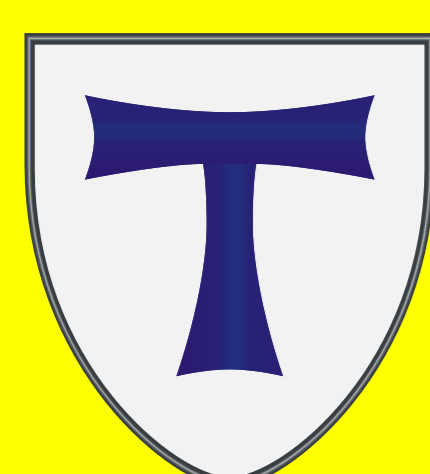


# New mutation of the PDX-1 gene causes MODY Type 4 Diabetes in a 17 years old girl with good response to oral Antidiabetics



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## Introduction

A 17 years old female adolescent was referred to our clinic due to hyperglycemia. She complained of dizziness and nausea. Her blood pressure was 160 /100 mmHg; she had hyperglycemia (208 mg/dl), glycosuria and ketonuria without ketoacidosis. Some members of the family were on insulin therapy and others were treated with oral antidiabetics. The HbA1c was 8.2 % .

## Hospitalisation and start of insulin treatment

We started an intensified therapy with basal and bolus insulin, to which the patient responded well. The insulin requirement was about 1 IU/kg body weight and the measured HbA1c 2 months afterwards was 6 %.

**The diabetes autoantibodies were negativ**

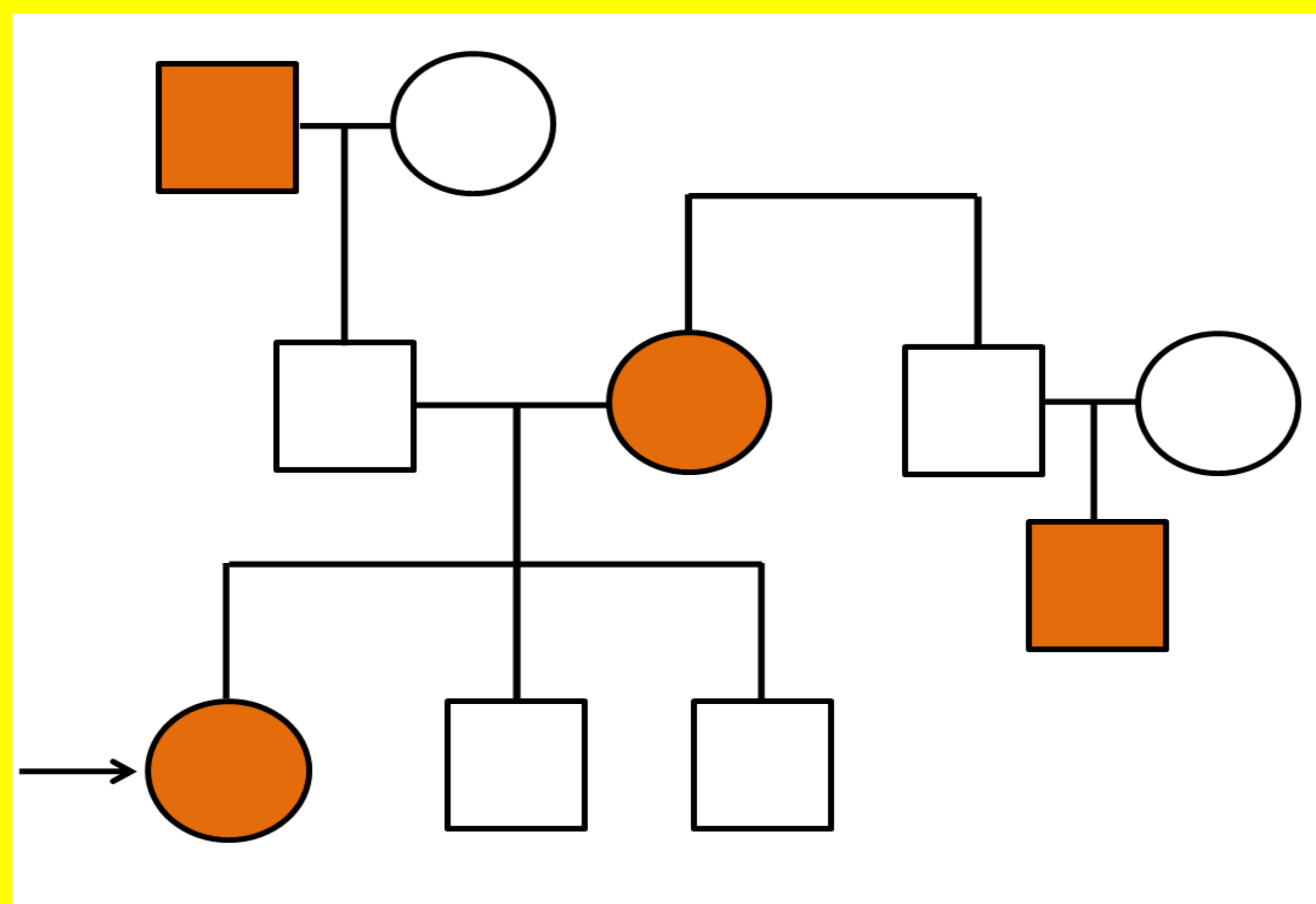
(Islet Cell Cytoplasmic Autoantibodies (ICA), Insulin Autoantibodies (IAA), Glutamic Acid Decarboxylase Autoantibodies (GADA), GAD65 Autoantibodies )

## Molecular examination of the MODY genes

Our patient had a mutation in the PDX-1 gene

(c.479A>T; p.Glu160Val) . Mutations in the PDX-1 gene are responsible for MODY diabetes type 4. This mutation however has not yet been detected in other MODY diabetes patients.

## Family tree



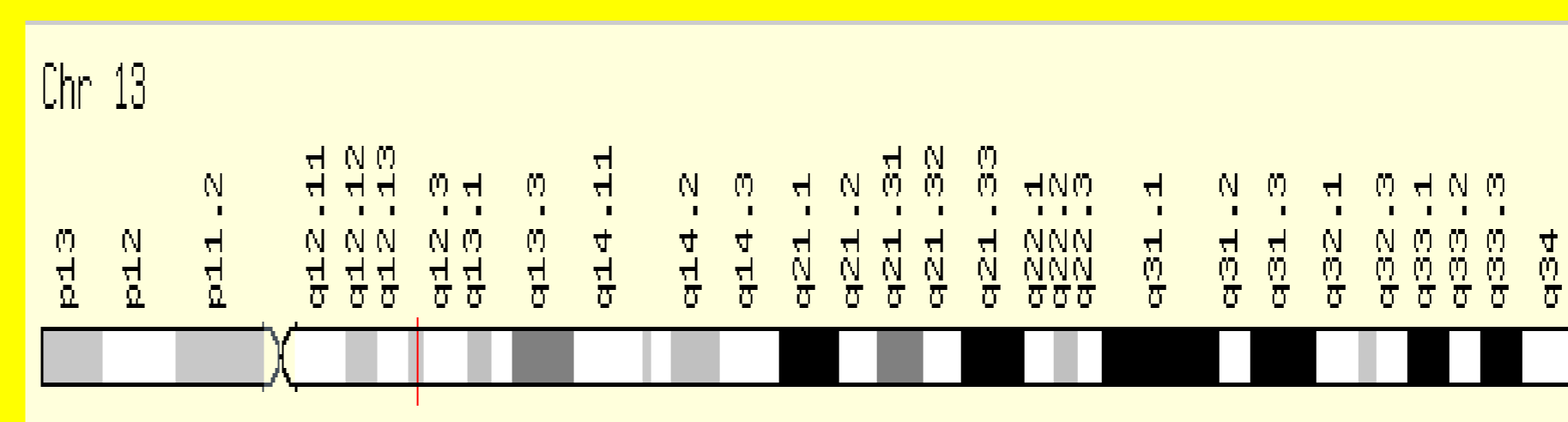
The orange-marked members of the family have diabetes

## Conclusions

We suggest that the detected mutation of the PDX-1 gene is the cause of the MODY diabetes in our patient. It is important to recognize MODY diabetes type 4 in children and adolescents, because this type of diabetes can be treated with oral antidiabetics instead of insulin .

## PDX1 / IPF 1 : pancreatic and duodenal homeobox 1 Gen /insulin promoter faktor-1

This gene encodes a protein which is a transcriptional activator of several genes, including insulin, somatostatin, glucokinase, islet amyloid polypeptide and glucose transporter type 2. The encoded core protein is involved in the early development of the pancreas and plays an important role in glucose-dependent regulation of insulin gene expression. Homozygous defects in this gene are a cause of pancreatic agenesis, and defects in the gene can lead to early-onset insulin-dependent diabetes mellitus (IDDM) and MODY diabetes.

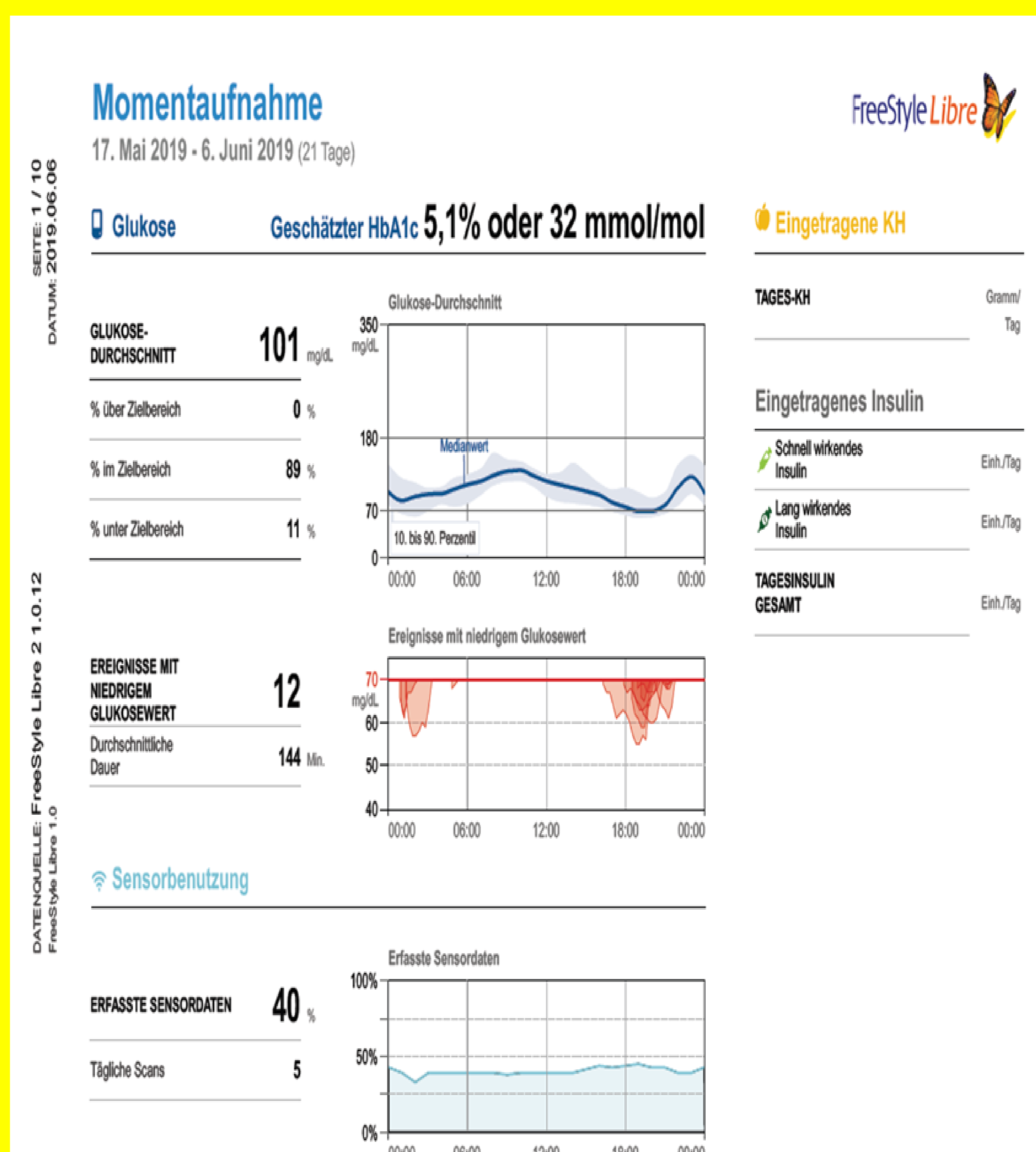


Gene location:13q12.2

## Switch from insulin to oral antidiabetics

**Glibenclamide**, starting dose 0,875 mg once daily

- Blood sugar levels normal
- No side effects
- HbA1c 2 months after treatment with glibenclamide 6 %



The authors have nothing to disclose.

