Abstract

Introduction: We examine a presumptive case of complete androgen insensitivity syndrome (CAIS) with certain unusual features. Clinical Picture: A woman with early onset osteoporosis gave a history of primary amenorrhoea and surgery for intraabdominal gonads. She subsequently defaulted follow-up and hormone replacement therapy. Endocrinological evaluation revealed hypergonadotrophic hypogonadism associated with a 46,XY karyotype. Treatment: Therapy included reinforcement of the female phenotype and oestrogen replacement. Outcome: There was gradual development of her secondary sexual characteristics and improvement in her bone mineral density. Conclusion: Patients with CAIS need proper counselling and education according to their psychosexual make-up and sociocultural factors. The importance of long-term oestrogen replacement in a young subject post-gonadectomy cannot be overemphasised as illustrated in our case.


Keywords: Androgen insensitivity syndrome, Atypical presentation, Male pseudohermaphroditism, Management, Testicular feminisation syndrome