# A rare case of Congenital Hyperinsulinism associated with Hypopituitarism due to Pituitary Stalk Interruption Syndrome

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## Introduction

Congenital hyperinsulinism (CHI) is a rare genetic disorder that is characterised by persistent hypoglycaemia in infants and children. We are reporting a rare case of diffuse CHI who was also found to have hypopituitarism and several other congenital anomalies. A similar association has not been reported in literature.

## Case Report

A female baby was born at 42 weeks gestation with a birth weight of 4.185Kg (1.72SDS). She suffered shoulder dystocia and was ventilated for 12days.

She had persistent

- **Hypotension**
- **Hyponatremia and**
- **Hypoglycaemia**

It was found that she had low ACTH and cortisol levels, subsequently she failed short Synacthen test. She also had hypothyroidism and low growth hormone.

The glucose requirement was up to 20mg/kg/min

CHI was managed by

- high concentration dextrose infusion and intravenous glucagon.

Subsequently

- Diazoxide was started; but she developed cardiac failure,

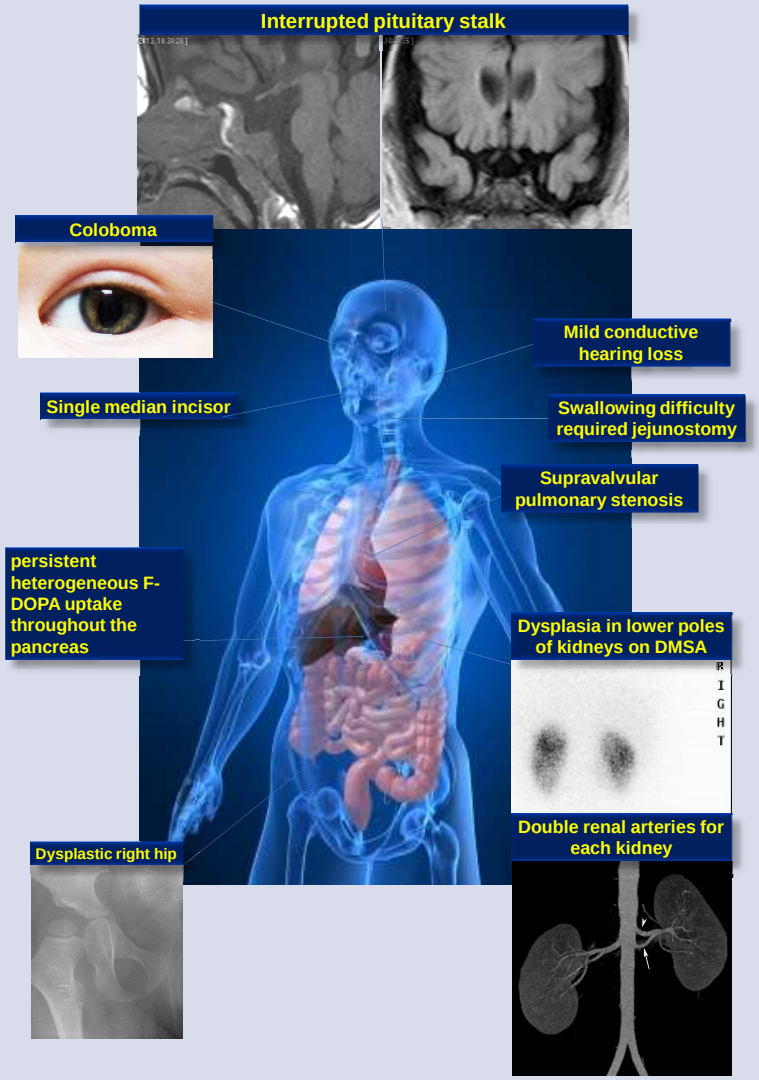
Therefore replaced by

- Subcutaneous Octreotide injections. This was later discontinued due to liver dysfunction.

She is currently 3 years old settled on continuous feeds, hydrocortisone, Levothyroxine and growth hormone.

Table1.Investigations confirming hypoglycaemia and Hypopituitarism

| **Investigations** | Results | Normal range |
|---|---|---|
| ***Hypoglycaemia screen (age 13 days)*** | | |
| Glucose (mmol/l) | 2.5 | 3.5 - 7 |
| Insulin (pmol/l) | 90 | < 13.8 |
| C-peptide (pmol/l) | 712 | 190 - 990 |
| Free fatty acides (umol/l) | 50.1 | |
| 3-β-hydroxybutyrate (umol/l) | < 100 | |
| Lactate (mmol/l) | 1 | 0.7 - 2.1 |
| Ammonia (umol/L) | 51 | 0 – 70 |
| Cortisol (nmol/l) | 120<br>< 50 | 140 - 500 |
| Growth hormone (ug/L) | < 0.05 | > 7.7 |
| Free T4 (pmol/l) | 5.3 | 10 – 30 |
| TSH (mu/l) | < 0.03 | 0.3 – 3.8 |




## Conclusion

We report a rare association of diffuse persistent CHI and hypopituitarism in a patient with several other associated anomalies with probably an unidentified genetic aetiology.