

LONG-TERM RENAL OUTCOME OF A LARGE COHORT OF PATIENTS WITH TUBEROUS SCLEROSIS COMPLEX

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Background

Tuberous Sclerosis Complex (TSC)

= autosomal dominant neurocutaneous disorder characterized by the growth of hamartomas in multiple organs

Renal involvement in TSC

= second most important cause of morbidity and mortality at all age

= first common cause of mortality after the age of 30 years

= however, very little is known about the natural history of these renal features in adults and even less in children and adolescents affected by TSC

Materials and methods

-We assessed the clinical records and the renal imaging of TSC patients from two tertiary hospitals in a cross-sectional study

-Demographics, renal phenotype, renal outcome and co-morbidity data were assessed retrospectively

-The purpose of this study is to explore the renal phenotype and long-term outcome in a large TSC cohort

Results

Demographics

Population: 82 TSC patients

50 females (61%) with a male/male sex ratio of 1.6

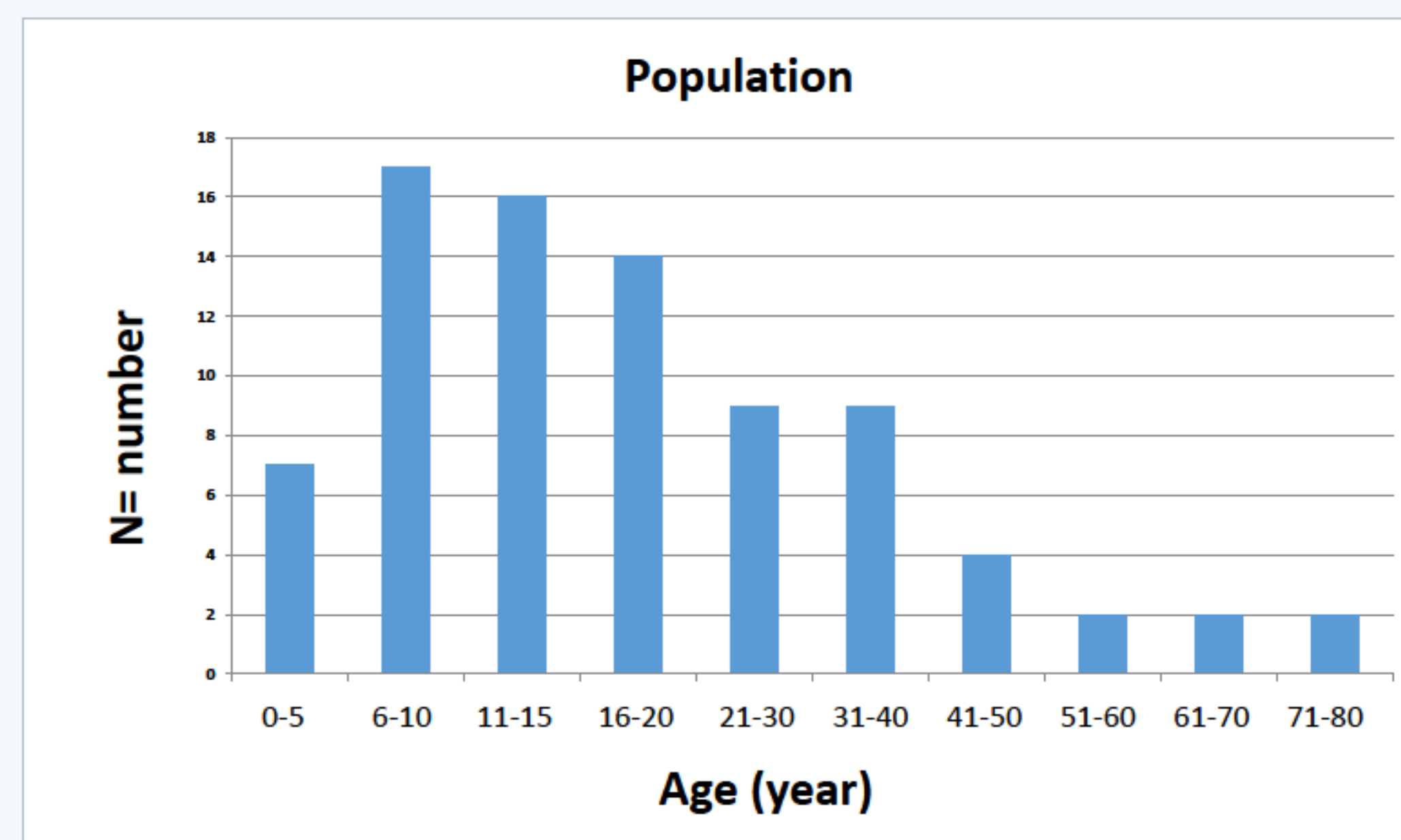
Children (< 18 y): 51 (62%)

Median age at last follow-up was 15.4 y (1.4-72.5)

Median follow-up duration of 11.1 y (0.9-56.6)

Positive family history of TSC in 44%

Prenatal diagnosis in 7 (9%) patients (6 pt with Cardiac rhabdomyoma and 1 pt with renal cysts)



Presenting symptom	N= 82	%
Neurology (epilepsy)	45	55%
Nephrology (bleeding, cysts)	4	5%
Dermatology (skin lesions)	10	12%
Cardiology (rhabdomyoma)	8	10%
Genetic screening	2	3%
Unknown	13	16%

Genetics	N=82	N=62
TSC1	17	21%
TSC2	37	45%
No TSC1/TSC2	8	10%
PKD1/TSC2	2	2%
Unknown/Not done	20	24%

Renal imaging at last FU

-Ultrasound

All ages: 65 patients (82%)

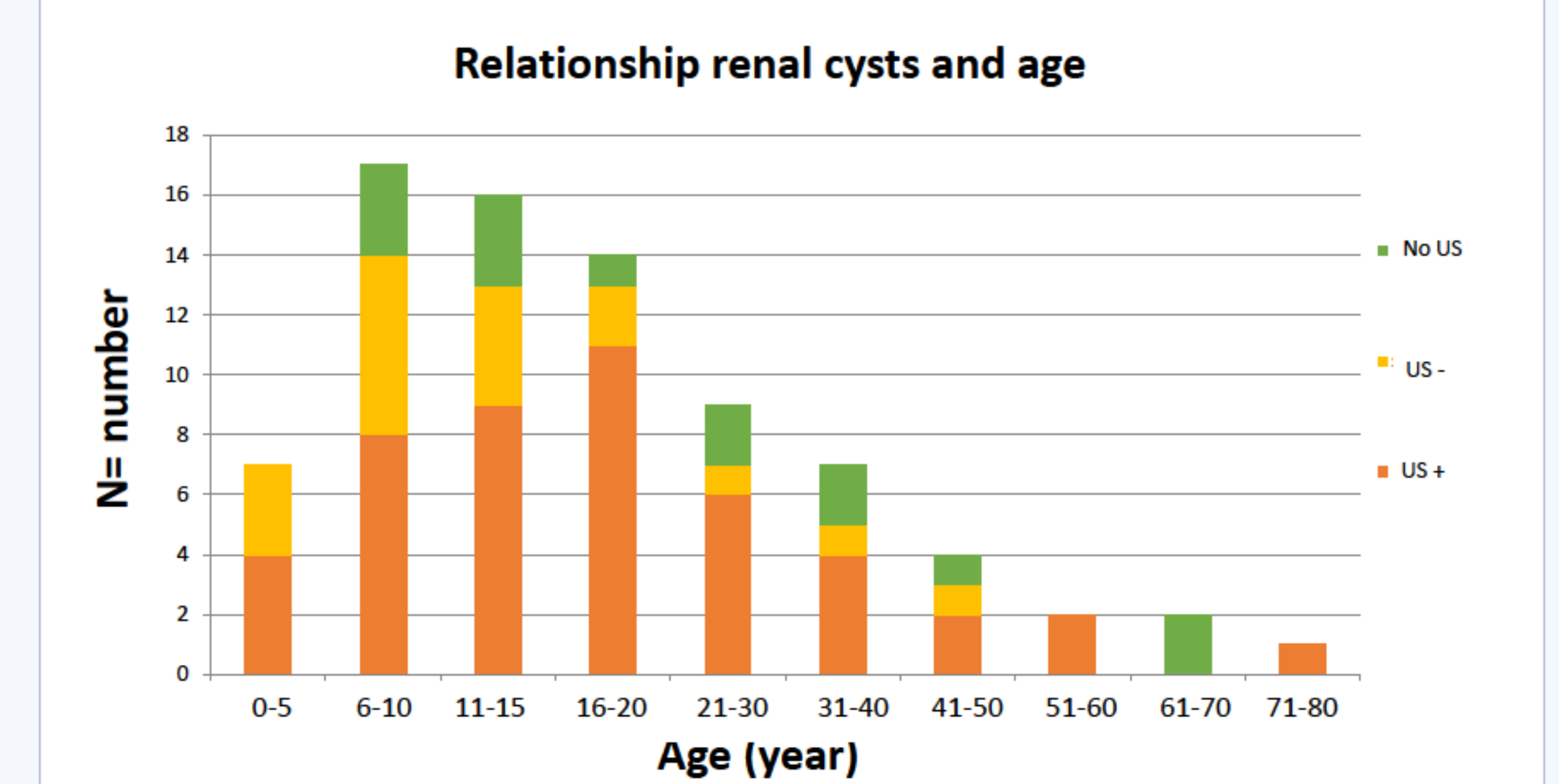
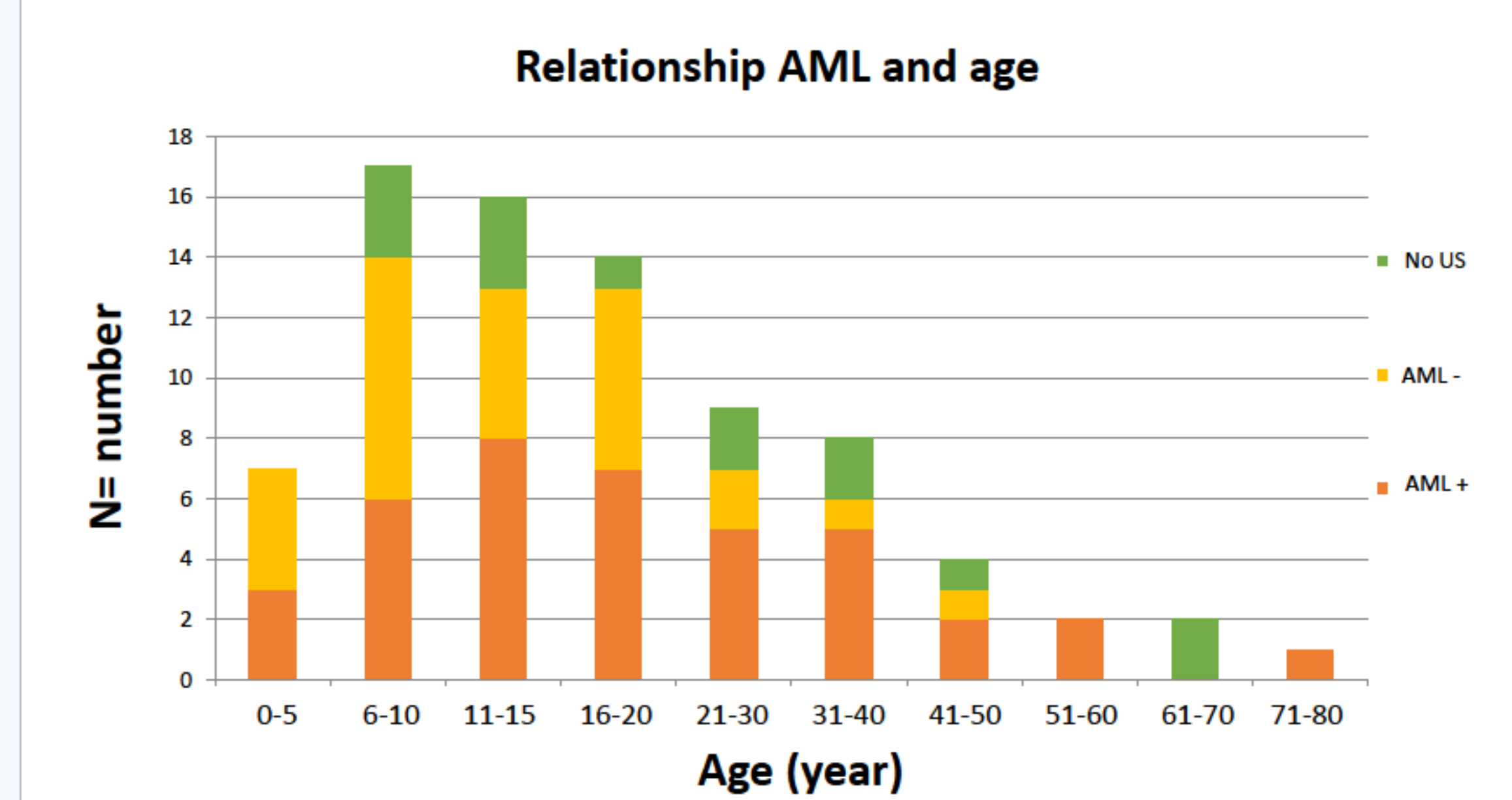
<18 year: 44 patients (86%)

>18 year: 21 patients (75%)

-Renal lesions

AML: 39 (59%) patients

Renal cysts: 36 (55%) patients



Renal phenotype

-Renal function (N=73)

-eGFR <90 ml/min/m² in 24 (33%) TSC patients

-ESRD in 6 (8%) pts with median age of start RRT: 47.0 y (21.0-64.0)

3/6 patients had a nephrectomy due to RCC

-Hypertension (N=60)

in 14 (23%) of all TSC patients and in 5 (15%) TSC children

-Proteinuria (N= 41)

in 6 (15%) of all TSC patients and in 1 (5%) TSC children

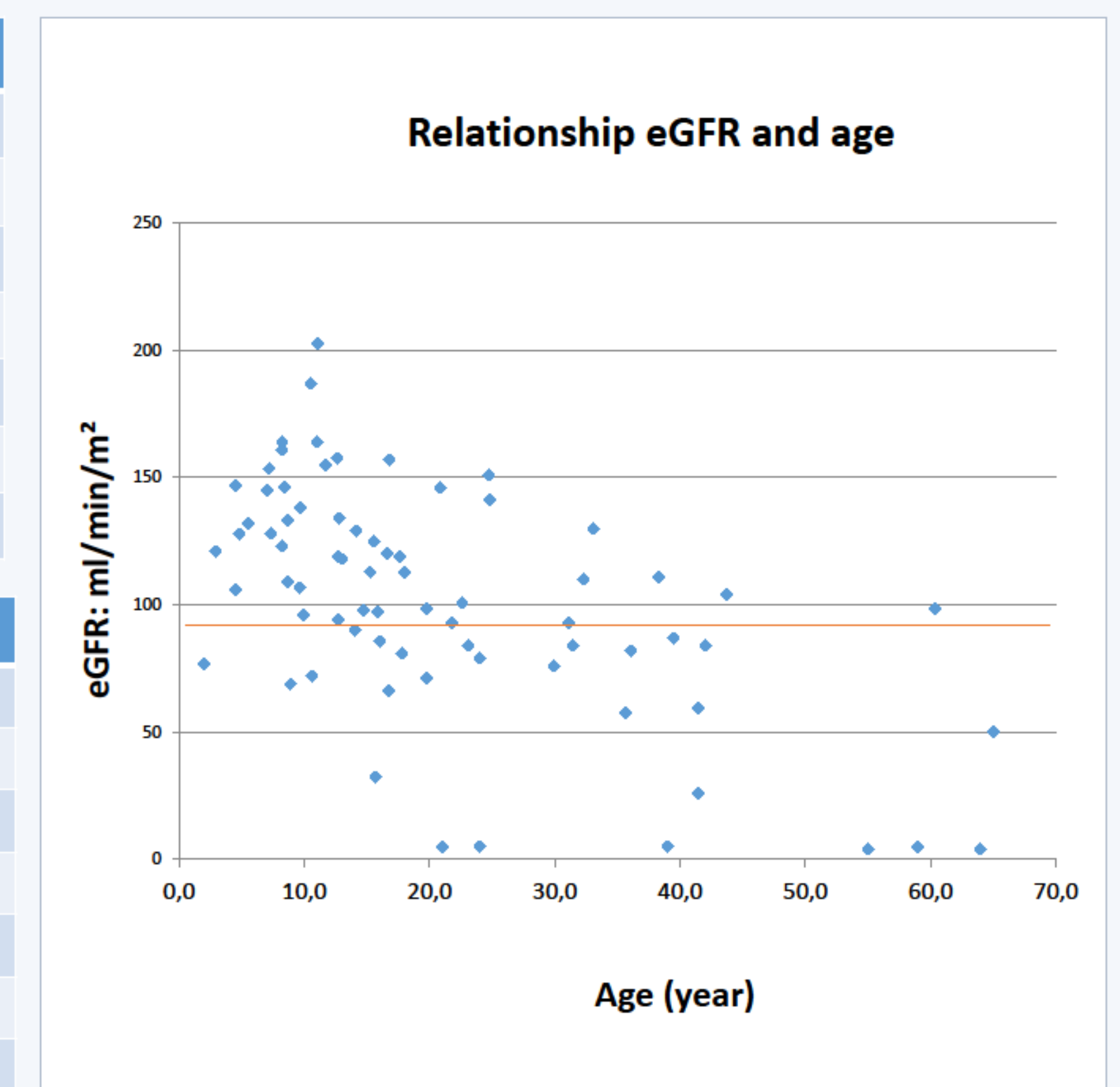
-Renal complications: in 9 (11%) patients

-bleeding AML (4 pts; need of embolization), RCC (7 pts; need of (part) nephrectomy, marsupilation (1 pt due to renal cyst of 20cm)

-eGFR <90 ml/min/m²: 7 patients, eGFR <30 ml/min/m²: 5 patients

CKD stage	N=73	%
No renal lesion	21	29%
CKD ≥90	27	37%
CKD2 ≤89- ≥60	14	19%
CKD3 ≤59- ≥30	3	4%
CKD4 ≤29- ≥15	1	1%
CKD5 ≤15	6	8%
eGFR ≤90	24	33%

CKD stage	All ages	Children	Adults
No renal lesion	29%	33%	23%
CKD ≥90	37%	51%	17%
CKD2 ≤89- ≥60	19%	14%	27%
CKD3 ≤59- ≥30	4%	2%	7%
CKD4 ≤29- ≥15	1%	0%	3%
CKD5 ≤15	8%	0%	20%
eGFR ≤90	33%	16%	60%



Conclusion

This study describes the long-term renal outcome in a large cohort of TSC patients.

Our findings confirm the high rate of renal involvement in TSC.

The number of renal lesions at last follow-up accounts for 47 (72%) patients. Renal AML were seen in 39 (59%) and renal cysts in 36 (55%) patients.

Renal function (N=72) showed that the eGFR was <90 ml/min/m² in 24 (33%) TSC patients. ESRD was found in 6 (8%) patients.

Hypertension (N=52), proteinuria (N=31) and renal complications (bleeding, RCC or surgery) were found in 12 (23%), 5 (16%) and 8 (10%) patients.

Therefore, we advocate regular renal surveillance of these patients for the timely and optimal managing of the renal co-morbidities.

