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Pycnodysostosis and the making of an artist

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ABSTRACT

Henri de Toulouse-Lautrec, a 19th century artist celebrated for his depictions of the Moulin Rouge and Parisian nightlife, suffered from an unknown disorder. His symptoms were not only rare, but also difficult to determine. Both during his lifetime and following his death potential diagnoses have proved controversial, including the most popularly supported suggestion of pycnodysostosis. Addressing the ongoing debate of Toulouse-Lautrec's diagnosis, this article reconsiders the evidence. It summarises multiple perspectives and draws on more recent medical research, while acknowledging that the available sources are often unreliable. Ultimately, while there may be no definitive solution to the mystery of Toulouse-Lautrec's diagnosis, it is possible to draw one conclusion. Observing its impact on his life and work, it is clear that the condition formed the foundation of Toulouse-Lautrec's artistic career, shaping the way he perceived the world and defining the artworks that are now so widely admired.

1. Introduction

Henri de Toulouse-Lautrec was an artist, a drunk and a self-fashioned bohemian. He is best known for his posters and paintings of cabaret performers in the nightclubs of late 19th century Paris. Picturing dancers of the Moulin Rouge, prostitutes and radicals, his images have come to define our understanding of life on the periphery of French society.

Yet Toulouse-Lautrec is not only renowned for his dissident lifestyle and avant-garde works. Suffering from a rare genetic disorder, he is also known as a medical curiosity. Triggering a debate that has spanned decades and disciplines, his symptoms have been a challenge to identify. However, despite ongoing disagreement and flaws in the evidence, one diagnosis has been in favour. In fact, the acclaimed artist is now so closely associated with a form of dwarfism – pycnodysostosis – that the condition is often described as the 'Toulouse-Lautrec Syndrome'.

Whatever the diagnosis, the title 'Toulouse-Lautrec Syndrome' is apt. For in many ways the artist not only defined the condition, but also the condition defined him. It played a critical role in his life and works. Impacting his character and choices, the disorder shaped the way Toulouse-Lautrec envisioned the world and those around him. The still undiagnosed syndrome was the making of an artist.

2. The short life of Toulouse-Lautrec

Toulouse-Lautrec was born in 1864 to an aristocratic family in the South West of France. His grandmothers were sisters and his parents, Comte Alphonse-Charles de Toulouse-Lautrec and Comtesse Adèle Zoë Céleyran de Toulouse-Lautrec, were first cousins (Frey, 1995c; Sweetman, 1999). Gaining wealth, status and privilege from his parents, it was also from their consanguineous marriage that Toulouse-Lautrec inherited a genetic disorder that left him short statured and crippled.

From a young age it was apparent that Toulouse-Lautrec was not an entirely healthy child. Besides the typical colds and flues extensively described in the letters of his concerned mother, the young artist suffered with severe pains in his legs (Frey, 1995a). At the age of seven he was withdrawn from horse-riding lessons and taken to Lourdes to pray for a cure (Frey, 1995a). Teachers worried that he would be injured while playing in the schoolyard, and thus he was only able to attend school for one year (Frey, 1995a).

By the age of ten, the pains in his legs and thighs worsened. At times he was able to walk with a cane or assisted by a tricycle, while on some occasions he was unable to walk at all. In 1875, in the hope of finding a cure, his mother placed him under the care of a Dr Verrier for 18 months (Leigh, 2013). During his stay with the doctor in Neuilly, he likely underwent painful treatments, including hours of traction every day (Frey, 1995a).

Throughout childhood and adolescence, Toulouse-Lautrec suffered from sinus headaches so painful he would wake up crying. Described by many of his friends as having a distinctive lisp, his lips were enlarged and red, often the cause of slight drooling. He developed a large nose and impaired vision, which required him to wear *pince-nez* (glasses) (Frey, 1995a).

Significantly it was in May 1878 at Alby, and in August 1879 at Barèges, that Toulouse-Lautrec suffered two falls, breaking both his femurs in the process (Herbert, 1972). Only able to walk with the assistance of two canes, and with a waddling gait, it was clear that he would

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be unable to participate in the outdoor pursuits so popular among his aristocratic peers. Excluded from the past-times of hunting or riding, he turned instead to art.

Now at his full height of 1.52 m, with significant mobility issues and visible deformities, a career in art seemed an acceptable solution for Toulouse-Lautrec's future. Employing the help of deaf–mute painter, René Princeteau, the portrait painter Léon Bonnat and then Fernard Cormon, the Toulouse-Lautrec family trained the young boy until he was ready for a formal artistic education in Paris (Leigh, 2013).

In Paris, Toulouse-Lautrec embraced a new world of art, scandal and bohemianism. In 1884, he set up his own studio in Montmartre near the Basilica Sacré Coeur, a neighbourhood renowned for its poverty and illicit activity. Loitering in nightclubs, cafés and galleries, he quickly earned a reputation for his outrageous behaviour. Frequently drunk, he would dress in costumes, while disrupting dance halls and bars (Leigh, 2013).

Despite his health and tumultuous life-style, Toulouse-Lautrec was quick to find fame. Critically acclaimed throughout Paris, in 1889, his work was hung for the grand opening of the Moulin Rouge and for the next ten years he continued to produce paintings, drawings, posters and advertisements to be seen across the city (Frey, 1995c, 1995a; Sweetman, 1999).

However, by 1893, the artist's alcoholism had grown worse. Likely suffering from syphilis and bouts of depression, he lived briefly in several brothels, narrowly escaping brawls and arrest. In 1899 he was institutionalised for six weeks. Two years later he suffered a stroke and died at his mother's house, shortly before his 37th birthday (Frey, 1995c, 1995a).

3. The diagnosis of Toulouse-Lautrec

There has been substantial debate over Toulouse-Lautrec's diagnosis. From surviving documents and images, there is little doubt that the artist suffered with disproportionate dwarfism, with short limbs and a normal trunk length (Fig. 1). However what remains unclear is the cause of this condition, as the precise features of his disease and its underlying pathology have proved difficult to decipher.

During his lifetime, initial diagnoses attributed his symptoms to dampness, nerves, poor nutrition and rheumatism (Frey, 1995a). In the decades following his death, many more suggestions were put forward, including achondroplasia, pseudo-achondroplasia, osteogenesis imperfecta, polyepiphyseal dysplasia and rickets (Seedorf, 1949; Sejournet, 1955; Krabbe, 1956; Levy, 1957).

In 1965, in response to a history of accounts that failed to consider all available evidence, and given inadequate descriptions of Toulouse-Lautrec's disease aetiology, French doctors Maroteaux and Lamy diagnosed the artist with pycnodysostosis (Maroteaux and Lamy, 1965). They had first described this form of autosomal recessive dwarfism in a paper published only three years earlier (Maroteaux and Lamy, 1962).

Pycnodysostosis can be characterised as clinically consisting of: brittle bones resulting in spontaneous fractures, particularly of the legs, feet, jaw and clavicles; short distal phalanges, often resulting in short stubby hands; a short-stature; open fontanelles and sutures; a large beaked nose; chronic respiratory airway infections and obstruction; and maxillofacial features such as micrognathia, mid-facial hypoplasia, enamel hypoplasia, delayed or premature eruption of mature teeth and a grooved palate (Alves and Cantin, 2014; Periera et al., 2008; Mujawar et al., 2009; Ramaiah et al., 2011; Puri et al., 2013).

In accordance with the diagnosis of pycnodysostosis, Toulouse-Lautrec was certainly short in stature and suffered from weak, brittle bones. In his painting *At the Moulin Rouge* (c. 1892–5), the artist appears in the background standing beside his tall, robust cousin, Gabriel Tapié de Céleyran. It seemed that Toulouse-Lautrec enjoyed the comic effect of his size and fragility when directly compared to others. Intentionally picturing himself beside tall, strong and agile people, he light-heartedly exaggerates his own condition.

Fig. 1. Henri de Toulouse-Lautrec aged 26, 1890, photograph, private collection.

In several caricatures of himself, Toulouse-Lautrec also plays upon his large nose and lips. In his self-portrait (Fig. 2), though satirically exaggerated, the artist highlights his protruding features with a bold black line. Combined with reports of his pronounced lisp, prematurely erupting teeth and drooling (Leigh, 2013; Herbert, 1972), such symptoms are compatible with the maxillofacial characteristics of pycnodysostosis (Mujawar et al., 2009; Alves and Cantin, 2014; Sudarshan and Vijayabala, 2012).

We now know that pycnodysostosis is an autosomal recessive disorder caused by a mutation in a gene on chromosome 1q21 encoding for the enzyme cathepsin K, which is important for the normal functioning of osteoclasts and the re-absorption of bone organic matrix (Gelb et al., 1996; Mujawar et al., 2009). Given the fact that his parents were first cousins, it seems highly likely that Toulouse-Lautrec inherited an autosomal recessive bone-related disease. Indeed five of the artist's cousins also suffered from painful skeletal disorders (Frey, 1995c; Maroteaux and Lamy, 1965), and it is possible that some/all of them also were afflicted by the same autosomal recessive disease as Toulouse-Lautrec. Sadly very little is definitively known about the nature of the cousin's pathology, meaning any comparisons with Toulouse-Lautrec are problematic.

However, while the diagnosis of pycnodysostosis came to be initially widely accepted by historians and clinicians, in the last two decades the assessment has received substantial criticism. In 1995 literary historian and academic, Julia Frey, contested the validity of Maroteaux and Lamy's case (Frey, 1995a). Frey claimed that many of the classical features of

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Fig. 2. Self-portrait caricature, Henri de Toulouse-Lautrec, 1885, pen and ink on paper, Musée Toulouse-Lautrec.

pycnodysostosis – stubby hands, open fontanelle and micrognathia – were not present in Toulouse-Lautrec.

Challenging Maroteaux and Lamy's description of the artist's hands as small and square, Frey reconsidered a photograph referenced by the doctors. While they claimed that the image illustrates a small, disproportionate hand, Frey suggests that an unusual perspective distorted it. According to Frey, the photograph displays a normal sized hand, but with the fingers extended backwards. She continued to quote Toulouse-Lautrec's friend and author, Jules Renard, who described the artist's hands as 'bony with widely spaced fingers and semi-circular thumbs' (Frey, 1995b).

However, the quote from Renard does not entirely disprove the diagnosis of pycnodysostosis. In fact, the description of 'semicircular thumbs' could even support the hypothesis, given the description by Mujawar et al. (2009) of a patient with 'short, spoon-shaped digits.' Furthermore, Toulouse-Lautrec's own reference to his hands as 'grosses pattes' [fat paws] (Frey, 1995b), could also be considered as evidence (particularly given his avoidance of the word *grands* (Leigh, 2013)). Thus, while it is impossible to be certain, it seems that there is no substantive proof that the morphology of Toulouse-Lautrec's hands was not consistent with a diagnosis of pycnodysostosis.

A key feature of pycnodysostosis is unclosed fontanelles. According to Maroteaux and Lamy, Toulouse-Lautrec likely suffered from this symptom. They gained their evidence through conversations with French biographer, Henri Perruchot, who had known friends of the artist. They also noted that Toulouse-Lautrec frequently wore a hat to 'protect an imperfectly ossified skull'. Finally, they quoted Sir Terrence Cawthorne, in his address to the Royal Society of Medicine: '…and when he [Toulouse-Lautrec] was five it was noted that his fontanelle had not yet closed' (Cawthorne, 1970). However, according to Frey there is no mention of Toulouse-Lautrec's fontanelle in Perruchot's writings or in the accounts of his friends and family (Frey, 1995b). Cawthorne, she continued, did not provide any references for his claim in 1970.

Certainly Frey casts doubts on the validity of Maroteaux and Lamy's evidence. Yet even if the sources had been exaggerated, or worse, falsified, it would not necessarily counter the diagnosis of pycnodysostosis. For it remains to be said that a minor failure of the fontanelles to close might not be noticed by friends and family, and may not have had an appreciable effect on the shape of the skull.

Finally, in opposition to Maroteaux and Lamy, Frey (1995b) contested the proposition that Toulouse-Lautrec had a receding chin — a significant feature, given Alves and Cantin's (2014) recent review revealing one of the most frequent clinical features of pycnodysostosis is mandibular hypoplasia. Highlighting photographs of Toulouse-Lautrec in his youth, Frey argues that a deformity of the chin is not immediately obvious. In later portraits, Toulouse-Lautrec's thick, black beard disguises the lower part of his face, preventing any definitive conclusion.

Yet in numerous photographs of the artist, micrognathia does seem to be apparent, as his jaw is set back with little definition (Maroteaux and Lamy, 1965; Maroteaux, 1995; Herbert, 1972). Additionally, in his caricatured self-portraits (Figs. 2 and 3), the artist draws attention to his strikingly small chin. Behind protruding lips and enlarged nose, his jaw is depicted with little definition and only a tuft of black hair. While caricatures such as these cannot be relied on for objective scientific information, it is of course their purpose to exaggerate any obvious or differentiating features.

In further support of the diagnosis of pycnodysostosis it seems that there are few alternative bony-related congenital disorders consistent with the artist's symptoms. One condition that could cause unclosed fontanelles together with some of Lautrec's other features is cleidocranial dysostosis. Yet this is an unlikely diagnosis as it presents with normal height (Sudarshan and Vijayabala, 2012) and hypermobile shoulders (Karagüzel et al., 2010). It is also inherited in an autosomal dominant pattern, whereas Toulouse-Lautrec's disease was inherited in an autosomal recessive pattern (Mujawar et al., 2009). Another possibility could be the autosomal recessive craniomandibular dermatodysostosis. Yet this is a cutaneo-skeletal

Fig. 3. Self-portrait caricature, signed 'Lost', Henri de Toulouse-Lautrec, circa 1882, charcoal on paper.

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disorder, involving both the skin and blood vessels, and produces features such as haematemesis that are not apparent in Toulouse-Lautrec (Danks et al., 1974; Pedagogos et al., 1995). Other autosomal recessive conditions that result in brittle bones and short-stature (Stark and Savarirayan, 2009) could also be considered, but their clinical features also do not seem obviously applicable to Toulouse-Lautrec.

It seems that perhaps the only way to definitively diagnose Henri de Toulouse-Lautrec would be to examine the artist's remains. Another option, given the prevalence of painful bony skeletal disorders being reported in 5 of Toulouse-Lautrec's cousins, would be the sequencing of his distant relatives DNA. Having to rely on a range of photographs, caricatures and verbal accounts, rather than the more precise diagnostic tools of radiography or molecular genetics, results in uncertainties. To unearth the pathognomonic feature of acro-osteolysis with sclerosis of the terminal phalanges (Ramaiah et al., 2011), open fontanelles/sutures, and a hypoplastic mandible, would provide long sought after solutions. To carry out genetic testing of a small tissue sample revealing the appropriate alleles on 1q21 would end the debate all together.

For now, while acknowledging the unreliability of our evidence, a diagnosis of pycnodysostosis is the most reasonable conclusion to draw. This diagnosis would be based upon: (a) the shortening of the legs after repeated fractures following minor trauma; (b) the probable micrognathia; (c) the parental consanguinity and its genetic implications and; and (d) the absence of other persuasive diagnoses.

4. The making of an artist

Toulouse-Lautrec tackled the difficulties of his condition head on. Visibly deformed, physically handicapped and marginalised from the lifestyle of his aristocratic class, the artist made his home among the outsiders of Paris. With a group of loyal friends, from artists and intellectuals, to prostitutes and coachmen, it seemed that he did not wish to hide from public view. Instead, he used his art to satirise his deformities and attack any patronising sentimentality directed towards him.

Arguably, if it weren't for his crippling genetic condition, Toulouse-Lautrec would not have been driven to the outskirts of Paris to observe the gritty nightlife around him. He would not have sat and endlessly observed the movements of dancers, or bitingly revealed the daily suffering of prostitutes. In multiple ways the 'Toulouse-Lautrec syndrome' was the making of an artist.

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