Congenital Hyperinsulinism in association with Poland Syndrome and chromosome 10p11p13 duplication

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Background

- Poland syndrome (PS) is characterized by unilateral absence or hypoplasia of the pectoralis muscle, most frequently involving the sternocostal portion of the pectoralis major muscle, and a variable degree of ipsilateral hand and digit anomalies, including symbrachydactyly.
- Congenital Hyperinsulinism (CHI) is the result of unregulated insulin secretion from the pancreatic β-cells leading to severe hypoglycaemia.
- We report a baby with Poland's syndrome and 10p11p13 duplication in association with CHI, which has not been previously described in the literature.

Case

- Baby girl, born at term by emergency C-section due to abnormal CTG to non-consanguineous parents with a birth weight of 2kg (-3.3SDS) in good condition.
- Recurrent hypoglycaemic episodes since birth.
- Absence of pectoralis major on the left side of the chest.

Investigations

- Inappropriately high plasma insulin (77pmol/l) and low plasma free fatty acids (447µmol/l) and β-hydroxy butyrate (<29µmol/l) during hypoglycaemia (blood glucose 1.0mmol/l) confirming a diagnosis of CHI.
- X-ray and MRI imaging of the spine revealed the absence of left upper thoracic ribs, sprengel deformity of the left scapula and myelomeningocele at the cervico thoracic junction of the spine.
- MRI brain did not reveal any abnormality
- Microarray revealed duplication in the 10p11p13 region.

Management

- Commenced on diazoxide to which she was responsive.
- Currently 6 months old and her hyperinsulinism is well controlled on diazoxide.

Conclusion

- CHI can be an associated feature of several genetic syndromes.
- This is the first reported case of CHI in association with Poland Syndrome and 10p11p13 duplication.
- The genetic mechanism(s) in this syndrome that leads to dysregulated insulin secretion is unclear.

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Hypoglycaemia

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