Original Research Paper

Volume - 14 | Issue - 08 | August - 2024 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Paediatrics

"SMALL BUT MIGHTY: UNDERSTANDING MICROPENIS"

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ABSTRACT Isolated micropenis is a rare congenital condition characterized by a penis that is significantly smaller than the normal size for the infant's age, without any associated anatomical or systemic anomalies. This condition presents unique diagnostic and management challenges due to its isolation from other disorders. Although isolated micropenis is a rare and challenging condition, early and accurate diagnosis followed by appropriate hormonal treatment can lead to favorable outcomes. Ongoing monitoring and support are essential to ensure optimal development and quality of life for affected infants. Early identification and appropriate management of isolated micropenis are crucial to optimize developmental outcomes. Treatment often involves hormone therapy, specifically testosterone replacement, aimed at promoting normal penile growth and development. Long-term follow-up is essential to monitor the effectiveness of treatment and to address any potential psychosocial concerns.

KEYWORDS : Isolated Micropenis, Genetic Evaluation Micropenis, Testosterone Replacement Therapy, Newborn Penile Length

INTRODUCTION

Micropenis is a clinical term used to describe an unusually small penis relative to the age and size of the infant, often defined as a penile length that falls more than -2 standard deviations below the mean for the patient's age. While micropenis can be associated with a range of underlying conditions, including genetic syndromes, endocrine disorders, and congenital anomalies, isolated micropenis is characterized by the presence of a small penis without any additional anomalies or systemic conditions.

Isolated micropenis, although rare, presents a unique diagnostic and management challenge. It is essential to differentiate it from other causes of penile underdevelopment, which may include congenital adrenal hyperplasia, Klinefelter syndrome, or hypothalamic-pituitary-gonadal axis dysfunction. Accurate diagnosis typically involves a comprehensive evaluation, including hormonal assays, genetic testing, and imaging studies, to exclude secondary causes and confirm the diagnosis. The etiology of isolated micropenis often relates to conditions affecting testosterone production or action during critical periods of fetal development. Hypogonadotropic hypogonadism, where there is insufficient stimulation of the testes due to inadequate levels of luteinizing hormone (LH) and follicle-stimulating hormone (FSH), is a common underlying mechanism.

Case Report

A Baby born via elective LSCS at gestational age of 38W+4D, to non-consanguineous parents, was noted to have a small penis. Baby cried immediately after birth, umbilical cord clamped and cut 2A+1V present, NG tube measured and passed to check for choanal and anal patency both patent. APGAR at 1min-8/10 , 5 min- 9/10. Baby started on breast feeding sucking and feeding good.

Vitals at birth- Heart rate-136/min, Respiratory rate- 49/min, Temp-36.6F, SpO2-97% on RA.

Anthropometry at birth- Length-48cm, Weight-2.727kg, head circumference- 34cm, chest circumference- 32cm. Penile length was measured- Relaxed length- 1.1 cm, stretched length- 1.3cm which was significantly below -2SD for his age. The penis appeared proportionately smaller compared to other infants, but the scrotum was well-formed, and both testes were descended and palpable.

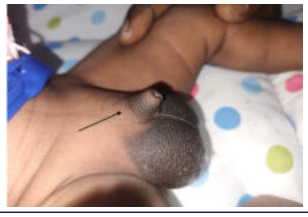

Image 1- Penile Length < -2SD

Image 2- Rugged Scrotum with both testis palpable

The pregnancy was normal, with no significant maternal illnesses or exposures. Ultrasounds during pregnancy did not reveal any anomalies. No significant family history of congenital anomalies, endocrine disorders, or genetic syndromes.

Diagnostic Workup was done which included variety of investigations

- **Complete blood count, LFT, RFT** – Within normal range
- **Thyroid function test** – Normal
- **Bilirubin** - Normal
- **Hormonal Evaluation:**
- Serum testosterone: Low (suggestive of hypogonadism)
- Luteinizing hormone (LH): Normal
- Follicle-stimulating hormone (FSH): Normal
- **Genetic Testing:** Karyotype analysis showed a 46,XY chromosomal pattern with no detectable anomalies.
- **Imaging Studies:** Ultrasound of the pelvis and abdomen was normal, showing no structural abnormalities.

The diagnosis of isolated micropenis was made based on the physical examination and the absence of other anomalies or syndromes. Hypogonadotropic hypogonadism was considered due to the low serum testosterone level and normal LH and FSH levels.

The patient was initiated on testosterone replacement therapy, with regular follow-up planned to monitor growth and response to treatment

DISCUSSION

- **Etiology:** Isolated micropenis can be attributed to various causes, including:
- **Hypogonadotropic Hypogonadism:** Reduced testosterone production due to insufficient stimulation from the pituitary gland.
- **Endocrine Dysfunction:** Conditions affecting the hypothalamic-pituitary-gonadal axis may be involved.

Management:

- **Hormone Therapy:** Referral to a pediatric endocrinologist is recommended for managing isolated micropenis. Testosterone replacement therapy was initiated for Baby B to encourage penile growth and development.
- **Follow-Up:** Regular follow-up appointments were scheduled to monitor growth and assess the response to therapy. Adjustments to the treatment plan were made based on clinical progress.
- **Prognosis:** With appropriate hormonal treatment, the prognosis for isolated micropenis is generally favorable. Early intervention can help achieve more normal penile development and improve future psychosocial outcomes.

CONCLUSION

Isolated micropenis, though rare, is a condition that demands careful evaluation and intervention. This condition, characterized by a penile length significantly below the expected percentiles for age, presents a unique diagnostic challenge as it occurs without any associated anomalies or systemic disorders. Accurate diagnosis relies on a thorough workup, including hormonal evaluations, genetic testing, and imaging studies, to rule out secondary causes and confirm the isolated nature of the condition.

The etiology of isolated micropenis often involves disturbances in testosterone production or action during critical periods of fetal development. Hypogonadotropic hypogonadism is a common underlying factor, necessitating targeted hormonal therapy to promote normal penile growth. Early initiation of testosterone replacement therapy is crucial for improving developmental outcomes and reducing potential psychosocial impacts.

Management of isolated micropenis requires a multidisciplinary approach, including pediatric endocrinology, urology, and regular follow-up care. This ensures effective monitoring of growth, development, and response to treatment, and provides support for any emerging psychosocial issues.

In summary, while isolated micropenis presents significant challenges, early diagnosis and intervention are key to achieving favourable outcomes. Continued research and clinical experience will further refine treatment strategies and improve our understanding of this condition. Ensuring timely and comprehensive care can lead to improved developmental trajectories and overall quality of life for affected infants.

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