Tables

Table 1: Markers used for microsatellite homozygosity mapping

Loci	Markers
NPHP1	D2S1890, D2S1893, D2S1888, D2S1896
NPHP3* and *NPHP5	D3S3513, D3S3709, D3S1267, D3S1292, D3S1596, D3S1290, D3S1238
NPHP4	D1S2660, D1S2795, D1S2633, D1S2870
NPHP6	D12S88, D12S1719, D12S1598, D12S1678

Table 2: Clinical and Biological features of the report cases at time of NPH diagnosis

Families	***A***	***B***		***C***	***D***	***E***	***F***	***G***	***H***	***I***
Patients	1	2	3	4	5	6	7	8	9	10
Sex /Agge (years)	F/11	F/11	M/9	M/15	F/9	F/14	M/11	H/24	F/11	F/11
Consanguinity	(+)	(+)	(+)	No	(+)	(+)	(+)	(+)	(+)	(+)
Growth failure	(+++)	(+)	(++)	(-)	(-)	(+++)	(++)	(-)	(+)	(++)
Polyuria	(-)	(-)	(-)	(-)	(-)	(-)	(+)	(-)	(+++)	(++)
Enuresia	(+)	(-)	(-)	(+)	(-)	(-)	(+)	(-)	(-)	(-)
Blood pressure	Nle	Nle	Nle	Nle	Nle	Nle	Nle	Nle	Nle	Nle
Hyponatremia	(+)	(-)	(++)	(-)	(-)	(+)	(-)	(-)	(-)	(+)
diurese cc/Kg/d	3.3	3.4	ND	3.6	2	2.6	4.7	ND	3	3.4
Hb g/dl	7.7	8	12	12.9	8.9	5.4	6.3	12	7.3	9
Proteinuria and Hematuria	(-)	(-)	(-)	(-)	(-)	(-)	(-)	(-)	(-)	(-)
Clr-creat mmol/min/1.73m^2	16	23.4	19	23	51	19	51		10	12
Cysts	(-)	(-)	(+)	(+)	(+)	(-)	(-)	(-)	(-)	(++)
Osmolarity mmol/L	241	158	ND	222	290	242	290	ND	186	120
Extra-renal manifestations	No	RP, D	OC, D, JS, FD	(-)	D, RP MR, VH	(-)	MR FD	(-)	(-)	D, RP MR
Gene alteration	*NPHP1* (HD)	*NPHP1* (HD)	*NPHP1* (HD)	*NPHP1* (HD)	*NPHP1* (HD)	*NPHP1* (HD)	*NPHP1* (HD)	*NPHP1* (HD)	*NPHP4* p.H1363P, Hom	*NPHP4* exon 2&3 HD
Actual state	*Hd*	*Hd*	*Hd*	*Hd*	*Hd*	*Hd*	*Hd*	*Hd*	*Hd*	*Hd*
families members detected/ ages (years)	***3*** *(2/4/8)*	**1** *(4)*		**1** *(25)*	**3** *(25,27, 32)*	**1** *(8)*	**1** *(9)*	**O**	**3** *(25,27,28)*	**6** *(18,20,21, 16,20,26)*

FIGURES

F female **; M** Male**; MR**, : Mental retardation **; VH :** Vermis Hypoplasia **; D** : Deafness **; FD** : facial dysmorphy**; RP**: retinal pigmentosa**; JS**: Jouber Syndrom**; HD** homozygous deletion**; Hd** hemodialysis**; OC:** Ocular alterations.

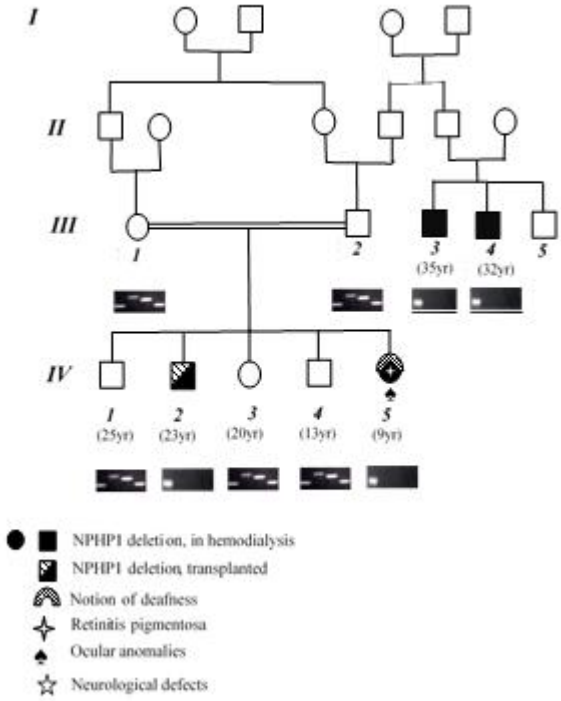

Figure 1: Pedigree analysis in family ***D***, presence of homozygote deletion of *NPHP1* gene with a notion of deafness, RP, neurological defects in patient IV-5. The cases III-3, III-4 and IV-2 developed ESRD in adulthood.

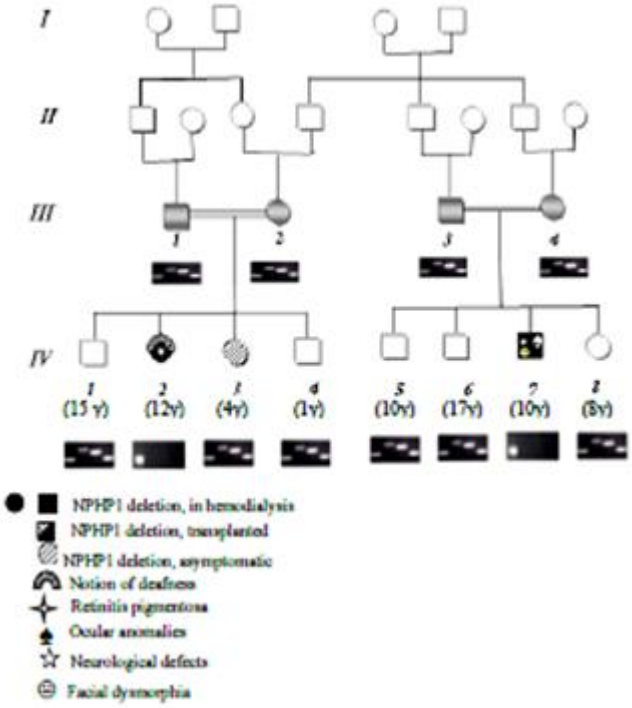

Figure 2: Pedigree analysis in family ***B***, presence of homozygote deletion of *NPHP1* gene with extra renal anomalies. A notion of deafness, RP, mental retardation were detected in patient *FB*-1 (IV-2). Facial dismorphia, ocular anomalies and neurological defects (JS) were present in patient *FB*-2 (IV-7).

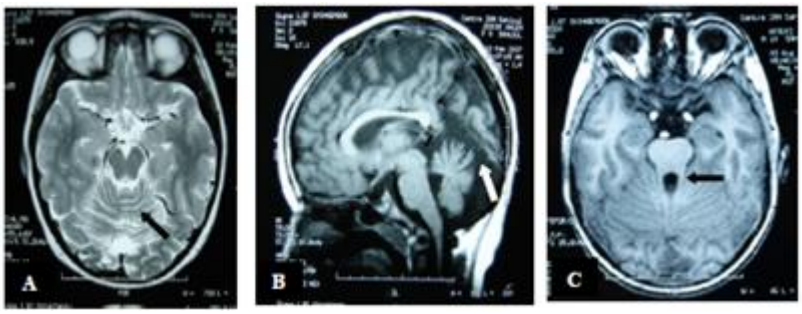

Figure 3: MRI of patients *FD*-1 (A, B) and *FB*-2 (C). **A**: MRI axial sections at the pontomesencephalic level. **B**: Paramedian sagittal sections showing cerebellar vermis hypoplasia in patient *FD*-1. **C** MRI of patients *FB-2*: Paramedian sagittal sections showing a "molar tooth sign" characteristic of Jobert syndrome.

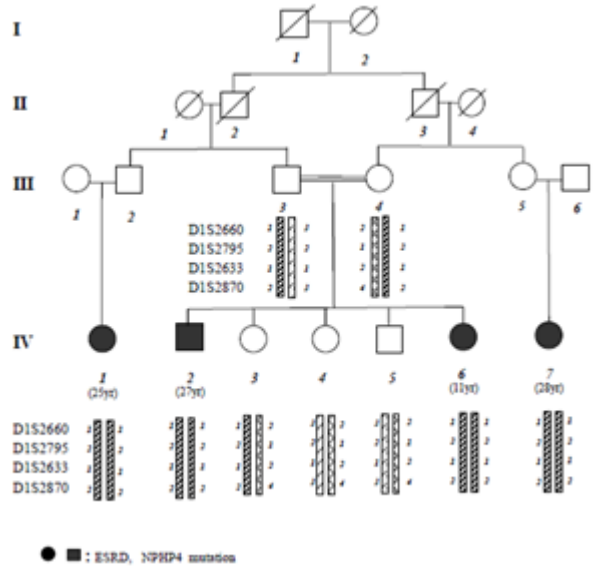

Figure 4: Pedigree analysis in family ***H*** microsatellite homozygosity mapping for *NPHP4* gene, and presence of the mutation His1363Pro in homozygous state in exon 29.

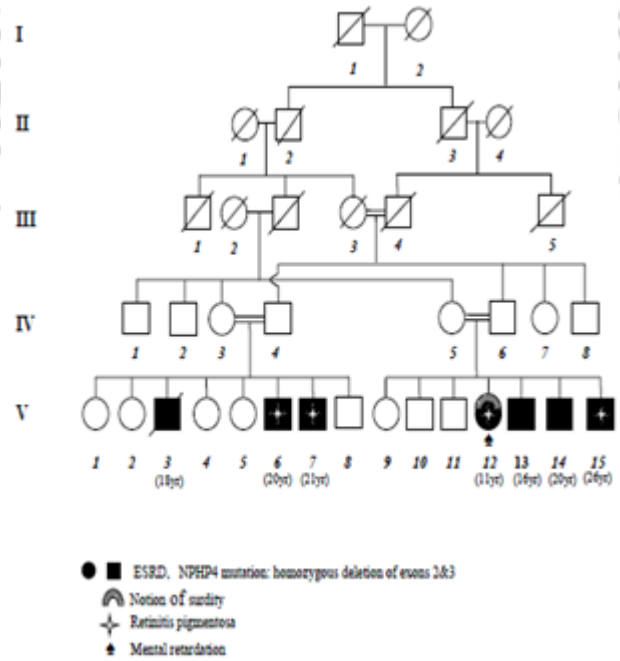

Figure 5: Pedigree analysis in family ***I***, presence of homozygote deletion of exons 2and 3 in *PHP4* gene. RP was present in 4 patients. The index case V-12 have RP, mental retardation and deafness