

# Early diagnostics of Wolfram syndrome(Case Report)

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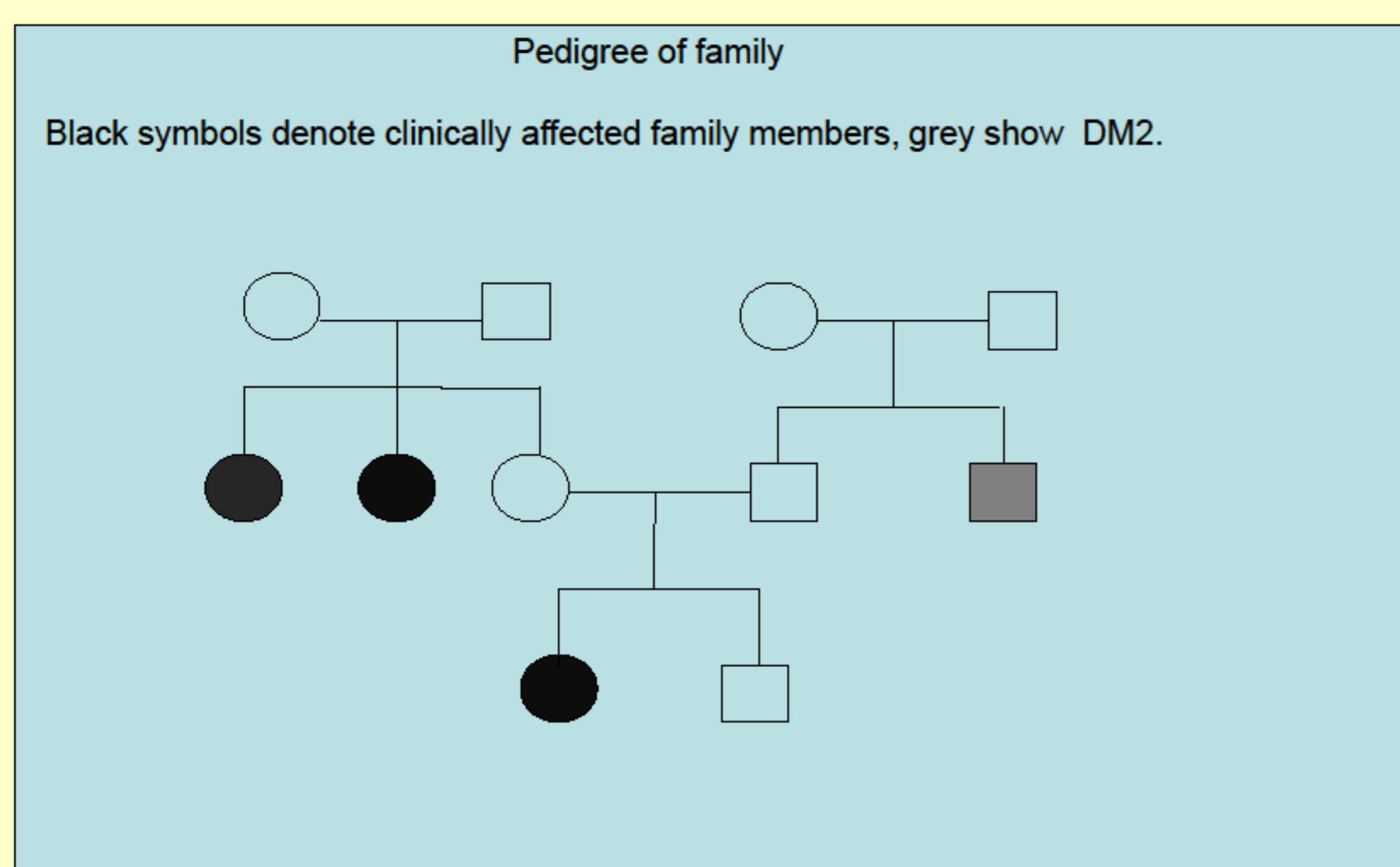


## OBJECTIVES

Wolfram syndrome is rare, progressive autosomal recessive disease with characteristic neurological and endocrine features. Signs and symptoms appear with different combination during the lifespan in different patients. Here we report the family case of Wolfram syndrome with different phenotype variable.

## METHODS

Patient 4 year 4 month old girl with Diabetes Mellitus since the age of 2 year 3 months Born term, healthy, with no previous medical problems. Was hospitalised in clinic for Diabetic Ketoacidosis. Treatment with human insulin was started. Physical examination -no significant findings. Insulin daily requirement 0,21U/kg. Patient' s parents are healthy, no consanguinity. Patient's two maternal aunts had Wolframs syndrome. The older of the two had DM, optic atrophy, deafness, mental retardation(psychiatric problems). The younger aunt had DM, Diabetes Insipidus, optic atrophy, a large bladder. Both died at the age of 15 an 13 years with complications(hypoglycemia, infection). It is significant that, in both cases diseases started at a young age(DM by the age of 6, optic nerve atrophy, Diabetes Insipidus and deafness by the age of 10 years).



## Graphs and tables

	Diabetes Mellitus	Diabetes Insipidus	Optic atrophy	Deafness	Mental retardation	Large bladder
Patient	+	-	-	-	-	-
Aunt1	+	-	+	+	+	-
Aunt2	+	+	+	-	-	+

## RESULTS

Fasting C-peptide 0,50ng/ml, Islet-cell-ab<5 JDF-U, Islet cell Titer 1:<10Titer. GAD<5IU/ml, TSH -1.00mIU/l, Cortisol -231,48ng/ml. Mean HbA1c-5,9%. Genetic testing for WFS1 gene mutation was performed, with both PCR as well as whole genome sequencing. c. 1523-1524deLAT(p.Tyr508Cysfs\*34) homozygous mutation in the 8<sup>th</sup> exon of WFS1 gene was found.

## CONCLUSIONS

Characteristic features of Wolfram Syndrome are variable even in the same family. Course of DM is quite mild in our patient with minimal insulin requirement, compared to other patients. Early identification of the syndrome gives us a chance to for early detection and proper management of associated conditions and its complication with maximal efficacy.

## References

1. Tanabe K. et al. Nihon Rinsho. Wolfram syndrome clinical features, molecular genetics of WFS1 gene. Review. Japanese 2015Feb;73(2):341-9.PMID:25764693.
2. Rigoli L and Di Bella C: Wolfram syndrome 1 and Wolfram syndrome 2. Curr Opin Pediatr 2-12, 24:512-7./article/PubMed