



# Patients Perspectives on Common Elements in Uncommon Kidney Diseases

Daniel Renault Chair

FEDERATION OF EUROPEAN ASSOCIATIONS OF PATIENTS  
AFFECTED BY  
GENETIC and/or RARE RENAL DISEASES

**FEDERERG**

# Disclosure of Interests

NO interests to declare..... apart the interest for patients

FEDERG = volunteers

Budget = fees only & EURORDIS grant

KDIGO



Polycystic  
Nephrotic Syndrome  
Alport Syndrome  
Barrter and Gitelman  
Cystinosis  
aHUS  
Hypomagnesemia



FEDERG: Federation of  
15 national European associations  
– single pathology or umbrella  
organizations

## 8000 Rare Diseases.....IMPORTANCE of UMBRELLA ASSOCIATIONS

- **Solidarity** among patients: different conditions but **commonalities**
- **Together stronger:** sharing resources
- **Single patient representative** (not 100+) for Public Health authorities and for Professionals
- **Independence** (Not relying on a single supporting Pharma)

National Associations for Research and Information on Renal Genetic Diseases: AIRG-France in 1988 then Belgium, Spain, and Switzerland.

European Federation FEDERG was launched in 2012 to cover Rare/ Genetic Renal diseases in Europe.

# FEDERG Organization: Volunteers with a professional purpose

## FEDERG Registration Legal seat



### ADDRESS

info@federg.org



### WEBSITE, FACEBOOK & MORE

federg.org

Federg aisbl



### AISBL BRUSSELS

Association internationale sans but lucratif  
Legal seat: Brussels Cliniques  
Universitaires

*KDIGO Controversies Conference on Common Elements in Uncommon Kidney Diseases*  
June 16 - 19, 2016 | Amsterdam, Netherlands



**FEDERG**

# Governance: Board of FEDERG - Monthly

Teamwork is everything



Daniel Renault  
President – France



Claudia Sproedt  
Vice President – Germany



Bruno Woitrin  
Treasury – Belgium



Michel Schenkel  
Secretary – Belgium



Susie Gear  
United Kingdom



Francisco Montfort  
Spain



Tess Harris  
United Kingdom



# Scientific Council

Knowledge from the specialists

## Scientific Council

- Chair: Olivier Devuyst
  - Vice-Chair: Francesco Emma
  - Dominique Chauveau
  - Pierre Cochat
  - Oliver Gross
  - Jean-Pierre Guignard
  - Katharina Hohenfellner
  - Nine Knoers
  - Elena Levtchenko
  - Rachel Lennon
  - Albert Ong
  - Yves Pirson
  - Franz Schaeffer
  - Vladimir Tezar
  - Roser Torra
- ERA EDTA  
Italy  
France  
International INPA  
Germany  
Switzerland  
Germany  
The Netherlands  
Belgium and ESPN  
United Kingdom  
United Kingdom  
Belgium  
Germany  
Czech Republic  
Spain

# Membership – Assembly – Members' meeting

## Membership

- Full members are legal patient organization of the EU28: one vote per full member
- Associate members are members not from EU28: they are fully associated to federation activities (without vote)





# FEDERG Main Supports

We are part of the movement

## Well connected:

- EURORDIS (European Organization for Rare Diseases) strong support to develop our capacity
- ERA-EDTA European Renal Association – European Dialysis and Transplant Association
- ESPN: European Society of Paediatric Nephrology
- KDIGO: Kidney Disease Improving Global Outcomes -Guidelines



*KDIGO Controversies Conference on Common Elements in Uncommon Kidney Diseases*  
June 16 - 19, 2016 | Amsterdam, Netherlands





AIRG/FEDERG movement = a long experience of sharing common concerns, hopes and commitments by patients affected by R/G Renal disorders.

FEDERG has been involved in several previous KDIGO conferences:

Gitelman, aHUS, ADPKD, Cystinosis.

In 2015, EUROPEAN initiative for rare diseases: launching the ERNs (European Reference Network)

One of the 21 themes of ERN is **Rare Renal**: Thus FEDERG is legitimate and fully engaged to represent the European patients within the ERKNET project (European Reference Network for Rare Renal).



# Introductory Remark 1: The Race between Aggregation and Partition

Recent progresses in genetic investigation (NGS) lead to divide the conditions, into sub categories.

Must be counterbalanced by a strong effort to aggregate and synergize forces.

Heavy strain on the numbers of patients for study and trials to reach critical mass for each sub-category.

Pressure on the prognosis side (lagging behind diagnosis)!

We need to work hard on the commonalities! We need to involve much more patients to address these challenges.



## Introductory Remark 2: Common and Uncommon: the Type of Gene Transmission

One generation ago we were all sharing the same “out of the blue” diagnostic no matter what the process link to the default.

Today our patients are **partitioned into 3 groups**:

The group of “dominant transmission” for which the disease is known and will be transmitted.

The group of “recessive transmission”: typically not known till the first child affected

The group of de novo mutation: not known at all

And then grouped with the later, the idiopathic rare renal disease.



## Introductory Remark 3: The Fight is for both Common and Uncommon

Common? What does that mean? Common to all conditions ? To few ?

Identification of the commonalities is critical BUT should not lead to put aside the specificities OR to isolate ultra-rare diseases.

Commonalities and specificities are our common fight in FEDERG.

As father of a young woman affected by ALPORT syndrome, I am concerned by the problem of bad breath of teenagers with cystinosis, by the discomfort of polycystic women with a belly of pregnancy type.

**OUR main commonality is a sense of solidarity**

**We fight for each one and for all !**



# Common Issues, Common Questions and Common Responses !

Mapping common issues of Rare/Genetic kidney disorders

*Questions:* My future? How do I see/build it? How do I overcome the hurdles?

*Responses:* Medicine improves progressively medical responses!

However beyond GFR, how about my life? Which part relies on Public Health policy and which part on the social system?

What is my contribution in these responses?



# Mapping the Common Issues of Uncommon Kidney Diseases

## Features of the Conditions: Rare/Orphan-Genetic-Pediatric

### **RARE/ORPHAN**

Not well known !

Not always known by local doctors !

Often a diagnosis odyssey – Weak prognosis

Isolation

Often no treatment yet

High cost medicine (if any)

Insurance (high fees)



# Mapping the Common Issues of Uncommon Kidney Diseases Rare-Genetic-Pediatric

## GENETIC

Complex/multigene pathologies,

Multi-organ pathology

Family problem

Transmission, Medically Assisted Procreation (MAP)

Long life disease before ESRD

Declaration: Problems of insurance, access to job, ...

Sharing of genetic information: family, incidental findings





# Mapping the Common Issues of Uncommon Kidney Diseases Rare-Genetic-Pediatric

**PEADRIATRIC** (for about 2/3 of the R/G Kidney patients)

Infant (sometimes before birth)

Education, sports

Diets (schools, cantinas)

Frequent and numerous medicines

Side effect of treatments (fatigue, bad breath, ...)

Transitions (teen-age adult, dialysis to transplantation)

Parents involvement, strain on the family

Growing up with renal diseases (RRT)



# Mapping the Common Issues of Uncommon Kidney Diseases

## Commonalities in Treatments

### **Existing treatments:**

Dialysis, transplantation, nephroprotection, compensation.  
The heavy load of treatments on personal life.

**The hope for improvement of treatments, and for innovative treatments.**



# Mapping the Common Issues of Uncommon Kidney Diseases

## Commonalities in Quality of Life

**Quality of life (all aspects).....**

**SURVEY ARE RARE**

Paediatric, Adult and the (g)old age.

Education (Constraints, support, prospects & achievements when already under RRT).....

Education for adult transplanted in their childhood (308 patients) Chantal Loirat et al..	Reported rate %	General population %	p
<b>No diplôme</b>	<b>20.5</b>	<b>12.3</b>	<b>&lt; 0.01*</b>
CEP, BEPC	7.5	5.7	0.22
BEP, CAP	20.1	20.1	0.97
Bac ou équivalent	21.4	22.2	0.81
Bac+2	16.6	18.2	0.53
<b>≥ Bac+3 (High education BSc +)</b>	<b>14</b>	<b>21.6</b>	<b>&lt; 0.01#</b>

# Mapping the Common Issues of Uncommon Kidney Diseases

## Commonalities in Quality of Life

### Quality of life (all aspects)..... WORK

Access, remuneration, declare health conditions, treatments? (Pb with health insurance). Coping with work stress?

Employment for adult transplanted in their childhood Chantal Loirat et al..	Reported %	General population %	p
<b>Unemployed (N=325)</b>	<b>18.5</b>	<b>10.4</b>	<b>&lt;0.01</b>
<b>Employed with contract (N=199)</b>			
•Apprentissage (young training)	7	3.1	<0.01
•Short term	21.1	11.8	<0.01
•Long term	66.8	81.8	0.02
<b>Part-time employee(N=184)</b>	<b>22.8</b>	<b>16.7</b>	<b>0.05</b>



# Mapping the Common Issues of Uncommon Kidney Diseases

## Commonalities in Quality of Life

### Quality of life (all aspects).... Personal

Personal life (single, married)

Impact (psychological aspects)

Impact on the whole family (pediatric)

Genetic & ethics : share or not to share Information ? Duty of knowledge

# Common Questions/Responses for Uncommon KD

## Where are they?

**In the hands of the clinicians and researchers**

**In the hands of Public Health authorities**

**In the hands of pharmaceutical companies**

**In my hands (patients and associations)**

# Our Hope for the KDIGO Workshop

Mapping of the Commonalities of issues/questions with:

- Responses per type of actor
- Priorities of action
- Prepare for a roadmap

KDIGO



*A message from the workshop ?*  
**In my Hands (Patients and Associations)**

**Right & Duty**  
**Health & Life**

RIGHT for the best health possible,

RIGHT for a “quasi-normal life”

NEED for Data on QoL

DUTY to contribute to the knowledge  
development on Health issues and QoL, and on  
the steps for improvements





Thank you for your attention

FEDERGO