

# The Virgin and Child with the Infant St. John the Baptist by Sandro Botticelli (1445-1510): does the child have Pompe Disease?

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## Short Report

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# Abstract

**Purpose:** Current literature describes that art can be used to teach observation skills in medical students. In this way, many medical schools have developed formal observational training on works of art to improve their students' visual diagnostic skills. In this context, this description presents unprecedented evidence that Sandro Botticelli (1445-1510) may have represented a rare neuromuscular disorder, known as Pompe Disease (*accumulation of lysosomal glycogen primarily in the heart, skeletal muscles, and the nervous system*) in one of the characters that make up *Virgin and Child with the Infant St. John the Baptist* (1490-1500).

**Methods:** Observational study.

**Results:** The painting reveals that the Infant Jesus has facial features consistent with some of the main clinical manifestations of Pompe Disease (*poor head control, facial weakness with open mouth posture, tongue protrusion and eyelid ptosis*).

**Conclusion:** These results may indicate that Botticelli in 1500 may have made the first pictorial representation of Pompe's Disease that was only described in the medical literature in 1932. Furthermore, this description demonstrates the importance of the medico-artistic field for the study of any disease during the Renaissance period, which will be essential for the learning process of visual diagnostic.

## Background

The specialized literature asserts that art can be utilized to teach observational skills in medical students, a skill that is integral to patient examination but seldom taught directly within medical curricula [1]. Thus, many medical schools have developed formal art observation training in conjunction with nearby art museums to enhance the visual diagnostic skills of their medical students [2]. In this context, previous studies have pointed out that some important diseases/congenital anomalies existing in antiquity can be perceived through the figurative works of artists who may not have intended to highlight any pathological manifestations. This is especially due to the fact that during the Renaissance, the growing interest in the study of human anatomy led artists to produce increasingly lifelike portrayals. The close collaboration between medical science and artistic skill arose in-part because patrons and viewers expected artworks to be a real representation of what artists observed [3, 4]. Bearing this in mind, the description offered here provides unprecedented evidence that in one of his works the famous Renaissance artist, Sandro Botticelli (1445–1510), may have represented a figure with a rare neuromuscular disorder known as Pompe Disease (glycogen storage disease type II).

## Analysis

Through detailed observation of the work entitled *Virgin and Child with the Infant St. John the Baptist*, painted by Botticelli in 1490-1500, it is possible to perceive that the Infant Jesus is represented with tongue protrusion, bilateral eyelid ptosis and a poor head control (Figure 1). According to the specialized

literature [5-7], which are characteristics typically seen in the clinical presentation of Pompe Disease. Classically, the literature describes that Pompe Disease results from mutations in the gene encoding acid- $\alpha$ -glucosidase (GAA) which is responsible for lysosomal glycogen degradation. Lack of GAA activity results in pathologic glycogen accumulation creating deposits that ultimately disrupt cardiac, skeletal muscle, and central nervous system architecture and function. The most widely accepted classification of Pompe Disease distinguishes between the infantile-onset (classic and non-classic) and late-onset (childhood/juvenile and adult) forms. These forms vary according to clinical presentations, the extent of organ involvement, and GAA activity levels. The recent introduction of newborn screening for Pompe Disease has raised estimates of prevalence to 1 in 8,700 [6, 7]. Patients with classic Infantile-Onset Pompe Disease usually become apparent during their first 2 months of life, typically presenting marked muscular hypotonia together with rapidly progressing muscular weakness. Indeed, muscle weakness and motor delay are the presenting manifestations in 40% of cases. The common neurologic/musculoskeletal manifestations include absent or delayed motor milestones and, in some patients, even deterioration, poor head control, hypotonia, facial weakness with open mouth posture and tongue protrusion, eyelid ptosis, and generalized muscle weakness mostly involving proximal and truncal muscles [5, 6]. In this context, it can be inferred that Botticelli may have used a child with Pompe Disease to represent the Infant Jesus, as the physical characteristics of that figure are in accordance with some of the main clinical manifestations of this disease (*poor head control, facial weakness with open mouth posture, tongue protrusion and eyelid ptosis*).

## Discussion

This description is the first to suggest a possible representation of Pompe Disease by a Renaissance artist. However, it should be noted that many other artists from this period, including Botticelli [8, 9], have also unequivocally portrayed some of their characters evidencing diseases, physical anomalies [3, 4], and neurological signs [10, 11]. Also, we should bear in mind that Renaissance artists did not usually leave detailed descriptions as regards the real intentions behind their works or the representation of the figures [12]. Because of this, any such interpretation is always controversial because the artist's intention to reproduce a deformity should be distinguished from the stylistic feature of the artistic movement of that era [3]. As for the *Virgin and Child with the Infant St. John the Baptist* by Botticelli, although the current literature does not provide any description of the artist's real intentions regarding this work, undeniably the Infant Jesus is clearly distinguished, especially the facial features, from all the other representations [13] of this character produced by Botticelli.

Therefore, although the interpretation that Botticelli incorporated a figure representing the Infant Jesus with Pompe Disease will remain speculative, there is plausible evidence to that effect. Furthermore, it should be noted that the work described here belongs to Botticelli's late period. During which, the literature suggests, the artist emphasized religious themes focusing pain and suffering, especially those related to the Virgin [14]. It would therefore be plausible to suppose that, in seeking to create a highly realistic work, Botticelli may have used a child who in fact had a disease that would have led the mother suffer due to her son's situation, as is narrated in the story of Jesus and the Virgin. This can be seen both in the Virgin's

own melancholic physiognomy, and in the way she brings her face closer to the child's, seemingly expressing care for someone who is really weakened (Figure 1). Thus, it is possible to infer that Botticelli, when representing the *Virgin and Child with the Infant St. John the Baptist* in 1500, may have in fact provided the first pictorial description of a neuromuscular disorder that was historically described in medical literature only in 1932 by Joannes Cassianus Pompe [6].

## Conclusion

As a starting point for future investigations, this description demonstrates the importance of the medico-artistic field for the study of Pompe Disease, and any other disease (or neurological disorder), during the Renaissance. Moreover, this description, at least in part, may in fact explain the artist's real intentions in elaborating this work, which will be useful to many researchers, including medicine students who wish to improve their skills in observing the anatomical details that are fundamental in the learning process of visual diagnostic. "Art whether it manifests as paintings, sculptures, poems, symphonies, or the care of patients, is a uniquely human quality. It signifies the unquenchable human quality of hope. [...] It acknowledges the human need to communicate, to share both the knowledge and the art of living well, to confront together the uniquely human enigma of pain, suffering, and death. [...] Painters and physicians know this well. Observation is not merely data collection such as any computer can record. It is not merely looking, it is seeing. It is not only listening, it is hearing. Its task is to enable the artist to complete what nature has not." M. Therese Southgate (1928-2013)

## Declarations

### Ethics approval and consent to participate

Not applicable.

### Consent for publication

Not applicable.

### Availability of data and materials

Not applicable.

### Competing interests

The author confirm that this report or any of its contents have not been previously published or presented, and that it is not currently under consideration by any other journal. The author also declare that there is no conflict of interest concerning this report or any of its findings, nor are there any financial disclosures required. Patient consent was not required for this description as there are no patient identifiable details or protected health information included within this manuscript.

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## Figures

### Figure 1

(A) *Virgin and Child with the Infant St. John the Baptist* (1490-1500) created by Sandro Botticelli (1445-1510). (B) Note in detail that the Infant Jesus is represented with poor head control, and facial weakness, with an open mouth, a protruding tongue and eyelid ptosis. Tempera on wood [*Diameter: 74 cm*], [São Paulo Museum of Art – São Paulo, Brazil].