A case study on a rare disorder presenting as polycystic kidney disease

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Keywords. Polycystic kidney disease; Orofaciodigital syndrome; Systemic lupus erythematosus.

Introduction. Although loss of renal function in polycystic kidneys is mostly caused by autosomal dominant polycystic kidney disease (PKD), it is important to consider the broader differential diagnosis for this finding.

Case description. A forty-year-old female patient (BMI 24.7 kg/m2) with few years long anamnesis of arthritis presented with unclear worsening of kidney function (eGFR 65-43 ml/min CKD-EPI) and I grade arterial hypertension (148/93 mmHg). The patient had a history of multiple kidney cysts identified with ultrasonography (Fig.1), congenital heart disease with Fallot tetrad correction in youth, seronegative rheumatoid arthritis, and negative family anamnesis for PKD. Diagnostic evaluation revealed normal urine analysis and significantly elevated anti-dsDNA (>240 IU/mL) and ANA (11.5 IU/mL), suggesting PKD or systemic lupus erythematosus. A kidney biopsy was performed, showing non-specific findings of glomerular hypertrophy with focal segmental glomerulosclerosis and no immune deposits. To obtain a specific diagnosis, genetic testing was performed, and OFD1 gene mutation was identified, which confirmed the diagnosis of orofaciodigital syndrome type 1 which is a rare X-linked genetic condition mainly presenting with anomalies in oral cavity, face and digits, as well as being associated with polycystic kidney disease.

Conclusions. This case report emphasizes the crucial importance of thorough patient evaluation, even when the initial diagnosis appears to be known.

Fig.1 US of the left kidney performed in 2022.