Hereditary Spastic Quadriparesis Associated with Proteinuric Kidney Disease

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INTRODUCTION

- In 1990 a familial association of proteinuric kidney disease with spastic quadriparesis was reported in two sisters.
- Both the proband and her sister presented with advanced chronic kidney disease and spastic quadriparesis in their mid-30s.
- They progressed to end-stage kidney disease 2 years after presentation.
- We report a follow-up of this family and describe the phenotype across 3 generations.

OBJECTIVE

- We aimed to identify any family members who exhibited any renal or neurological manifestation of this syndrome.

PATIENTS & METHODS

Study Design:
- Family based screening study.

Sample Size
- In December 2012 all family members were invited to participate in the study.
- 20 of 28 family members agreed to participate and provided informed consent.

Data Source
- Electronic patient database, hospital laboratory & radiology systems, biopsy reports.

Data instrument collected data on
- Demographics
- Clinical health at biopsy
- Laboratory values
- Biopsy details

Renal Biopsy Details
- Indications for biopsy
- Operator details
- Pathological diagnosis

Clinical Variables

Demographic
- Age, sex,

Laboratory values
- Creatinine (µmol/L) pre-biopsy
- eGFR (MDRD)

Disease Manifestations:
- Renal: Proteinuria if found on urinalysis and quantified by albumin-to-creatinine or protein-to-creatinine ratio.
- Neurological: Presence of any abnormal findings on neurological examination especially features of spastic paresis.

RESULTS

- The pedigree is presented in *Figure 1* and *Table 1* describes the clinical details of affected members.
- Seven members were identified as having a phenotype with subnephrotic range proteinuria, impaired renal function and features of spastic quadriparesis.
- Renal ultrasonography demonstrated small kidneys in cases with advanced renal disease but no structural abnormalities were present.

CONCLUSIONS

- There are seven affected members across three generations with potential autosomal dominant transmission.
- This phenotype of progressive kidney disease associated with spastic quadriparesis represents a unique disease association.

RESULTS

Figure 1.

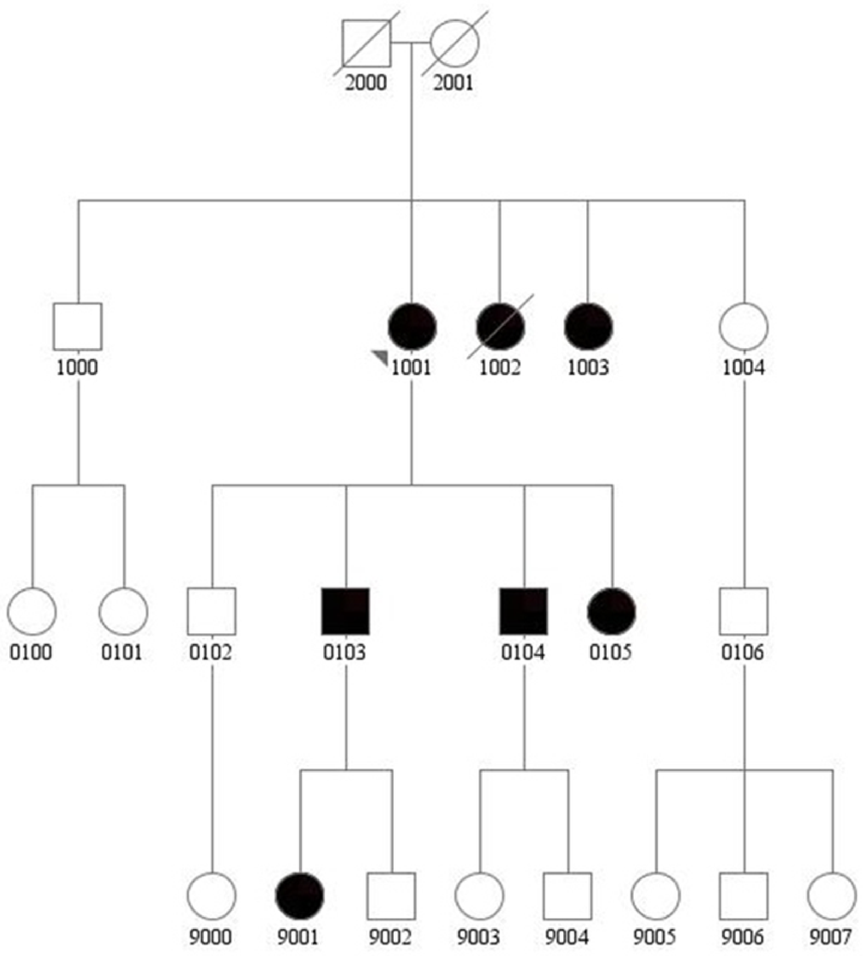

Table 1.

ID	Age at Diagnosis	Renal Status	Proteinuria on Urinalysis	Renal Pathology	Neurological Examination
9001	18	CKD	3+	NA	Spastic Quadriparesis
1002	40	ESKD	3+	NA	Spastic Quadriparesis
1001	36	ESKD	3+	Interstitial Fibrosis & Tubular Cystic dilatation	Spastic Quadriparesis
0103	31	ESKD	3+	Thrombotic Microangiopathy	Normal
0104	32	ESKD	3+	NA	Spastic Quadriparesis
1003	42	ESKD	3+	NA	Spastic Quadriparesis
0105	30	CKD	3+	Thrombotic Microangiopathy	Spastic Quadriparesis

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