INTRODUCTION

▪ CHARGE is an autosomal-dominant syndrome which includes a variable combination of coloboma of the eye, heart malformations, atresia of the choanae, retardation of growth and development, and genital and ear abnormalities
▪ CHARGE syndrome has rarely been associated with anterior pituitary dysfunction and with structural abnormalities of the pituitary gland only twice.
▪ We report the case of a child with CHARGE association and congenital hypopituitarism due to structural abnormalities of the pituitary gland

CASE PRESENTATION

▪ The patient was a boy born with IUGR (birth weight 2020g, 37 weeks’ gestation)
▪ Clinical features included
  ▪ retinal coloboma and microphthalmia,
  ▪ choanal atresia, dysplastic auricles with small accessory auricle,
  ▪ multicystic dysplastic kidney and hypospadias with cryptorchidism.
▪ Endocrine testing revealed central hypothyroidism and secondary hypoadrenalism. There was inadequate response to low-dose intravenous Synacthen stimulation, with serum cortisol peaking at 10.3 μg/dl at 1 hour. He was started on thyroxine and hydrocortisone replacement.
▪ Because of severe growth impairment by the age of 3.4 years (HSDS: −4.71, HVSDS: −3.62), growth hormone secretion was evaluated. A severe GHD was detected (peak GH 1.56 ng/ml in both tests) and rhGH therapy was initiated. MRI revealed ectopic posterior pituitary

CONCLUSION

▪ We describe the case of a boy fulfilling criteria of CHARGE association (Blakes 1998, Verloes 2005) presenting with multiple anterior pituitary hormone deficiencies and structural pituitary abnormality.
▪ To our knowledge this is the 3rd case in the literature where congenital hypopituitarism in CHARGE syndrome is associated with pituitary structural abnormalities and especially ectopic posterior pituitary