidance of his teacher Professor J.-A. Lièvre, performed extensive research and diagnosed Henri de Toulouse-Lautrec with achondroplasia-related dwarfism. Indeed, he found that up to 13 years the development of the child was perfectly normal; he reached a 147 cm stature at the age of 13 years. Thereafter, the growth rate slowed down sharply, and from age 14 years to 18 years, stature was permanently fixed at 152 cm, an increase in only 5 cm in contrast to approximately 30 cm increase in normal subjects [2, 7].

Comparative studies of the growth of the trunk and the lower limbs, between 14 and 22 years, showed that in a normal subject the trunk only increases by 5 cm, while the lower limbs lengthen from 20 to 25 cm. From this, it can be deduced that in the case of Henri de Toulouse-Lautrec the growth of the lower limbs had completely stopped at the age of 13 years, whereas the trunk continued to increase normally. It was unclear whether stunting had also affected upper limbs from the age of 13 years. However, the examination of the photographs taken after the age of 18 years showed that his arms barely reached the root of the thighs. His niece said that at the age of 5 years, the train controller refused to consider Henri as less than 7 years old and claimed to pay him a full seat ticket [1].

From 1878, everything had changed. There was an almost total loss of growth at that date, but at the same time a progressive deformation of the head and the face took place in the following few years. A photograph taken at the age of 15 years clearly shows a large and deformed head, a slightly amazed nose, and thick lips. At the age of 18 years, Henri described himself as follows: "……Look at this absolutely unattractive twist, this potato nose!……" At the age of 20 years, his niece describes him in these terms: "……head fat, covered with black hairs hiding huge lips and retaining a thick saliva, he goes waddling on his little legs of nabot whose bust emerges disproportionately……" [3, 4]. In the famous painting "Le Bal du Moulin Rouge" (Fig. 3), the artist imagined himself next to his cousin. One can see him in profile; the nose flattened at the root, the nostrils wide open, the lips thick and deformed; the bowler masks the deformity of the head [3]. In a self-portrait with the painter walking next to his cousin, one may clearly notice that Toulouse-Lautrec is just at the level of the armpit of his neighbor, his arms hidden in the sleeves and pockets of an ample overcoat barely reaching the top of the thighs, the legs are ridiculously small, and the famous checkerboard pants are the size of a child's underpants [3]. With respect to this portrait, at the beginning of the twentieth century, Pierre Marie, in a masterly description, fixed in him the features of the dwarf with achondroplasia. The master of the Salpetriere thus describes Lautrec "……of small size, arms and legs of child grafted on an adult trunk, the enormous head, the frontal and parietal bosses disproportionately projecting, the deformed face, the nose flattened at the root wide, at the flared end, the thick lips, the projecting buttocks, undulating at every step. In a playful, joking mood; boastful, he has a high idea of himself, he is polite, helpful, but pushes too far the love of the drink. Extremely salacious, particularly endowed by nature for this kind of jousting, he boasts of his feminine success……" [3, 6].

Fig. 3 Painting "At the Moulin Rouge" as depicted by Henri de Toulouse-Lautrec in 1892 (Art Institute of Chicago)

In these circumstances, it seemed reasonable to establish that, at the age of 13 years, Henri de Toulouse-Lautrec had all the characters of achondroplasia, micromelic dwarfism, cranial and facial deformity. It is not up to the seemingly excessive size of the genitals, common among achondroplases that cannot be found at home since it compared his image at times to that of a coffee maker [1, 3]. The consanguinity of the parents, the near-constancy in their family of cousin marriages for many generations, was in agreement with this hypothesis, because achondroplasia could be the expression of a recessive character [1, 2].

One may wonder why the diagnosis of achondroplasia which, Pierre Marie said, is obvious to those who once saw one of these patients was not mentioned in the books and articles consulted by the painter. Of course, the diagnosis could not be made at the time when Henri de Toulouse-Lautrec lived since the publication of Pierre Marie who described the disease in depth and made it known dates from 1900. In the nineteenth century, dwarfs were often attributed to the heredosyphilis and it is to presume that the doctors preferred to incriminate a trauma at the origin of the infirmity [1, 2, 7]. In addition, most authors describe achondroplasia as a gross malformation easily recognized at birth, with extremely premature growth arrest in case of survival. This is true when it comes to very severe cases occurring in newborns, such as those observed by Parrot in 1878, but there are degrees in achondroplasia, and when we reconstruct the history of adults by interrogation or entourage, one realizes that the signs of achondroplasia cannot manifest until rather late in life [7].

What was the role of the fractures of the femurs? Left femur first in May 1878 at the age of 13 years, and right femur 1 year later? The conditions of appearance were as follows: The fall in a staircase is pure invention. The first fracture occurred during a minor trauma, falling from a 30-cm chair; the second occurred during a walk when he rolled into a gully 1 m deep [1, 2]. These fractures did not induce shortening of the limb by malunion, as evidenced by the study of the measures mentioned above. However, they substantially coincided with the onset of the disease. Did these fractures give impetus to dystrophy? There is nothing to be said at present, but the coincidence is disturbing and this point remains mysterious [1, 2].

Epilogue

It is necessary to appreciate the repercussions of his deformity on the life and work of Henri, Count of Toulouse-Lautrec. His dystrophy had a decisive influence on the destiny of the artist. In adolescence, the occurrence of his physical disgrace meant for him the annihilation of all that he could hope for in his life. Henri, eldest son of the Count of Toulouse-Lautrec, heir of the title, was quite naturally destined to perpetuate the traditions of his family. At the age of 18 years, because he could not marry a person of his rank, in full agreement with him, his father passed the title to his elderly sister. From a physical point of view, at the age of 12 years, Henri was passionate about sports, especially riding. From the age of 14 years, his claudication and shortness of his thighs forbid any physical activity. Finally, his relations with his fellows and especially the people of his community fell back; he quickly became alienated, his associates were limited to a small number of friends, and soon, in brothels, he would find an atmosphere where his deformity is admitted. He also presented a very nasty defect of speech, and if we add that he also suffered from a hypertrophy of the genitals, we will agree on the relevance of the nickname "Little Priap." Then, he finds in alcohol a fictitious compensation, a vicious habit which would have led to his death at the age of 37 years. However, he always kept the exact notion of his race and rank. His alcoholism was good company "drinking little but often" we never saw him scandalously drunk. The infirmities with which Henri de Toulouse-Lautrec was affected undoubtedly influenced his vocation as draftsman and painter, and the nature of the work he produced [8, 9].

Compliance with ethical standards

Conflict of interest No benefits have been or will be received from a commercial party related directed or indirectly to the subject matter of this article.

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