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Diabetes Mellitus and Retinitis Pigmentosa

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CASE REPORT

Background:

Although the occurrence of diabetes mellitus and retinitis pigmentosa are independently common, four rare syndromes have been reported describing this combination. These include Bardet-Biedl syndrome, Alström syndrome, Kearns-Sayre syndrome, and Wolfram syndrome. Diagnosis can be done clinically based on other manifestations. Molecular genetic testing may be needed for accurate diagnosis and genetic counselling. We describe two cases that presented with diabetes mellitus and retinitis pigmentosa along with other manifestations.

Case 1 summary:

A 34-year-old gentleman presented with insulin resistant diabetes, hypertension, blindness, deafness, and primary infertility. Clinical examination revealed retinitis pigmentosa and normal cognitive functions. Investigations revealed C-peptide level of 2247 pmol/L and macroalbuminuria. His diabetes was gradually improved with increasing doses of insulin along with metformin and subsequently rosiglitazone. Both drugs were discontinued because of liver and renal impairment. Further investigations revealed non alcoholic steatohepatitis. He was diagnosed clinically as Alström syndrome.

Case 2 summary:

A 35-year-old gentleman presented with insulin treated diabetes, hypertension, dyslipidemia, impaired renal function, proteinuria, and blindness. Clinical examination revealed obesity, polydactyly, retinitis pigmentosa, normal cognitive functions, and ataxic gait. C-peptide level was 137 pmol/L. Abdominal ultrasound showed multiple renal cysts. He was diagnosed clinically as Bardet-Biedl syndrome.

Conclusion:

We report two rare cases of diabetes mellitus combined with retinitis pigmentosa. The cases were diagnosed clinically as Alström syndrome and Bardet-Biedl syndrome based on the coexistent clinical manifestations. The use of insulin sensitizers for the management of insulin resistant diabetes in Alström syndrome was restricted by the presence of renal and hepatic dysfunction.

Key Words: Diabetes; Retinitis pigmentosa; Kuwait