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Case report: Congenital central hypoventilation syndrome (CCHS)

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Introduction: CCHS is a rare disorder found in less than one per 100,000 new-borns. It affects the central and autonomous nervous system which controls many of the autonomic function in the body. The underlying cause is dominant mutation in the PHOX2B gene. The mutations are stable in transmission from generation to generation but penetrance and phenotype can still vary significantly.

Case Report: A new-born was born in good condition following induction of labour for polyhydramnios at term. She developed respiratory distress at 4 hours of age needing intubation and ventilation at local hospital. She failed extubation once at local hospital and twice at tertiary care with marked CO2 retention during sleep. Her elder sibling of 4 years is being investigated for obstructive sleep apnoea and awaiting adenotonsillectomy. Rest of the history and examination were normal except poor antigravity movements during first few days of life. CCHS was confirmed by genetic testing for PHOX2B gene mutation after excluding metabolic disorders and spinal muscular atrophy (SMA). She was switched to non-invasive ventilation and transferred to long term ventilation (LTV) center specialized for tracheostomy and long-term home ventilation. We avoided suxamethonium during intubation. Family was referred for genetic counseling and professionals involved in the care of elder sibling were made aware of this diagnosis after parental consent.

Conclusion: This case illustrates undiagnosed mild cases may be still at large and the importance of avoiding drugs causing prolonged neuromuscular blockade such as suxamethonium, atracurium, gentamicin etc. in hypotonia until spinal muscular disorders are excluded, to prevent unwarranted hyperkalemia; maintaining high index of suspicion in mildly symptomatic cases to avoid associated long-term morbidity and mortality and to permit early genetic testing and counseling for families; and referring to the local LTV specialist before a final decision to progress down the LTV pathway.

Biography

M Ranjan has completed MBBS and Postgraduate Master's degree in Sri Lanka and Membership of the Royal College of Pediatrics and Child Health in UK. She is currently working as Specialty Trainee Doctor at ST7 level in Yorkshire and Humber deanery rotation in United Kingdom.

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