

# Primary prevention of hereditary disease and the economic sustainability of public health systems go hand in hand

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## Introduction

Hereditary disease (HD) is still an unknown entity in terms of incidence and prevalence. We recognize some data of some diseases such as autosomal dominant polycystic kidney disease (ADPKD),<sup>1</sup> although others such as Alport syndrome or Fabry disease are masked. In the population of kidney patients, Genetics tries to identify cases by using generation sequencing (NGS) technology, although a better horizon is already offering exome sequencing,<sup>2</sup> which will have to be strengthened in the future.

Today we have NGS panels for hereditary renal disease (HRD) at the healthcare level. About 15 years ago, if you suspected HRD, you could request a gene study; if the result was negative, you could request another one and so on, and frequently you did not reach a good diagnosis. Shortly after came the narrow NGS panels investigating 8-12 genes: Congratulations if your patient's HRD was in that! If not, you still didn't get a diagnosis. Since 2018, our center used the Nephropathy Solution (NES) of Sophia Genetics, which includes 44 most clinically relevant genes related to a broad range of nephropathies, cases where other panels failed.

Patients suffering from HD share a common approach horizon primary prevention (PP). All individuals affected by HD at reproductive age should have genetic counseling and access to assisted human reproduction (AHR) techniques that favor the birth of healthy children and, therefore, will no longer transmit it to their offspring.

Our group has shown that preventive reproductive intervention, in addition to the health advantages indicated and the consequent reduction in the frequency of the disease, is capable of reducing the health costs derived from renal replacement therapy (RRT), which will help public health system (PHS) to be sustainable.

We present our study carried out in ADPKD individuals where the preventive strategy using Preimplantation Genetic Testing (PGT) is compared with the current panorama of arrival on RRT (Figure 1).<sup>3</sup> For this, a simulated design was used, applying the health intervention of the PGT in couples with one of its affected members, and the measure of effectiveness used was the live births per transfer rate (LBTR). This study arose as a consequence of the need to assess the economic impact of the implementation of the non-legal proposal (10-16/PNLC-000248) approved in 2016 in the Andalusian Parliament regarding the creation of the Primary Prevention Plan for ADPKD, which in its point 5 urged the Government of the Junta de Andalucía to promote the most appropriate reproductive options that would prevent the transmission of the disease and specifically the PGT.<sup>4</sup> For this analysis, information was collected from deceased ADPKD patients with complete data on the start and end dates of each RRT modality up to the time of death (n=143), and an average cost was imputed, considering exclusively the costs of the stages of RRT according to the average annual costs published patient/year.<sup>5</sup> Regarding the reproductive strategy, the option of elective transfer

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of a single fresh embryo and two more attempts with cryopreserved embryos was chosen in case pregnancy was not achieved, considering a LBTR of 25% for the transfer of the fresh embryo. and 20% for transfers after cryopreservation. Given the good results obtained, this strategy was recommended to the health authorities of the Andalusian Public Health System (APHS) through the Preventive Project of ADPKD, and in 2022 PGT was authorized in Granada and the first girl free of disease from an ADPKD mother through PGT has already been born in the PHS.<sup>6-8</sup>

	No. Cases	Embryo Transfer	Children Born	Health Condition	Treatment	Unit Cost	Total Cost	Cost Period	Net Present Value	Cost-Effectiveness Ratio
Current Strategy	100	-	50	Healthy		€ -				
		-	50	ADPKD	TCKF	€ 170,800.54	€ 8,540,027.00	58 a 66	€ 1,370,299.19	
			50				€ 8,540,027.00		€ 1,370,299.19	€ 27,405.98
Preventative Strategy	100	1	25	Healthy		€ 5,520.00	€ 138,000.00	1	€ 138,000.00	
		1	75	Not pregnancy		€ 5,520.00	€ 414,000.00	1	€ 414,000.00	
		2	15	Healthy		€ 500.00	€ 7,500.00	1	€ 7,500.00	
		2	60	Not pregnancy		€ 500.00	€ 30,000.00	1	€ 30,000.00	
		3	12	Healthy		€ 500.00	€ 6,000.00	1	€ 6,000.00	
		3	48	Not pregnancy		€ 500.00	€ 24,000.00	1	€ 24,000.00	
		-	24	Healthy		€ -	€ -			
		-	24	ADPKD	TCKF	€ 170,800.54	€ 4,099,212.96	58 a 66	€ 657,743.61	
			76				€ 4,718,712.96		€ 1,277,243.61	€ 16,805.84

**Figure 1** Comparative cost-effectiveness analysis of two health management strategies for autosomal dominant polycystic kidney disease (ADPKD): current vs. preventive intervention with PGT<sup>1</sup>

<sup>1</sup>PGT, preimplantation genetic testing; TCKF, treatments for chronic kidney failure

## Summary

The diagnosis of HD is clarified thanks to the impulse of Genetics, and its approach from the cost-efficient scenario of the preventive strategy through PGT supports the economic sustainability of PHS. All this should be better known by health professionals.

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None.

## Conflicts of interest

Authors declare that there is no conflict of interest exists.

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