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Case Report

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AN INTERESTING CASE OF DIABETES WITH SYNDROMIC MANIFESTATION-MODY -5

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INTRODUCTION- MODY(Maturity Onset Diabetes of the Young) is defined as a dominantly inherited young onset non autoimmune diabetes that occurs in adolescence or young adulthood (usually <25 yrs) due to primary defect in pancreatic beta cell function. First reported in three families by Tattersall in 1974. The term MODY was used in 1975 for the first time following further clinical description. Presents 3-5% of all diabetes cases. Current prevalence is 70-110 per million that is roughly about 1 person per 10000.Out of it 5-10% cases are MODY 5- about 1 person per lac.[1-3]

CASE REPORT-A 27 year old unmarried man(fig-1) presented with head ache for 10 days. The patient was diagnosed with type 1 diabetes due to high blood sugar about 10 years ago and was started with insulin. Following 9 years the patient was completely asymptomatic. About a year back the patient was diagnosed to have some kidney disease as well as hypertension and anemia, 4 months back the patient was diagnosed with NSTEMI. The patient's father was diabetic, one maternal aunt and paternal uncle's son was diabetic and detected in 3rd decade.

On examination-BP-230/120 mmof Hg, GRBS-170 mg/dl. pallor, puffiness of face and pedal edema was present. On systemic examination free fluid was present in the abdomen, other systems being normal.

Routine investigations showed-normocytic normochromic anemia, LFT-ALT-104,AST-55,ALP-129, Lipid profile normal, blood urea-85 mg/dl, serum creatinine-2.5 mg/dl, FBS-139 mg/dl, PPBS-180 mg/dl, HbA1C-5.6%,24 hour urine albumin-1.5g,C -peptide 0.79, anti GAD and anti Islet cell antibody negative, serum uric acid-8 mg/dl, USG abdomen showed bilateral multicystic kidneys, 2D ECHO- hypertensive heart disease, MR angiogram of brain was normal, CT abdomen and pelvis-polycystic kidneys(fig-2), calcific atrophic pancreas, mild pericardial effusion

Fig-1

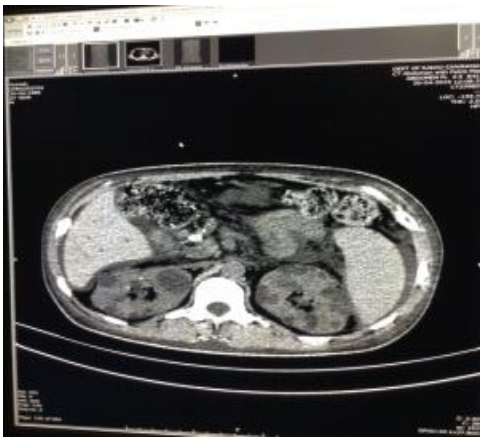

Fig-2

DISCUSSION

MODY type 5 is an uncommon cause of monogenic diabetes (12%) and was described in 1997 for the first time in a Japanese subject.

This type of MODY is early onset before age 25.

It is caused by a mutation in the gene encoding the transcription factor **HNF1β,** located at the locus gene **17q21.3.**[4-6]

COMPONENTS

1. Diabetes 2. Renal cystic disease 3. Hyperuricemia 4. Raised liver enzymes 5. Atrophic pancreas 6. Dental enamel defect.

Other features

1. Prognathism
2. Pyloric stenosis
3. Ligament laxity
4. Hearing loss
5. Learning difficulties

CONCLUSION

MODY is a rare type of diabetes which is usually undiagnosed. Mody type 5 is extremely rare variety of MODY with typical involvement of kidneys. Proper diagnosis is must to Classify, Plan management., Prognostication and genetic counselling.

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