



**SATELLITE MEETING
OF THE 40TH COURSE
ON ADVANCES IN NEPHROLOGY,
DIALYSIS AND TRANSPLANTATION
DECEMBER 5, 2008
ROOM WASHINGTON A**



**FIRST JOINT MEETING ON CLINICAL GENETICS
OF RENAL DISEASES**

BY FONDAZIONE D'AMICO PER LA RICERCA SULLE MALATTIE RENALI
AND ASSOCIAZIONE ITALIANA RENE POLICISTICO

(CME credits have been requested)

Morning	POLYCYSTIC KIDNEY DISEASE
Chairmen:	<i>L. RAMPOLDI (Milan)</i> <i>F. SCOLARI (Brescia)</i>
09.00-09.35	A.B. CHAPMAN (Atlanta) Renal manifestations
09.35-10.10	T. WATNICK (Baltimore) Extrarenal manifestations
10.10-10.45	G. GERMINO (Baltimore) Animal models
10.45-11.20	A. BOLETTA (Milan) Unravelling the function of polycystins
11.20-11.55	G. REMUZZI (Bergamo) Therapeutic clinical trials
11.55-12.45	DISCUSSION
12.45-14.00	AIRP LUNCH MEETING
Afternoon	COMPLEX TRAITS
Chairmen:	<i>D. CUSI (Milan)</i> <i>F. SCOLARI (Brescia)</i>
14.00-14.35	A.G. GHARAVI (New York) Genetics of IgA Nephropathy
14.35-15.10	S. SANNA CHERCHI (New York) Congenital Anomalies of the Kidney and Urinary Tract
15.10-15.45	G.M. GHIGGERI (Genoa) Genetics of FSGS
15.45-16.45	DISCUSSION

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Associazione Italiana Rene Polmonare
ONLUS



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*Nevertheless, in the
much promise is expected
effective therapy for a
untreatable disease*

86





Marriott
MILAN

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CENTRO CONGRESSI

AIRP

G. Rizzuto
MILAN

untreatable
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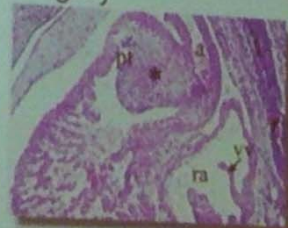
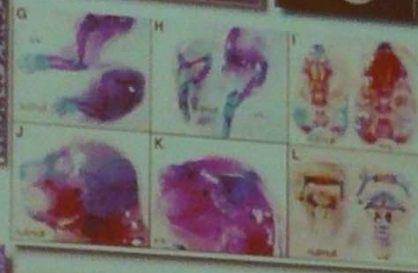
F. Scolari
M.D.

L. Rampoldi
M.D.

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Associazione Italiana
Riabilitazione Psichiatrica

Pkd1 is important for:

- Placenta
- Kidney
- Heart
- Skeleton
- Pancreas
- Biliary system
- Vascular system
- Abdominal wall integrity



Lu et al, *Hum Molec Genet* 10:2385-96, 2001
Boulter et al, *Proc Natl Acad Sci* 98:12174-79, 2001
Unpublished observations



Missense mutations can result in functionally "null" alleles

- ENU Mutant
 - M3091R
 - Transcribed into mRNA
 - Situated within first TM
 - Phenotype: looks like *Pkd1*^{-/-}
 - Edema
 - Renal cystic disease
 - Embryonic lethal



Hemon et al, Nature Genet 30:185-189, 2002

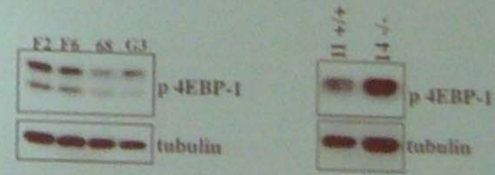
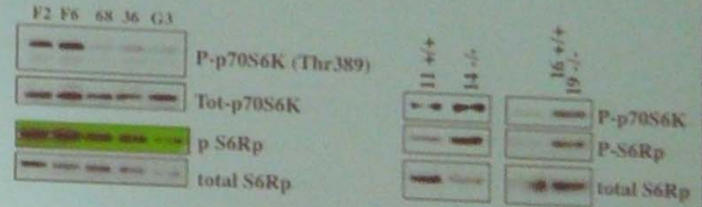
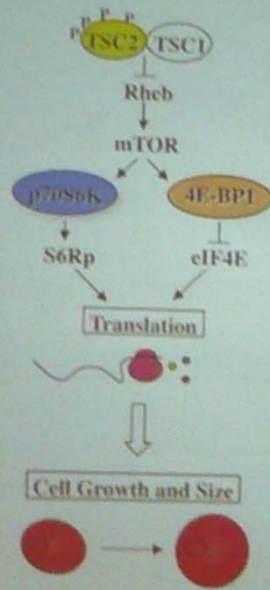




- Respiratory epithelium
- Epididymis, vas deferens
- Leydig cells
- Placental trophoblasts



PC-1 Controls Cell Size Through p70S6K/S6Rp and 4EBP1/eIF4E



Marriott
MILAN

AMP



A. Boletta
M.D.



AIRP



G. Remuzzi
M.D.

*Nevertheless, in the next decade
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effective therapy for a so
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A.R. Chapman
M.D.

T. Wainick
M.D.

AIRP

Germino
M.D.

A. Boletta
M.D.

G. Amici
M.D.



Man on the left: Grey suit, light blue shirt, red tie. Name tag visible on the left lapel.

Man on the right: Grey suit, white shirt, red patterned tie. Name tag visible on the right lapel. The name tag contains the following text:
[Logo]
[Name]
[Title]
[Barcode]



G. Germino
M.D.

A. Boletta
M.D.





G. Germino

A. Boletta
M.D.



ANNUAL
FRA COSTA FELLOWSHIP

CME STUDIOS

A. B. Chapman
M.D.

