INTRODUCTION

• Prader-Willi syndrome (PWS) is a complex, multisystem disorder first described in 1956.
• This disorder occurs at a frequency of 1/10,000–1/20,000 births and is the leading genetic cause of marked obesity[1].
• Hypotonia, developmental delay, short stature, small extremities, characteristic facies, hyperphagia, obesity, hypogonadism, obstructive sleep apnea, and other behavioral problems characterize PWS.

• The three genetic forms of PWS:
  Although every case of Prader-Willi syndrome is due to the baby failing to receive active genes from a specific section of the father’s chromosome 15, there are three different ways that this can happen:
  ✓ Paternal deletion: 70% cases
  ✓ Maternal uniparental disomy (UPD): 25%
  ✓ Imprinting defect: less than 5%

• When a deletion of chromosome 15q11-q13 region is found on the mother’s chromosome 15, the result is an entirely different syndrome called Angelman syndrome (AS).
• All persons suspected of having PWS should be tested with a DNA methylation analysis.

CASE SERIES

We report thirteen cases of PWS (4 females, 9 males) in the age group of 1-18 years, being treated at Sir Ganga Ram Hospital, a tertiary care center in Northern India.

• 9 children (69%) were diagnosed during infancy, two between 3-5 years and two at 9 years of age.
• Eight out of nine (89%) boys had cryptorchidism; in females, one (25%) had hypoplastic labia minora & majora.
• All children in this study have some degree of cognitive impairment and behavioral issues.
• 12 children are obese and one is overweight.
• 46% (6 children) developed dyslipidemia.
• Two boys and two girls developed hypogonadism.
• Polysomnography was performed in five children, two had moderate degree of Obstructive Sleep Apnea-Hypopnea syndrome, mild variety in the other two while one had a normal study.
• One had oclocutaneous albinism, three developed hypothyroidism, one developed scoliosis and one developed Type 2 Diabetes.
• DNA methylation analysis revealed hypermethylated SNRPN gene in all children (but it doesn’t differentiate the three genetic forms)
• FISH was performed in four of them, 15q11.2-q13 region deletion confirmed in three while one had uniparental disomy (confirmed by DNA polymorphism analysis)

DISCUSSION

• Four children were started on growth hormone (GH) replacement at 9-12 years of age for growth hormone deficiency after performing polysomnography. Three showed significant improvement in height SDS and two showed reduction in BMI SDS. Case 4 failed to show any response in terms of height or BMI on GH therapy. Case 3 expired due to unknown reason after stopping GH.

<table>
<thead>
<tr>
<th>Case</th>
<th>Height SDS (onset of GH)</th>
<th>Height SDS (end of GH)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>-0.394</td>
<td>+2.314</td>
</tr>
<tr>
<td>2 (still on GH)</td>
<td>-2.045</td>
<td>+2.075</td>
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<tr>
<td>3</td>
<td>-1.794</td>
<td>+2.127</td>
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<td>4</td>
<td>-3.323</td>
<td>-3.013</td>
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<table>
<thead>
<tr>
<th>Case</th>
<th>BMI SDS (onset of GH)</th>
<th>BMI SDS (end of GH)</th>
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</thead>
<tbody>
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<td>1</td>
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<tr>
<td>2 (still on GH)</td>
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<td>3</td>
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• Earlier diagnosis, allowing for earlier access to developmental resources, Recombinant human growth hormone (hGH) therapy, and anticipatory guidance, has significantly improved the long-term health and developmental outcomes of children with PWS [2].

References