Poor weight gain in Prader-Willi Syndrome – not always over-restriction consider Coeliac Disease

Lateva M[1], Kassim A[1], Meade C[1], Maher R[1], McCrann A[1], Roche E[1,2]

The Dept of Paediatric Endocrinology and Diabetes, National Children's Hospital, Tallaght University Hospital[1] and Trinity College Dublin, The University of Dublin[2]

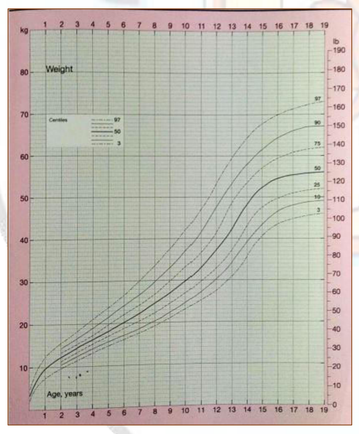
Introduction

Prader-Willi Syndrome (PWS) is a complex genetic condition associated with feeding difficulties, hypotonia, developmental delay in infancy; hyperphagia leading to extreme obesity, growth failure and behavioral problems in childhood (1).
Coeliac disease (CD), is an autoimmune disease characterized by gluten intolerance and a variety of symptoms most commonly diarrhea or constipation and failure to thrive (2). CD is common in the Irish population (3).
In PWS the challenge is to optimize growth while avoiding obesity. Dietary over-restriction can result from the careful regulation of food intake by families.
We present two children with an alternative cause of poor growth, to our knowledge not previously reported.

Case presentation

Case 1:

- a girl (3y 8m) born 2 kg, with poor feeding, diagnosed with genetically confirmed PWS (UPD) in neonatal period.

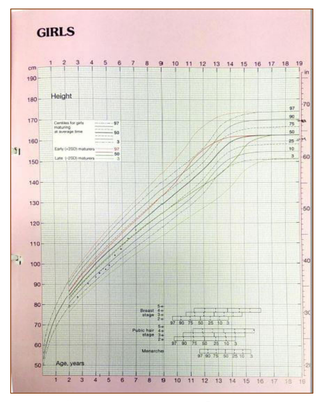


- she demonstrated poor weight gain, but no GI symptoms.
- no family history of CD.
- serum tTG was positive at 41 U/ml (0-6.99) with positive endomysial antibodies, the diagnosis of CD was histologically confirmed following intestinal biopsy.
- GH started 3 months prior the diagnosis of CD

Commencement of a Gluten Free Diet (GFD) was associated with a marked improvement in mood and general wellbeing.

Case 2:

- a girl (7y) with poor weight gain, who required NG tube feeding (until 7m of age), diagnosed with genetically confirmed PWS.



- GH commenced at 5 years of age.
- On recent review despite a good appetite, weight gain was poor and abdominal swelling was noted. There were no other GI symptoms.
- no family history of CD.
- investigation revealed an elevated tTG 124 U/ml (0.6-6.99) and positive endomysial antibodies, the diagnosis of CD was confirmed histologically.

Following commencement of GFD, the abdominal swelling resolved.

Conclusion

Other than poor weight gain, these patients were relatively asymptomatic and did not display the classic GI symptoms of CD. Poor weight gain in PWS is generally considered to be due to parental dietary over-restriction. However we believe that coeliac disease should be actively excluded in those children. We are currently monitoring for CD in our PWS patients.

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contact details: mina_pl@yahoo.com

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Growth and syndromes (to include Turner syndrome)

M Lateva

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