Subclinical endothelial inflammation markers in a family with type i familial hyperaldosteronism caused by a de novo mutation Marcadores de inflamación endotelial subclínica en una familia con hiperaldosteronismo familiar tipo i por mutación de novo

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Background: Type I familial hyperaldosteronism is caused by the presence of a chimaeric gene CYP11B1/CYP11B2 which encodes an enzyme with aldosterone synthetase activity regulated by a drenocorticotrophic hormone (ACTH). Therefore, in patients with FH-I is possible to normalize the aldosterone levels with glucocorticoid treatment. Recently it has been shown that aldosterone plays a role in the production of endothelial oxidative stress and subclinical inflammation. Aim: To evaluate subclinical endothelial inflammation markers, like Mtalloproteinase 9 (MMP-9) and ultrasensitive Creactive protein (usPCR), before and after glucocorticoid treatment in family members with FH-I caused by a de novo mutation. Patients and methods: We report three subjects with FH-I in a dngle family (proband, father and sister). We confirmed the presence of a chimaeric CYP11B1/CYP11B2 gene by long-PCR in all of them. Paternal grandparents were unaffected by the mutation. The proband was a 13 year-old boy with