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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