

Delayed Diagnosis of Disorder of Sexual Development-Impact on Quality of Life

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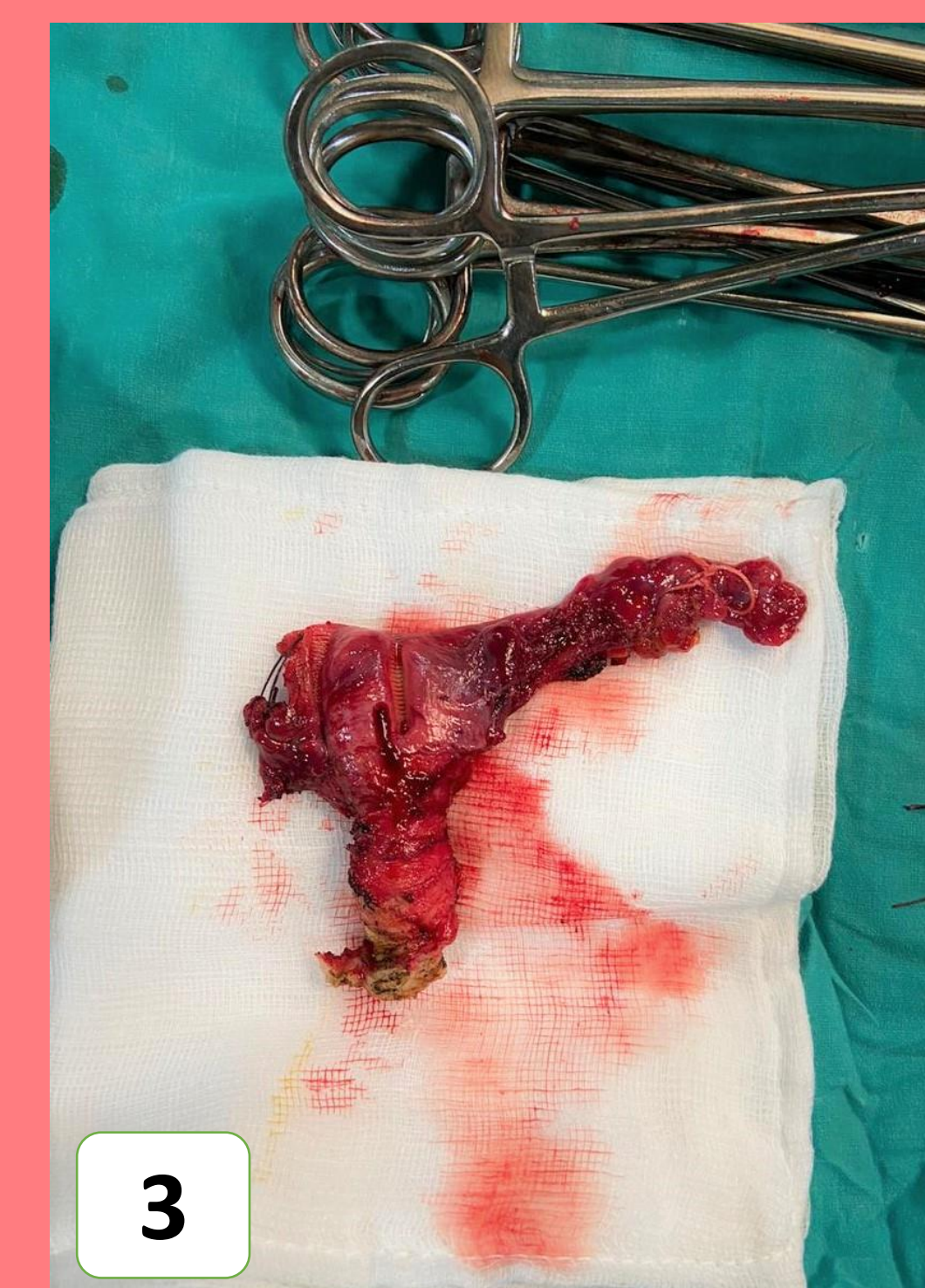
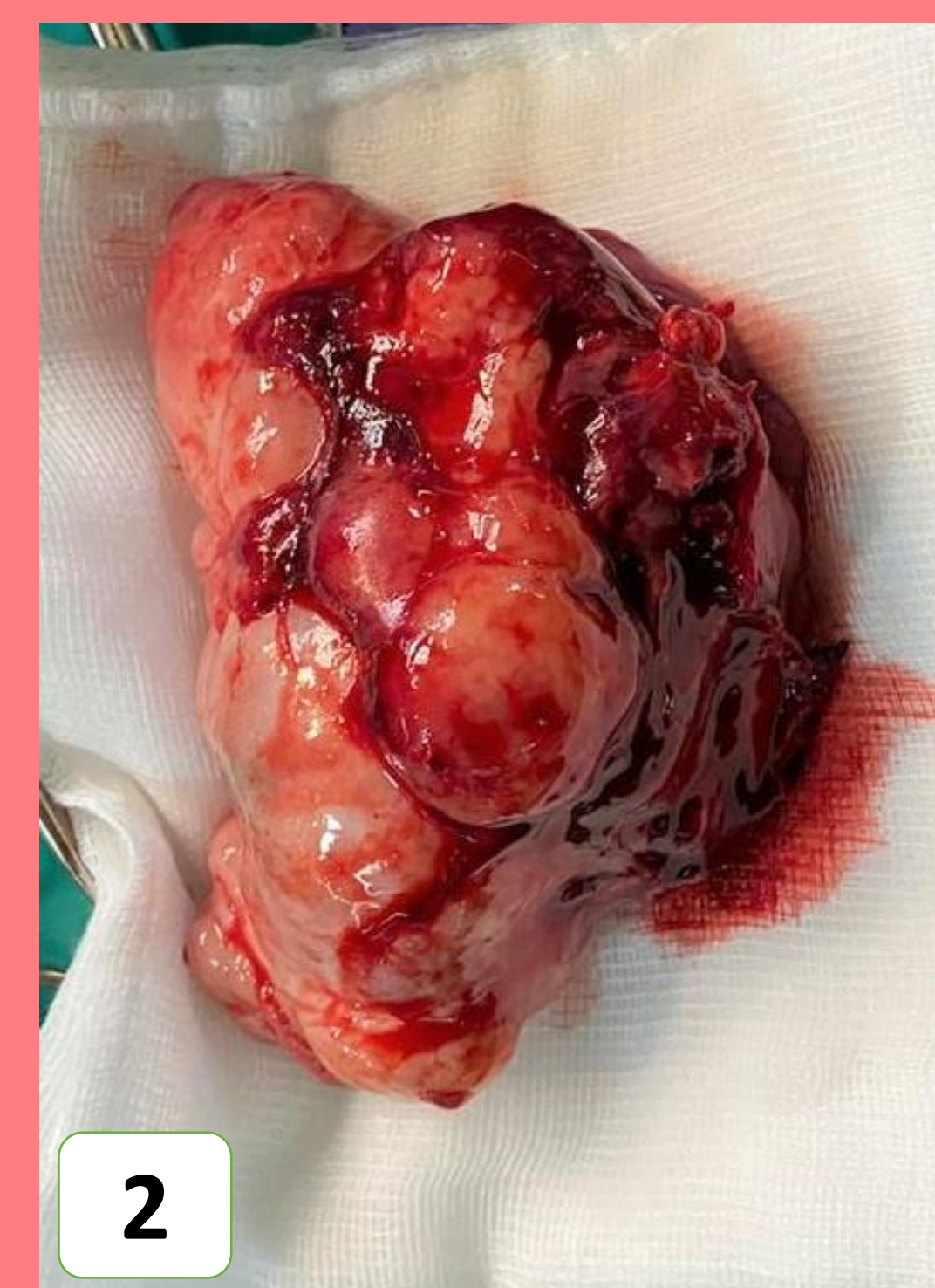
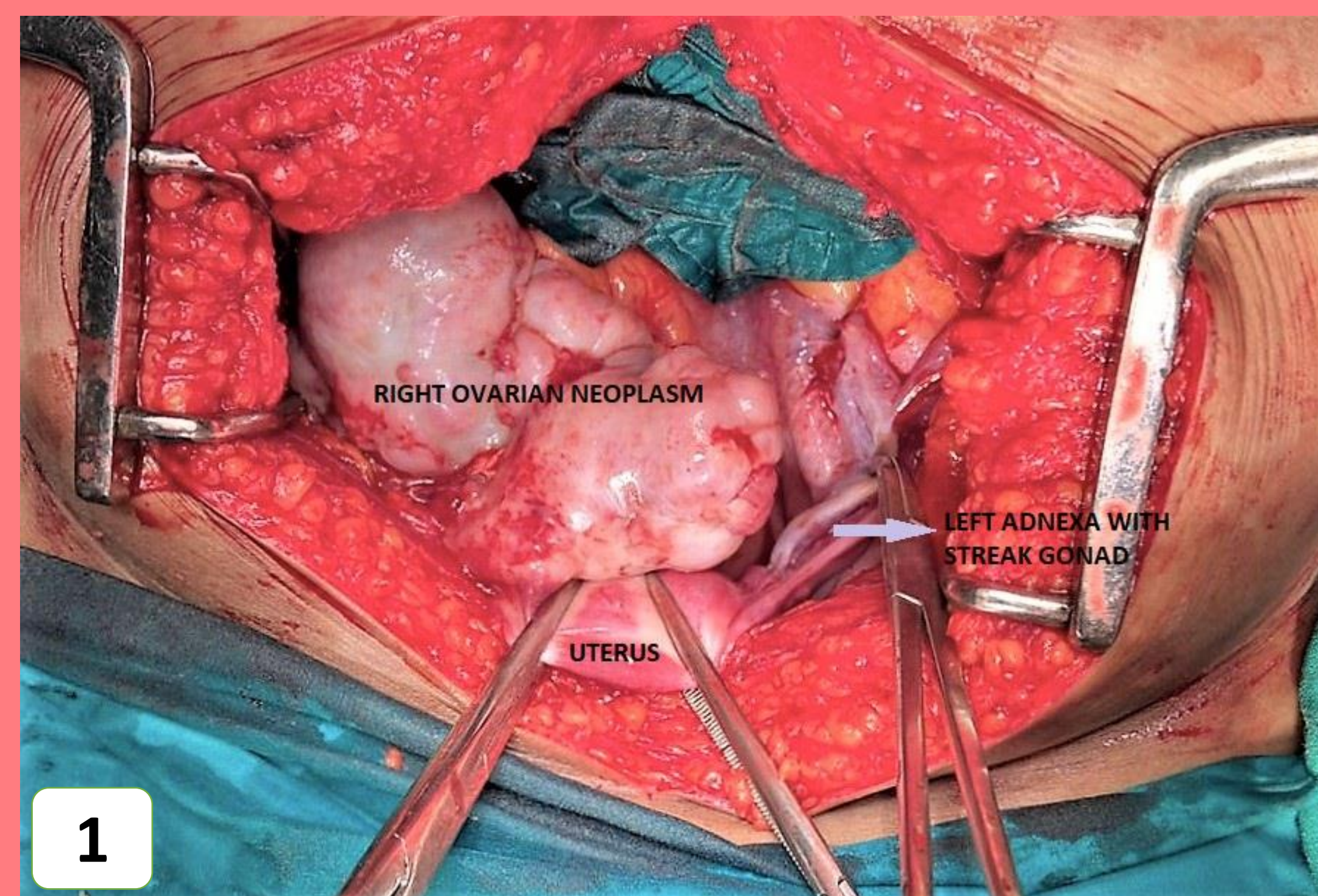
Objective

Disorders of Sexual Development (DSD) is many a time diagnosed later in life.

Disclosing that one's genetic sex differs from phenotype and sex of rearing can cause distress with personal and social repercussions.

This case highlights the emotional turmoil and breakdown of marital life of an individual diagnosed with Swyers Syndrome.

Figure 1. Intra-operative findings, Figure 2. Right sided enlarged gonad, Figure 3. Hypoplastic uterus with left streak gonad



Discussion

The Diagnosis of DSD leads to conflict within the individual as well as family. This is even more so in societies with strictly assigned roles for each gender.

These individuals have reduced quality of life and increased affective distress. Suicide ideation is as high as 20% compared to 9.2% in unaffected population.

Lack of psychological counselling services and awareness of its importance even among the medical fraternity negatively impacts their mental health

Case Report

A lady in her early 20s presented with history of chronic lower abdominal pain. On detailed history taking it was revealed that at the age of seventeen she was evaluated for primary amenorrhoea and ultrasound done then showed presence of uterus and B/L ovaries. She was prescribed Oral Contraceptive Pills and had withdrawal bleeding. However for unknown reasons she stopped the pills after two months and subsequently amenorrhea continued.

She got married at 19 years of age and one year later the couple sought medical opinion as they were unable to conceive. Ultrasound revealed hypoplastic uterus and absence of ovaries. Karyotype was XY. The option of pregnancy with IVF using donor egg was explained, but the couple refused it.

Her husband could not accept a wife with male genes and he deserted her without a lawful divorce. She fell in to depression which was compounded by being single, unemployed and dependent on elderly parents with limited income.

This lead to emotional distress and self neglect. Though the need of gonadectomy was explained, no follow up was done until she presented to us with symptoms. Treatment plan was decided after thorough counselling. She underwent hysterectomy along with Gonadectomy which revealed Dysgerminoma/Seminoma.

Conclusion

1. Any child presenting with primary amenorrhoea should be evaluated based on a **standard evidence based protocol** to ensure correct and timely diagnosis
2. The **impact of DSD** should be understood by the medical fraternity and besides conventional medical/surgical care **must compulsorily include psychosocial services** to both the patient and family. Community social workers involvement would aid in a productive and independent life

References

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