

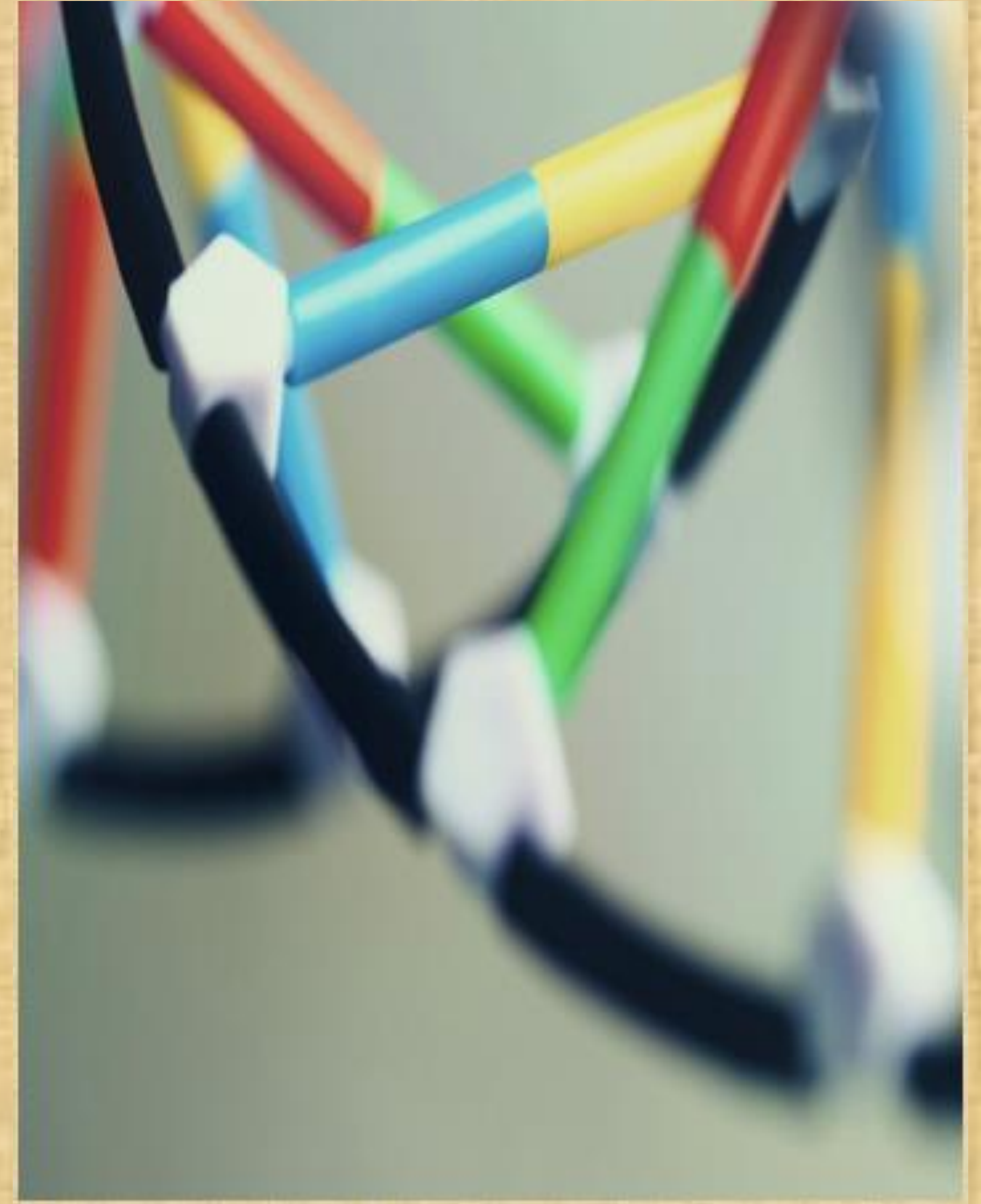
MONOGENIC DIABETES CAUSE BY MUTATION OF THE GENE HNF – 1A

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INTRODUCTION

The MODY diabetes (Maturity Onset Diabetes of the Young) belongs to monogenic alterations group, the mutation of the gene HNF-1 α is the most common and present an autosomal dominant inheritance that causes dysfunction of the Beta pancreatic cell and alteration in the reabsorption of glucose to renal level, with age of variable presentations, it often leads to a diagnosis as type 1 diabetes mellitus.



DESCRIPTION OF THE CLINICAL CASE

Present in a 3-Year-old patient that debuts with hyperglycemia without ketosis, that has been catalogued as type 1 diabetes mellitus, therefore, it initiates treatment with insulin (basal- bolus) with low dose (0.3UI/Kg/day) and continuous monitoring system of glucose, being relevant in her clinical history the presence of antibodies negative and the low requirements of exogenous insulin after 2 years of evolution, with HbA1 6,5 %. The genetic study showed a mutation of the gene HNF-1a in heterocycosis c.62 C> G described before. With these findings, the treatment began with sulfunilureas (0.07mg / kg / day) up to the maximum dose recommended (1.5mg / kg / day) without therapeutic awaited response.

CONCLUSIONS

We are with a patient that is showing a mutation of the HNF-1 α gen located at the region of the promoter in heterozygosis c.62 C>G of early beginning in the evidence, that this entity has a wide variability, not only in the age of beginning but in the response to the treatment with sulfunilureas.

