NEPHROCALCINOSIS AND MICROCYTHEMIA: AN EMERGING PROBLEM

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Objectives:

Nephrocalcinosis is a clinical-pathological entity caused by various hereditary or acquired diseases that lead to the deposition of calcium salts within the kidney parenchyma. Epidemiological data are scant; the protean presentation may explain the lack of data regarding its prevalence. The aim of this study is to report the prevalence and main clinical features of the cases of nephrocalcinosis diagnosed in a newly opened Nephrology Out-patient Unit with particular regard for the cases related to microcythemia.

Methods:

We carried out a retrospective analysis on the data we prospectively gathered from the start of our activity in December 2007 until December 2013. Clinical and laboratory data were collected from the medical records and from the general laboratory; diagnosis was based upon imaging data (mainly ultrasounds) reviewed by the same Radiologists.

Results:

About 2.5% of the patients referred to our Unit were diagnosed with nephrocalcinosis; the disease was associated with autoimmune disorders in 29% of patients and with microcythemia in 23%, while positive family history was present in 23% of patients.
Microcythemia patients were referred to us due to the presence of kidney stones, electrolyte derangements, mild proteinuria or history of kidney urinary tract infections or malformations. 17 patients are affected by beta thalassemia (10 males and 7 females), while 4 patients are affected by sickle cell disease (all females). Median age is low: 35 (22-46) for the subset with beta thalassemia and 35.5 (30-47) for the other subset. Kidney function is preserved in both groups with median Crs of 0.73 mg/dL (0.51-1.09 mg/dL) in thalassemic patients and 0.63 mg/dL (0.51-0.76 mg/dL) in patients with sickle cell disease.
The biochemical profile of Beta thalassemia patients is homogeneous and is characterized by severe hypercaciuria which is often concomitant with increased FePi. The four patients with sickle-cell disease do not display hypercalciuria, in keeping with the different pathogenesis (calcifications on parenchymal microinfarctions). (Table)

Name	Sex	Age	Underlying disease	Kidney stones	Other diseases	sCr mg/dL	GFR EPI/Shwartz mL/min	PtU g/day	CaU mmol/day	CaU mg/kg/day	Fe Ca %	Fe Pi %	Iron-chelator
AD	M	22	β thal	1 (Ca Ox)	Tetraplegia	0.59	147	0.13	6.22	7.54	2.2	26.7	Desferal
BM	M	35	β thal	1(Ca Ox)	Cryo III	0.73	120	0.14	6.53	4.75	1.9	11.3	Deferasirox
CL	M	35	β thal	1	Previous AKI	0.85	112	0.25	14.16	7.87	2.5	26.5	Desferal
CA	M	38	β thal	1	-	0.76	113	0.125	9	9	2.0	21.7	Deferasirox
DM	M	34	β thal	0	-	0.68	123	0.15	8.65	6.04	1.5	10.6	Desferal
DSM	M	33	β thal	0	-	1.01	113	0.8	10.36	7.97	3.7	22.1	Deferasirox
DE	F	41	β thal	0	Cryo III, previous APN	0.68	108	0.17	14.2	13.2	5.0	27.9	Deferasirox
FA	F	33	β thal	1 (Ca Ox)	-	0.64	120	0.07	9.53	9.39	3.6	30.4	Desferal
MV	F	31	β thal	0	Mental retardation	0.51	140	0.31	5.11	3.65	1.9	12.8	Deferasirox
SS	F	28	β thal	0	-	0.69	118	1.01	17.32	12.48	4.5	9.3	Deferasirox
VEM	M	34	β thal	1	Cryo III	0.85	117	0.16	10.3	5.8	2.4	14.2	Deferasirox
FG	M	44	β thal	0	IgAGN	0.73	104	1.7	6.43	3.96	1.5	13.6	Deferasirox
SR	F	46	β thal	1	HCV	0.6	109	0.1	7.5	4.4	1.7	8.3	Deferiprone
MM	F	35	β thal	0	-	1.05	69	0.3	8	6.6	4.2	47.9	Deferasirox
RL	M	35	β thal	1	HCV	1.09	88	0.4	5.4	4	1.7	23.7	Deferasirox
LPC	F	37	β thal	0	APS, HCV	0.8	94	0.9	9.4	5.6	2.2	15.4	Deferasirox
LA	M	36	β thal	0	Gilbert, HCV	0.79	116	0.1	6.7	4.2	1.7	14.3	Deferasirox
All pts β thal (median)	-	-	-	-	-	**0.73 (0.51-1.09)**	**113 (69-147)**	**0.17 (0.07-1.7)**	**8.7 (5.1-17.32)**	**6.04 (3.65-13.2)**	**2.2 (1.5-5)**	**15.4 (8.3-47.9)**	-
BP	F	31	SKD	0	-	0.51	129	0.455	1.95	1.22	0.5	11.0	Deferasirox
RMJ	F	47	SKD	0	-	0.68	116	1.3	n.a.	n.a.	0.3	12.7	Desferal
SPCM	F	30	SKD	0	-	0.58	124	0.2	3.9	2.4	0.9	7.7	-
UM	F	40	SKD	0	HIV	0.76	114	1.7	<0.5	0.4	0.3	9.7	Deferasirox
All pts SKD (median)	-	-	-	-	-	**0.63 (0.51-0.76)**	**120 (114-129)**	**0.9 (0.2-1.7)**	**1.95 (0.5-3.9)**	**1.22 (0.4-2.4)**	**0.4 (0.3-0.9)**	**10.35 (7.7-12.7)**	-

Legend: sCr serum Creatinine; **GFR** Glomerular Filtration Rate; **PtU** 24 h Proteinuria; **CaU** Urinary Calcium; **Fe Ca** Calcium Excretional Fraction; **Fe Pi** Phosphate Excretional Fraction; **Cryo** Cryoglobulinemmia; **VUR** Vesicoureteral Reflux; **AKI** Acute Kidney Injury; **SKD** Sickle Cell Disease; **APN** Acute Pyelonephritis; **GN** Glomerulonephritis

Conclusions:

Nephrocalcinosis is a rare, but not exceptional disease in the Nephrological practice. Microcythemia appeared to be a major cause, probably also better diagnosed on the account of the recent interest for a detailed analysis of kidney function related to the surveillance of the new oral iron chelators. Better awareness of this disease points to the need for an integrated approach involving various branches of Internal Medicine and Radiology.

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H) Clinical Nephrology, CKD. H1) Lab methods, progression & risk factors for CKD 1-5, nutrition in CKD, renal diseases (except GNs and cystic diseases).
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