

Introduction

Rapid-onset obesity with hypoventilation, hypothalamic dysfunction, and autonomic dysregulation and neuroendocrine tumours (ROHHADNET) is a rare disorder which present in early childhood. Underlying pathogenesis is unclear and many patients succumb to respiratory failure or sudden death. Survivors will have sociocognitive defects. Autoimmune and paraneoplastic mechanisms have been suggested for ROHHADNET syndrome. Approximately 20-30 % of patients will have neural crest tumours.

Case Presentation

Four year old girl was referred to endocrinology unit with a history of excessive weight gain. Parents noticed a rapid weight gain from 3 years of age with increase food seeking behavior and daytime somnolence.

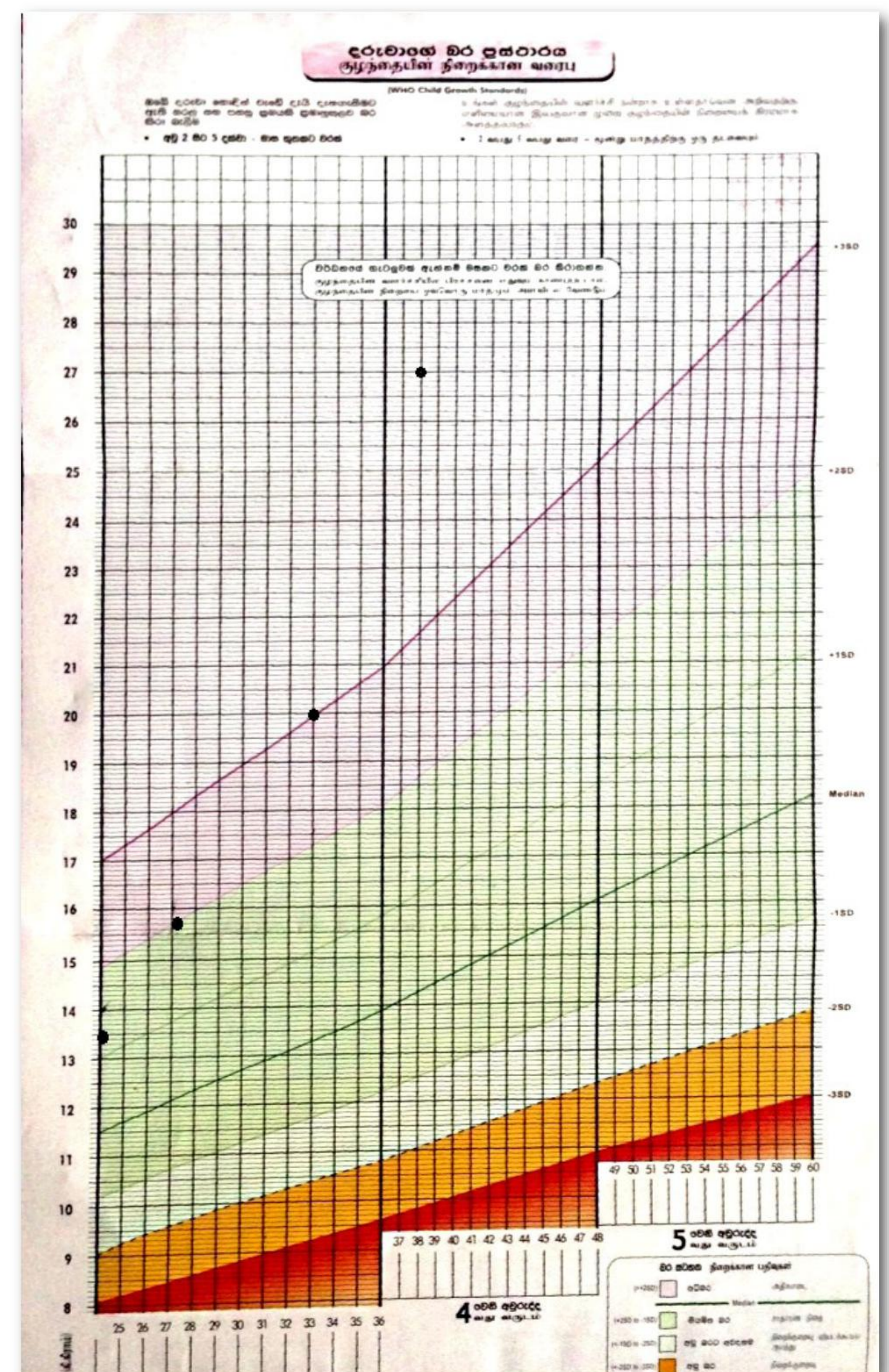
Her weight was >95 th centile and her height was on 25 th centile. Bone age was 1 year and 6 months at the chronological age of 4 years 2 months. Ultra-sound scan of the abdomen revealed a mixed echogenic lesion in right suprarenal area. Right kidney was 8.1 cm and left kidney was 7.6 cm.

She had normal endocrine work up prior to surgery. Urinary Vanillylmandelic acid levels were within normal range. Her blood pressure was normal.

She underwent tumour excision and histology confirmed ganglioneuroma. During surgery she had excessive bleeding. Post operatively she was found to have hypertension. Subsequent ultrasound scan revealed small kidney on right side and dimercaptosuccinic acid (DMSA) scan revealed a nonfunctioning right kidney most likely due to ischemic infarction of the right kidney during surgery.

She was admitted with repeated episodes chest infections due to hypoventilation and frequent febrile episodes without any focus of infection. Child's blood pressure was controlled with two anti hypertensives.

One of the episode of respiratory tract infection required assisted ventilation. She was ventilator dependent for 6 weeks and succumbed after 18 months of the diagnosis.



Conclusion

Child was under close follow up for developing endocrinopathies. Hypoventilation resulted in repeated infections and child needed oxygen and antibiotics during such episodes. Overall she had a poor quality of life. She developed hypertension as a surgical complication. She succumbed to a severe respiratory tract infection

References

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