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# Ambiguous Genitalia in 46 XY Disorder of Sex Development: a Challenging Case of Partial Androgen Insensitivity Syndrome

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**Abstract:** Ambiguous genitalia, phallus with clitoris-like underdeveloped glans; labia majora-like bifid scrotum; hypoplastic Mullerian duct; primary amenorrhea, gynecomastia in puberty, and assigned as women but a male karyotype is a clinical presentation in 46 XY disorder of sex development (DSD) due to partial androgen insensitivity syndrome (PAIS). We reported a case of ambiguous genitalia with under-masculinized male in PAIS. A case report with a literature review of ambiguous genitalia in 46 XY DSD PAIS at Nusa Tenggara Barat Academic General Hospital. A 14-year-old girl presented with ambiguous genitalia and primary amenorrhea. Physical examination revealed Tanner I breast development with Tanner IV pubic hair development, phallus (2,9 cm) along with external urethra orifice on the superior vestibulum, and hypoplastic major labia. Abdominal CT scan revealed a hypoplastic uterus and an indecisive interpretation between immature ovary or testes on both side where an ovary should be found in normal female. Laboratory result revealed normal testosterone level for male (332 ng/dL), normal estrogen level for female (130 pg/mL), high LH (54,7 mIU/mL), high FSH (>110 mIU/mL), and karyotyping 46 XY chromosome. We suggested comprehensive management to undergo hormonal examination, analysis gonad function, diagnostic laparoscopy for internal genitalia, DNA examination, and also with DSD team that distinguished differentials from 46 XY ovo-testicular disorder, 46 XY mixed gonadal dysgenesis, and 5α-reductase type 2 deficiency. The collaborative multidisciplinary approach with appropriate expertise and communication to the parent, are needed to decision regarding gender assignment and avoiding confronted patient.

**Keywords:** Ambiguous genitalia, 46 XY DSD, PAIS

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## Introduction

Androgen receptor (AR) sensitivity is crucial for the full development of the male phenotype in an XY individual during both the intrauterine and puberty phases of life. A collection of diseases with varying degrees of receptor insensitivity, from minimal to

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complete, is referred to as androgen insensitivity syndromes (AIS).[1,2] A person 46 XY chromosome with minimal androgen insensitivity (MAIS) displays traits associated with men, including gynecomastia, azoospermia, and male sterility. On the other hand, exhibit a phenotypically female appearance, are tall, have well-developed breasts, a vaginal fold that is blind, and have little to no pubic and axillary hair.[2] These people are completely insensitive to androgen.

The most pernicious type of androgen insensitivity is partial androgen insensitivity syndrome, or PAIS, which can cause diagnostic confusion. It is the range's midway. Although these patients have an XY karyotype, they have variable genitalia at birth, reduced spermatogenesis in otherwise normal testes, nonexistent or primitive Mullerian structures,[3] normal or elevated synthesis of testosterone, normal or elevated synthesis of pituitary luteinizing hormone,[1] or impaired activity of genital skin fibroblasts in binding androgen.[4] Partial androgen insensitivity syndrome, or PAIS, is estimated to affect between one in 130,000 and one in 20,400 XY newborn.[5]

It is estimated that one occurrence of complete androgen insensitivity syndrome (CAIS) occurs in every 20,000 to 64,000 male babies; the frequency of partial androgen insensitivity syndrome (PAIS) is unknown.[6] There would be differences in rates for moderate, mild, and total AIS. Studies indicate that the frequency of CAIS varies between 2:100,000 and 5:100,000.[7] A ten-year assessment carried out in the Netherlands using reported cases of AIS showed that the lowest incidence was 1:99,000.[8] PAIS and CAIS are similar in frequency. The incidence of PAIS is estimated to be 1 in 130,000.[9] The current prevalence of MAIS is unknown. However, it is recorded significantly less frequently than CAIS and PAIS.

## Case Presentation

A 14-year-old girl presented to Nusa Tenggara Barat Provincial Academic General Hospital with a chief complain of having no menstrual cycle since birth. In addition, the patient complained of ambiguous genitalia, which she had only recently been aware of a year before the current appointment. The patient had a girl's upbringing. No such complaint was found regarding her forebears. A physical examination revealed tanner III pubic hair and tanner I breast **(Figure 1)**. The both major labia were found to be hypoplastic, while the both minor labia were determined to be intact. The external genitalia were discovered to be a phallus (2,9 cm) with a urethra meatus on the superior vestibulum **(Figure 2)**. Due to the patient's upbringing as a female and the religious expectation of virginity, vaginal toucher examination was not performed. A differential diagnosis of testicular hypoplasia was made based on the findings of pelvic ultrasonography, which also revealed underdeveloped ovaries on both sides **(Figure 3)**. An MRI of the pelvis revealed a hypoplastic uterus with normal cervix and vaginal length, and both sides of the ovary were normal **(Figure 4)**. Testosterone level was found to be 332 ng/dL, estrogen 130pg/mL, LH 54,7 mIU/mL, FSH >110 mIU/mL, and karyotyping resulted in 46 XY chromosome. The findings of 46 XY Ovo-testicular Disorder and 5 Alfa Reductase Inhibitor Deficiency led to the final diagnosis of partial androgen insensitivity syndrome (PAIS) in the patient. A team of pediatrician, gynecologist, andrologist, and psychiatrist was collaboratively managing this case.

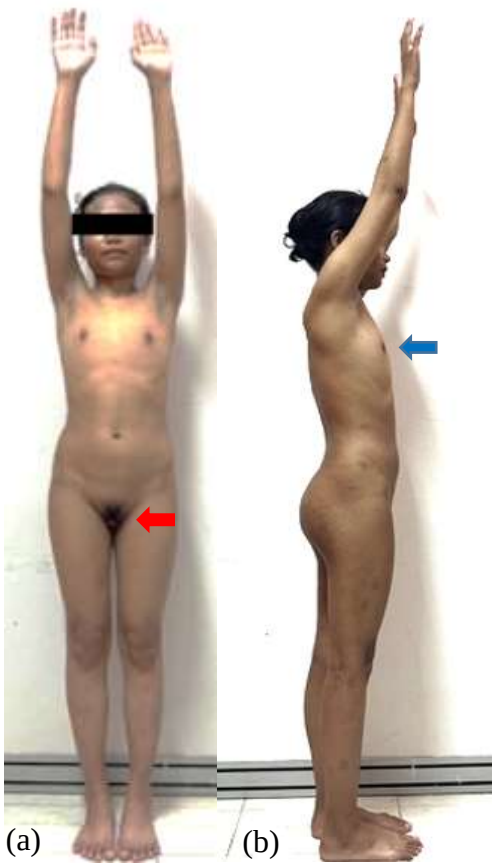


**Figure 1.** Tanner classification; (a) Tanner III Pubic (red arrow) and (b) Tanner I breast (blue arrow)

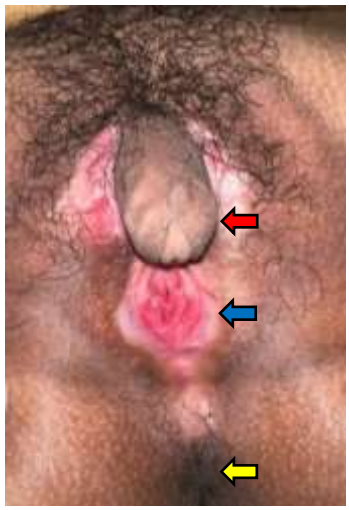
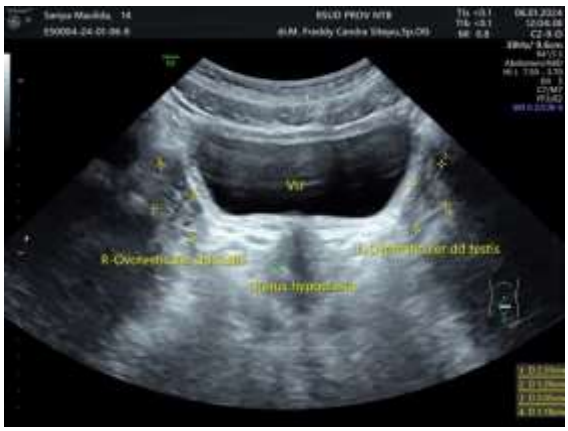


**Figure 2.** External genitalia revealed micropenis (red arrow), vulva with hypoplastic major labia (blue arrow), anus (yellow arrow)



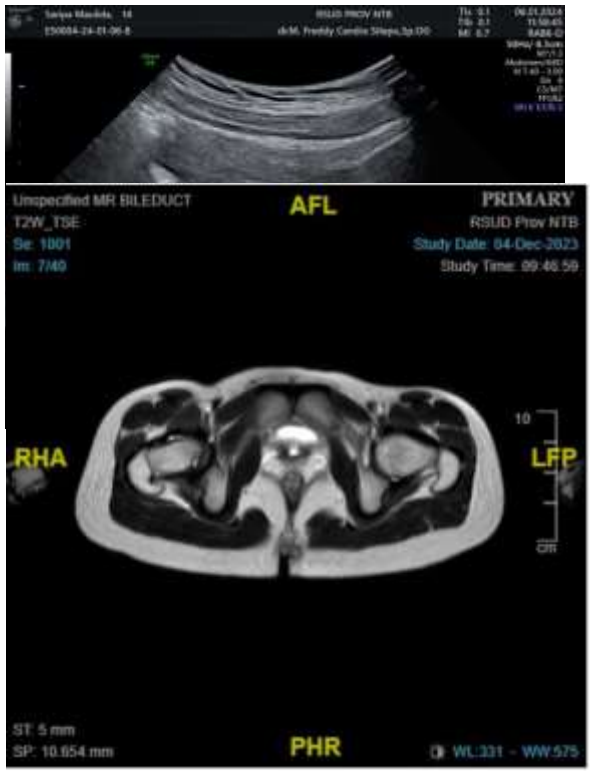



**Figure 3.** Transabdominal ultrasound reveals hypoplastic Mullerian duct; (a) axial view: Ovo-testiculer or testis and hypoplastic uterus; (b) sagittal view: hypoplastic uterus and vagina

**Figure 4.** Abdominal MRI reveals hypoplastic uterus (red arrow)

## Methods

### Search strategy

The Preferred Reporting System for Systematic Reviews and Meta-analyses (PRISMA) statement serves as the foundation for this systematic review of a case report.[10,11] We explored PubMed and ScienceDirect up to May 8th, 2024 using the following keywords or terms: ("partial androgen insensitivity syndrome" OR Androgen insensitivity syndrome OR Testicular feminization) AND "Case report".

### Eligibility criteria

Studies were screened according to the inclusion criteria as follows: 1) Published paper discussing about PAIS, and 2) Case report. Afterwards, exclusion criteria were also set, which include: 1) irretrievable full-text articles. Details of study search strategy are shown in **Figure 5**.

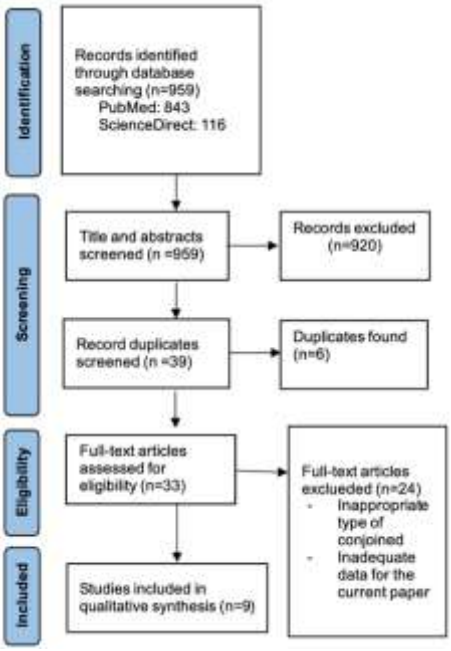



**Figure 5.** Diagram flow of literature search strategy for this systematic review

**Data extraction and quality assessment**

Subsequently, we extracted data from our selected articles, which included author and year of publication, first realization of the chief complain, chief complain that was later become the foundation of PAIS diagnosis, assigned gender at birth, physical features (external genitalia, testis, vagina, uterus, breast, body contour, pubic hair), hormone profile (testosterone, luteinizing hormone, follicle-stimulating hormone, estrogen, anti-Mullerian hormone), karyotype, radiological studies along with other diagnostic modality, therapy given and it's follow up **(Table 1)**.

**Table 1.** Systematic review of case report result

| Author | Year of publication | First realization | Chief complain | Assigned gender at birth | Physical features | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | Genitalia | | | | Uterus | Breast | Body contour | Pubic/axillary hair |
| | | | | | External genitalia | Testis | | Vagina | | | | |
| | | | | | | Right | Left | | | | | |
| **Mohan, CS et al.**[5] | 2011 | 4 years old | Ambiguous genitalia | Female | Micropenis (2,5cm) with penoscrotal hypospadias | No testis palpable | No testis palpable | 1 inch, ended blindly | Non palpable | Tanner III | Male | Tanner IV |
| **Zavaleta, MJC et al.**[12] | 2021 | 9 years old | Masculine behaviour | Female | Micropenis (0,5cm) with perineal hypospadias | Palpable at the level of inguinal canal | Palpable at the level of inguinal canal | Normal length | Non palpable | Tanner I | Male | Tanner I |
| **Turan, V et al.**[13] | 2010 | 20 years old | Ambiguous genitalia | Female | Clitoromegaly (4,6cm) with perineal hypospadias | Palpable in the labioscrotal fold | Palpable in the left inguinal canal (Cryptorchidism) | Blind vagina | Non palpable | Tanner I | Male | Tanner I |
| **Dung, PTV et al.**[14] | 2021 | 44 years old | Ambiguous genitalia | Female | Micropenis (4,5cm) with hypospadias | Palpable and looks like labia majora | Palpable and looks like labia majora | Blind vagina (1cm in length and 5 mm in width) | Non palpable | Tanner III | Female | Tanner III |
| **Sezgin, T et al.**[15] | 2019 | 68 years old | Male type hair distribution | Female | Penis (6cm) with hypospadias | Palpable and looks like labia majora | Palpable and looks like labia majora | Blind vagina (3 cm in length) | Non palpable | Tanner I | Male | Tanner V |
| **Finney, EL et al.**[16] | 2019 | 6 months old | Ambiguous genitalia | Female | Clitoromegaly with urogenital sinus | Palpable in the distal inguinal canal | Palpable in the distal inguinal canal | Blind vagina | Non palpable | Not mentioned | Female | Not mentioned |
| **Bhangoo, A et al.**[17] | 2010 | 14 years old | Micropenis | Male | Micropenis (2cm) | Within normal limit | Within normal limit | No vagina | Non palpable | Tanner I | Male | Tanner III |
| | | 3 years old | Micropenis | Male | Micropenis (2,5cm) | Within normal limit | Within normal limit | No vagina | Non palpable | - | Male | Tanner I |
| **Fulare, S et al.**[18] | 2020 | 17 years old | Bilateral inguinal swelling | Female | Micropenis | Palpable in the distal inguinal canal | Palpable in the distal inguinal canal | Blind vagina (2cm) | Non palpable | Tanner III | Female | Tanner III |
| **Vaidyanathan, P et al.**[19] | 2018 | 17 years old | Gynecomastia | Male | Penis (8cm) | Within normal limit | Within normal limit | No vagina | Non palpable | Tanner III | Male | Tanner III |

| Author | Hormone profile | | | | | Kariotype | Radiological studies | Other diagnostic modality | | Therapy | Follow up | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | Testosterone | Luteneizing hormone (LH) | Follicle-stimulating hormone (FSH) | Estrogen | Anti-Mullerian Hormone (AMH) | | | Modality | Result | | Follow up duration | Outcome |
| **Mohan, CS et al.**[5] | 109 ng/dL | 19,6 mIU/mL | 26,3mIU/mL | - | | 46XY | Hypoplastic uterus with no gonads (USG) | Diagnostic laparoscopy with histopathological sampling | Leydig cell proliferation with atrophic seminiferous fallopian tubules | Gonadectomy followed by Hormone replacement therapy | 6 weeks | Decreased testosterone level (3,8ng/dL) |
| **Zavaleta, MJC et al.**[12] | 20,8 ng/dL | 0,77 mIU/mL | 2,13 mIU/mL | | 55 ng/mL | 46XY | Testes in the rudimentary bags at the level of inguinal canal and the absence of uterus (USG) | - | - | Orchidopexy, phalloplasty, scrotoplasty, and hormonal therapy (Intramuscular testosterone) | 6 months | Appearance of pubic hair and penis growth |
| **Turan, V et al.**[13] | 10,3ng/dL | 13,92 mIU/mL | 36,01 mIU/mL | - | | 46XY | The absence of uterus and adnexa (USG and MRI) | - | - | Penoscrotal hypospadias reparation | - | - |
| **Dung, PTV et al.**[14] | 71.8 ng/dL | not mentioned | Not mentioned | - | | 46XY | The absence of ovary, fallopian tube, uterus, and superior part of the vagina, prostate, and seminal vesicle (MRI) | - | - | Breast augmentation, Gonadectomy, penectomy, clitoroplasty, labioplasty | 8 months | Normal sex function |
| **Sezgin, T et al.**[15] | 388,9ng/dL | 28,2 mIU/mL | 76,1 mIU/mL | 56,4 pg/mL | | 46XY | Bilateral testis under the labia majora with left varicocele (USG); Evidence of prostate and seminal vesicle and the absence of uterus and fallopian tube (MRI) | - | - | Psychological support | - | - |
| **Finney, EL et al.**[16] | 134 ng/dL | 1,4 mIU/mL | 2,5 mIU/mL | <2 pg/mL | | 46XY | Bilateral inguinal testes with no obvious uterus (USG) | | | Bilateral inguinal gonadectomy with gonadal tissue cryopreservation | 3 month | Decreased testosterone level (<12 ng/dL) |

| | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | followed by hormonal therapy | | |
| **Bhangoo, A et al.**[17] | 440.9 ng/dL | 0,72 mIU/mL | 3,9 mIU/mL | - | 20 ng/dL | 46XY | - | | | Testosterone replacement therapy | | |
| | **658.7 ng/dL** | **0,10 mIU/mL** | **0,6 mIU/mL** | **-** | **-** | **-** | **-** | **-** | **-** | **Testosterone replacement therapy** | **3 month** | **Increasing penile length to normal limit** |
| **Fulare, S et al.**[18] | Elevated | Upper limit | Upper limit | Within normal limit | - | - | Vagina, uterine body, ovaries, and cervix not visualized, suggestive of agenesis. Testis in distal inguinal canal (MRI) | | | Orchidectomy and Hormone replacement therapy | - | **-** |
| **Vaidyanathan, P et al.**[19] | 1660 ng/dL | 14µ/mL | 7,7µ/mL | 64 pg/mL | - | - | | | | Bilateral mastectomy | - | **-** |

## Discussion

### Initial characteristic of the patients

We found a wide variety of the time of first realization ranging from as early as 6 months old in the Finney, EL et al. study up until 68 years old in the Sezgin, et al. study.[15,16] This broad variety of first realizations demonstrates the characteristics of PAIS, a condition that falls between MAIS and CAIS and denotes a broad range of illnesses. The range of primary complaints corroborates that idea as well. While ambiguous genitalia are the primary complaint we identified, there are other complaints ranging widely, including amenorrhea and male behavior. Based on the included studies, we discovered that patients who complained of gynecomastia and phallus had been reared as males, whereas patients who complained of ambiguous genitalia had been raised as females. We discovered a trend where the majority of patients were reared more as females than as males, which emphasizes the fact that PAIS is an "in-between" illness that can present as a wide range of illnesses. Comparatively, our case demonstrated a comparatively early realization at 14 years old, with amenorrhea being a less common major complaint.

### Physical, hormonal, and radiological feature

There are 8 out of 10 patients included in our systematic review showed penis and phallus external genitalia, whereas 2 patients who did not have penis showed clitoromegaly with no patients showing normal female genitalia which is clitoris. Furthermore, nearly all of our patients have a blind ending vagina without palpable uterus, even in the presence of external genitalia such as phallus. Tanner I and Tanner III breast proportions were comparable among the study's subjects. In contrast to tanner I, the pubic hair had a trend toward tanner III. This finding suggests that while pubic hair formation is not substantially dependent on the degree of androgen insensitivity, breast development is, however more specific study is still needed to confirm this notion.

Examining the testes revealed a wide range of results: some patients had no testes perceptible in the groin area or in the inguinal canal, while others had testes that could be felt, and a small number of patients even had normal testes. According to the author, this manifestation serves as the primary gauge for the degree of androgen insensitivity. Our patient had phallus, a blind-ending vagina, non-palpable uterus, and no palpable testes—physical characteristics that were similar to the tendency identified in our systematic review.

Our case showed an increased hormone profile for all hormone that was tested (testosterone, LH, FSH, and Estrogen) based on the normal level of women. This outcome provides more evidence for the patient's "in-between" status within the androgen insensitivity spectrum. The idea is supported by the systematic review's findings, which shows a wide range of results for every hormone measure.

All of our included studies utilize USG as the main diagnostic tool with several of them also utilize MRI to confirm the diagnosis, similar to our case. The radiological study primarily revealed a hypoplastic uterus with accidental observations of pelvic testes, albeit in varying locations. A prostate and seminal vesicle were also visible on radiological examinations in multiple studies.

### Therapy

All of the included studies utilize a hormonal therapy as the basis of therapy for their patients, for the exception of patient with an advanced age. Based on the patient's and their family's counseling results, the preferred hormone therapy was determined for the patient's expected gender at maturity. Although the patient and her family in our case have not made a decision yet, our goal, as advised by the multidisciplinary expert, was to administer hormone therapy based on the patient's gender preference. After their patients have received hormonal therapy for several months, a few of the included studies also follow up with them. Following hormone therapy, every patient responds well and achieves their desired outcome.

The goal of PAIS surgery and hormone therapy is to achieve the predetermined gender assignment. According to the most recent guidelines, a patient who was reared as a female should have estrogen therapy and gonadectomy.[20] Conversely, if the patient was reared as a man, then orchidopexy and testicle preservation should be performed in addition to androgen hormone therapy.[21] The effectiveness of long-term testosterone therapy in male-raised PAIS patients is unclear. Androgen treatment may have a major effect on individuals with specific missense mutations in the DNA-binding domain of the androgen receptor. A recent study[22] supported the difficulty of accurately predicting the efficacy of androgen therapy.

Patients diagnosed with PAIS, in contrast to CAIS, typically have ambiguous genitalia at birth. This necessitates a comprehensive conversation with family and careers before making a decision regarding sex assignment. The doctor can then handle early management concerns after that.

For newborns allocated or choosing to be male, androgen supplementation is a part of the medical therapy during puberty. Surgical therapy includes

correction of hypospadias and undescended testes.[23] These procedures are best done in the second or third year of life. Gynecomastia can develop at any point throughout puberty, and in order to prevent tumor growth, it should be treated with a reduction mammoplasty. However, breast cancer incidence is not high in men with PAIS.

When a patient is designated as a girl, the treatment of PAIS involves gonadectomy and genitoplasty prior to the onset of puberty through laparoscopy, as well as estrogen supplementation at that time.[24] The true necessity of gonadectomy in patients who chose to be female is still up for debate. On the one hand, the syndrome is linked to an increased risk of testicular germ cell tumors (TGCT), so testicular cancer can be prevented by removing the gonads.[25] On the other hand, waiting until at least puberty to remove the gonads allows for spontaneous pubertal development because the retained testes produce estradiol, which is derived from the peripheral aromatization of testosterone.[26]

## Conclusion

The collaborative multidisciplinary approach with appropriate expertise and communication to the parent, are needed to decision regarding gender assignment and avoiding confronted patient. Our case is a 14 years old patient who was raised as a girl with amenorrhea and micropenis which later diagnosed as PAIS. Our systematic review showed a similar pattern to our case which illustrate the spectrum of disease of PAIS.

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## Contributions of Authors

**Gede Made Punarbawa** contributed to the co-ordination and design of the manuscript project and overall strategy.

**Muhammad Freddy Candra Sitepu** contributed to corresponding author, conceptualization, writing, editing, and revising the manuscript.

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**I Made Putra Juliawan** contributed to reviewing the manuscript.

**Cok Erly Merlin** contributed to help in writing the manuscript, collating information regarding medical record, and getting informed consent from our patient.

The authors declare that there are no conflicts of interest.

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