of polycystic kidney disease (OMIM #173900), which also has an autosomal dominant inheritance pattern.[1,5] This is the most common hereditary kidney disorder, with an incidence of one in every 400–1000 live births.[5]

A large deletion which may encompass the genes *TSC2* and *PKD1* causes so-called *TSC2/PKD1* contiguous gene syndrome (OMIM #600273).[1,2,5] It was first reported by Brook-Carter et al. in 1994 in 6 patients with tuberous sclerosis complex with severe childhood polycystic kidney disease.[6] It is characterised by large bilateral kidney cysts. These cysts may be congenital or of very early onset, which may alter the disease prognosis. It has been estimated that 5% of patients with tuberous sclerosis complex have polycystic kidney disease.[1]

Kidney complications represent the second leading cause of death after neurological impairment in tuberous sclerosis complex. Angiomyolipomas are the most common kidney abnormality in adults and children, and may be present in 16% of cases, in addition to small cysts and renal cell carcinoma.[1,3]

We present the case of a girl with a genetic and clinical diagnosis of *TSC2/PKD1* contiguous gene syndrome, with an emphasis on multidisciplinary assessment given her pleiotropy and disease severity, as well as appropriate genetic counselling with a 50% risk of recurrence in descendants of affected individuals.

REFERENCES

1. Llamas Velasco S, Camacho Salas A, Vidales Moreno C, Ceballos Rodríguez RM, Murcia García FJ, Simón de la Heras R. *TSC2/PKD1* contiguous gene deletion syndrome. An Pediatr (Barc). 2013;79:42–5.
2. Ismail NF, Nik Abdul Malik NM, Mohseni J, Rani AM, Hayati F, Salmi AR, et al. Two novel gross deletions of *TSC2* in Malaysian patients with tuberous sclerosis complex and *TSC2/PKD1* contiguous deletion syndrome. Jpn J Clin Oncol. 2014;44:506–11.
3. Alp A, Esener S, Gürsoy D. Segmental tuberous sclerosis in a patient with chronic kidney disease. Nefrologia. 2016, http://dx.doi.org/10.1016/j.nefro.2016.09.012. pii:S0211-6995(16)30147-3 [Article in English, Spanish].
4. Monteiro T, Garrido C, Pina S, Chorão R, Carrilho I, Figueiroa S, et al. Tuberous sclerosis: clinical characteristics and their relationship to genotype/phenotype. An Pediatr (Barc). 2014;81:289–96.
5. Furlano M, Barreiro Y, Martí T, Facundo C, Ruiz-García C, DaSilva I, et al. Renal angiomyolipoma bleeding in a patient with TSC2/PKD1 contiguous gene syndrome after 17 years of renal replacement therapy. Nefrologia. 2016, http://dx.doi.org/10.1016/j.nefro.2016.04.007. pii:S0211-6995(16)30058-3.
6. Brook-Carter PT, Peral B, Ward CJ, Thompson P, Hughes J, Maheshwar MM, et al. Deletion of the *TSC2* and *PKD1* genes associated with severe infantil epolycystic kidney disease – a contiguous gene síndrome. Nat Genet. 1994;8:328–32.

Francisco Cammarata-Scalisi[a,*], Concha Vidales Moreno[b], Carmen Zara-Chirinos[c], Ana Bracho[c], Diomar Pérez[d]

[a] *Unidad de Genética Médica, Departamento de Puericultura y Pediatría, Facultad de Medicina, Universidad de Los Andes, Mérida, Venezuela*
[b] *Área Diagnóstica, DNA Data, San Sebastián, Guipúzcoa, Spain*
[c] *Instituto de Investigaciones Genéticas, Facultad de Medicina, Universidad del Zulia, Maracaibo, Venezuela*
[d] *Estudiante de Medicina, Facultad de Medicina, Universidad de Los Andes, Mérida, Venezuela*

[*] *Corresponding author.*
E-mail address: francocammarata19@gmail.com (F. Cammarata-Scalisi).

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Barriers for the development of home haemodialysis in Spain. Spanish nephrologists survey☆

Barreras para el desarrollo de la hemodiálisis domiciliaria en España. Encuesta a nefrólogos españoles

Dear Editor,

Last decade, the number of patients treated with home haemodialysis (HHD) worldwide has increased considerably[1]; this is in part due to the clinical benefits of more intensive haemodialysis (HD) regimens in HHD.[2–5] In Spain, however, despite a growing interest and modern systems that simplify HHD, the number of patients using HHD has not increased

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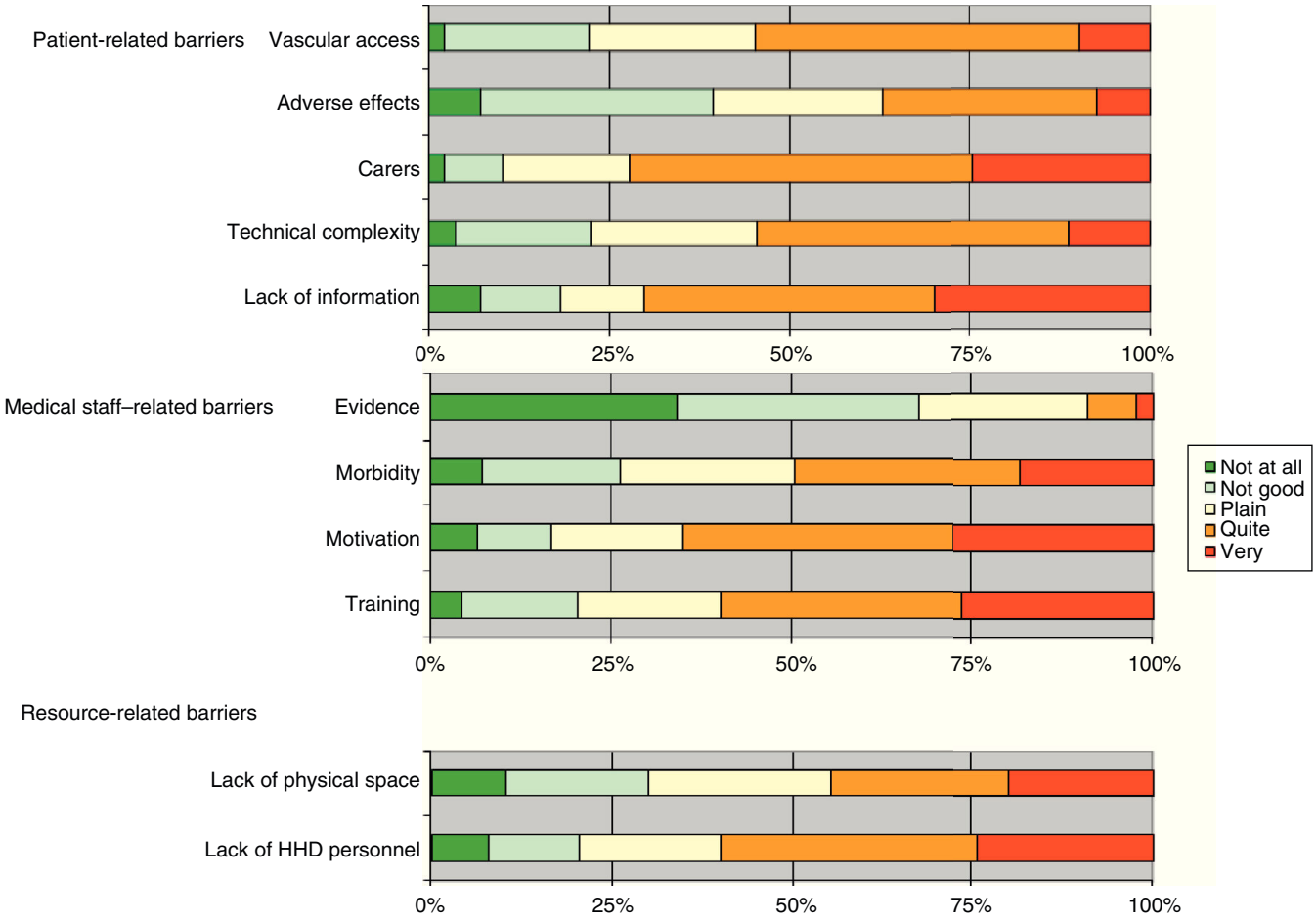

Fig. 1 – Barriers to implementing home haemodialysis programmes.

substantially.[6] We administered a survey to Spanish nephrologists, sent mainly by e-mail through the Spanish Society of Nephrology, to learn more about the barriers for implementing HHD in Spain.

Between February and November of 2015, we received 142 surveys from 76 different HD units, 60 of them hospital-based and 16 out-of-hospital (mean age of surveyed: 44.69 ± 10.55 years). HHD programmes were available in 32 of these centres (42.1%); 15 (19.73%) offered the option even though they did not have their own HHD unit, and in 29 (38.15%) the patients were not offered this possibility. All the HHD units were hospital-based, with no HHD programmes dependent on peripheral HD centres, in which it was only offered in 18.7% of cases (compared with 73.3% of HHD offerings performed in hospitals). 47% of HHD programmes were started after 2011; 23% between 2006 and 2010; 13% between 2001 and 2005; and 17% before 2000. In all, 60.5% of the nephrology units stated that they would be capable of offering their own HHD programmes within one year's period.

The median number of patients per HHD unit was 2 (range: 0–17); 59.4% of the centres had 2 patients or fewer; and only 12.5% had 6 patients or more. The nephrologist in charge of the HHD programme in 42% of the cases was also responsible for HD; in 32% for peritoneal dialysis (PD); and in 26% for both methods. In 46% of the cases the patients came from HD at the centre; 22% from PD; 25% from outpatient clinics; and 7% from renal transplantation.

We asked the nephrologists for their opinions about any clinical benefits of HHD. We obtained the highest percentage of affirmative responses in regard to quality of life (95.8%), and the lowest in regard to mortality (67.6%). As for anaemia, high blood phosphate levels, blood pressure, nutrition and morbidity, opinion was in favour of HHD in 80.3%, 84.5%, 87.3%, 88% and 76%, respectively. In addition, 85.9% of the surveyed said that HHD could produce economic benefits to the health service. In total, 45.8% of the nephrologists said that living far from the referral centre was a factor favouring implementing of HHD and, did not compete with PD, according to 52.8% of the responses.

We asked about possible barriers for the development of HHD by dividing them into patient-related factors (lack of information, technical complexity, need for carers, fear of

Table 1 – Patient-, medical staff- and resource-related barriers by size of centre.

	Type of centre per patient							
	3 or more			2–1			None	
	Yes	Yes %	*p*	Yes	Yes %	*p*	Yes	Yes %
Lack of information	31	79.5		24	68.6		41	65.1
Technical complexity	14	36.8	Sig.	21	61.8		38	61.3
Carers	20	51.3	Sig.	30	85.7		48	77.4
Adverse effects	3	7.7	Sig.	11	31.4	Sig.	37	58.7
Vascular access	14	35.9		19	55.9		41	66.1
Training	30	76.9	Sig.	17	48.6		35	55.6
Motivation	29	74.4		22	62.9		38	60.3
Morbidity	14	35.9		16	45.7		38	60.3
Evidence	1	2.6		3	8.6		8	13.1
Personnel	24	61.5		16	45.7	Sig.	42	66.7
Space	20	51.3	Sig.	10	28.6	Sig.	31	50.0

Sig: statistical significance between types of centre $p < 0.05$.

acute adverse effects of the method and difficulty with vascular access); medical staff-related factors (lack of properly trained medical personnel, lack of personnel motivated to perform home methods, consideration of excessive patient morbidity, lack of scientific evidence in favour of HHD); and resource-related factors (lack of equipment to work with HHD, lack of a physical space to conduct training) (Fig. 1).

Next, we divided the barriers according to the size of the HHD unit (Table 1), to see whether they varied depending on whether the nephrologist had more experience with the home method.

This survey showed that there is a growing interest in HHD on the part of Spanish nephrologists. The data must be analysed by considering that all surveys are answered mainly by those who are the most interested in the subject being investigated, and therefore we are aware that this real interest in the method may be overvalued and the possible barriers greater than what has been expressed. In any case, it seems that as HHD programmes develop, the initial barriers, which stem from a lack of practical knowledge of the method, are disappearing[7,8] and only resource-related barriers remain[9,10] (lack of information, training and personnel dedicated exclusively to HHD).

From the HHD Support and Promotion Group in Spain, we must focus our efforts on showing the results obtained in regard to generating HHD programmes, trusting that in the coming years the barriers stated in this survey – and which in many cases prevent this method of treatment from being offered to all patients as one more option to improve their quality of life – may be problems that will be overcome.

REFERENCES

1. Collins AJ, Foley RN, Chavers B, Gilbertson D, Herzoq C, Ishani A, et al. US renal data system annual data report. Am J Kidney Dis. 2013;63 Suppl.:A7.
2. Suri RS, Nesrallah GE, Mainra R, Garg AX, Lindsay RM, Greene T, et al. Daily hemodialysis: a systematic review. Clin J Am Soc Nephrol. 2006;1:33–42.
3. Walsh M, Culleton B, Tonelli M, Manns B. A systematic review of the effect of nocturnal hemodialysis on blood pressure, left ventricular hypertrophy, anemia, mineral metabolism, and health-related quality of life. Kidney Int. 2005;67:1500–8.
4. Nesrallah GE, Lindsay RM, Cuerden MS, Garg AX, Port F, Austin PC, et al. Intensive hemodialysis associates with improved survival compared with conventional hemodialysis. J Am Soc Nephrol. 2012;23:696–705.
5. Chertow GM, Levin NW, Beck GJ, Depner TA, Eggers PW, Gassman JJ, et al. In-center hemodialysis six times per week versus three times per week. N Engl J Med. 2010;363:2287–300.
6. Pérez-Alba A, Barril-Cuadrado G, Castellano-Cerviño, Martín-Reyes G, Pérez-Melón C, Slon-Roblero F, et al. Hemodiálisis domiciliaria en España. Nefrología. 2015;35:1–5.
7. Cornelis T, Tennankore KK, Goffin E, Rauta V, Honkanen E, Özyilmaz A, et al. An international feasibility study of home haemodialysis in older patients. Nephrol Dial Transpl. 2014;29:2327–33.
8. Pauly RP, Eastwood DO, Marshall MR. Patient safety in home hemodialysis: quality assurance and serious adverse events in the homesetting. Hemodial Int. 2015;19:S59–70.
9. Fluck RJ, Fouque D, Lockridge RS Jr. Nephrologists' perspectives on dialysis treatment: results of an international survey. BMC Nephrol. 2014;15:16.
10. Ludlow MJ, George CR, Hawley CM, Mathew TH, Agar JW, Kerr PG, et al. How Australian nephrologists view home dialysis: results of a national survey. Nephrology (Carlton). 2011;16:446–52.

Alejandro Pérez Alba[a,*], Fernanda Slon Roblero[b], Sandra Castellano Gasch[c], M. Auxiliadora Bajo Rubio[d]

[a] *Hospital General Universitario de Castellón, Castellón, Spain*
[b] *Hospital de Navarra, Pamplona, Spain*
[c] *FMC Madrid Dialcentro, Madrid, Spain*
[d] *Hospital Universitario La Paz, Madrid, Spain*

[*] *Corresponding author.*
E-mail address: aperezalba@gmail.com (A. Pérez Alba).

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