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Letter to the Editor

LETTER TO THE EDITOR

Reporting inherited kidney diseases: pick up the gauntlet

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On behalf of the Spanish Pediatric Non-End-Stage Chronic Kidney Disease Registry (REPIR II), we completely agree with Torra *et al.* [1], who highlight the important role of national and international registries to report inherited kidney diseases (IKDs) as a separate category.

Genetic forms of kidney disease are prevalent in paediatric populations. They are reported as 10–19% of causes of chronic kidney disease (CKD) and up to 22% of end-stage renal disease (ESRD) in children [2]. Diagnosis is also increasing in adults due to easy accessibility of genetic diagnostic technologies [3].

Paediatric CKD registries in children provide variable but very relevant epidemiological data. They are a clinical tool for early detection of the disease and identification of reversible or preventable causes and allow appropriate intervention for risk factors in order to improve prognosis and slow the progression of renal disease, including IKDs [4]. Despite the importance of early diagnosis in the progression of the disease, information on the epidemiology of the early stages of CKD is even more limited because the disease is usually asymptomatic and therefore underdiagnosed and unreported [5].

REPIR II is an Internet-based data collection system that annually records, since 2007, demographic, clinical, laboratory and treatment data of patients <18 years of age with CKD Stages 2–5 living in Spain [6]. Currently it collects data for >2100 children. According to the REPIR II, the annual incidence and prevalence of CKD Stages 2–5 in Spain are ~10 and ~100 per million of the age-related population.

In 2019, our registry adopted the ERA-EDTA coding system for Primary Renal Disease (PRD) thanks to the related web-based search tool (https://www.era-edta-reg.org/prd.jsp) [7] after translation and adaptation into Spanish [8]. The use of a coding PRD adjusted to international standards enhances quality and interoperability between registries.

REPIR II reports epidemiological data during the national annual scientific congress and eventually at the European congress [9]. Our report usually groups IKD as a specific entity (VI Group, Familial/Hereditary Nephropathies of the ERA-EDTA coding system) accounting for 14% of all registered children (Figure 1). Nevertheless, other genetic PRDs might be included in categories such as Group II: Tubulointerstitial diseases (multicystic dysplastic kidneys associated *HNF1B* gene mutation) or in Group I: Glomerular diseases (Finnish-type nephrotic syndrome with an *NPHS1* gene mutation).

Fortunately, this useful coding tool provides additional information for required diagnostic criteria such as clinical, biochemical, histology, imaging or gene testing. In the same way that the coding system includes the term 'histologically proven' and 'no histology' even for some PRDs, which should not reasonably be used without histological evidence, the term 'genetically proven' and 'no gene test' might be considered for some diseases such as congenital anomalies of kidney and urinary tract, cystic diseases or tubulopathies as a beginning of a path that integrates the burden of IKD in the epidemiology of CKD.

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CONFLICT OF INTEREST STATEMENT

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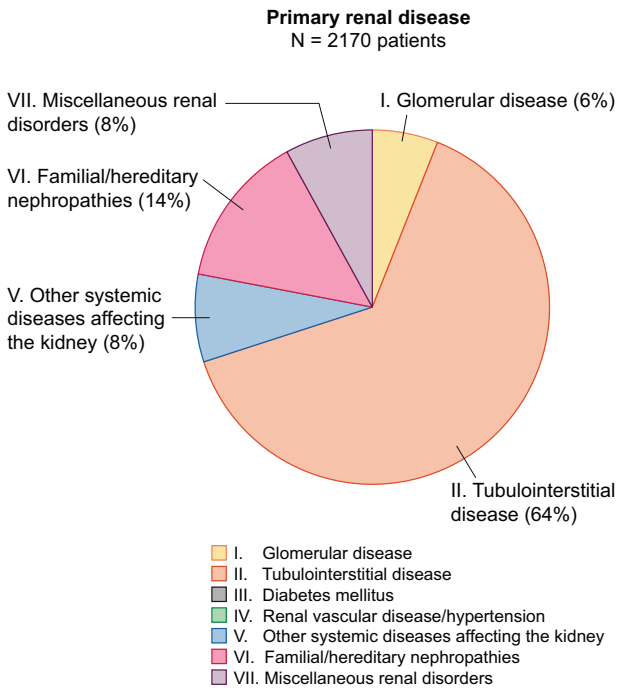

FIGURE 1: Primary renal disease.

REFERENCES

1. Torra R, Furlano M, Ortiz A *et al*. Genetic kidney diseases as an underecognized cause of chronic kidney disease: the key role of International registry reports. *Clin Kidney J* 2021; 10.1093/ckj/sfab056
2. Harambat J, van Stralen KJ, Kim JJ *et al*. Epidemiology of chronic kidney disease in children. *Pediatr Nephrol* 2012; 27: 363–373
3. Cocchi E, Nestor JG, Gharavi AG. Clinical genetic screening in adult patients with kidney disease. *Clin J Am Soc Nephrol* 2020; 15: 1497–1510
4. Plumb LA, Hamilton AJ, Inward CD *et al*. Continually improving standards of care: the UK Renal Registry as a translational public health tool. *Pediatr Nephrol* 2018; 33: 373–380
5. Liu FX, Rutherford P, Smoyer-Tomic K *et al*. A global overview of renal registries: a systematic review. *BMC Nephrol* 2015; 16: 31
6. Areses TR, Sanahuja IM, Navarro M. Epidemiología de la enfermedad renal crónica no terminal en la población pediátrica española. *Proyecto REPIR II. Nefrologia* 2010; 30: 508–517
7. Levey AS, Eckardt KU, Dorman NM *et al*. Nomenclature for kidney function and disease: report of a Kidney Disease: Improving Global Outcomes (KDIGO) Consensus Conference. *Kidney Int* 2020; 97: 1117–1129
8. Zurriaga Ó, López-Briones C, Martín Escobar E *et al*. Adaptación en español del nuevo sistema de codificación de enfermedad renal primaria de la European Renal Association-European Dialysis and Transplant Association (ERA-EDTA). *Nefrología* 2015; 35: 353–357
9. Melgosa-Hijosa M, Gamero Reyes AM, Ventura O. S. Predicting GFR in children with CKD: Application of artificial intelligent on REPIR II Project. Abstracts of the 18th Congress of the International Pediatric Nephrology Association, Venice (Italy), October 2019. *Pediatr Nephrol* 2019; 34:2039