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

FEDERG
Disclosure of Interests

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

FEDERG = volunteers

Budget = fees only & EURORDIS grant
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

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AISBL BRUSSELS
Association internationale sans but lucratif
Legal seat: Brussels Cliniques Universitaires

WEBSITE, FACEBOOK & MORE
federg.org
Federg aisbl
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 \( \text{ERA EDTA} \)
- Vice-Chair: Francesco Emma \( \text{Italy} \)
- Dominique Chauveau \( \text{France} \)
- Pierre Cochat \( \text{International INPA} \)
- Oliver Gross \( \text{Germany} \)
- Jean-Pierre Guignard \( \text{Switzerland} \)
- Katharina Hohenfellner \( \text{Germany} \)
- Nine Knoers \( \text{The Netherlands} \)
- Elena Levchenko \( \text{Belgium and ESPN} \)
- Rachel Lennon \( \text{United Kingdom} \)
- Albert Ong \( \text{United Kingdom} \)
- Yves Pirson \( \text{Belgium} \)
- Franz Schaeffer \( \text{Germany} \)
- Vladimir Tezar \( \text{Czech Republic} \)
- Roser Torra \( \text{Spain} \)
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)
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
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).
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.
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.
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!**
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?
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)
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
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)
Existing treatments:
Dialysis, transplantation, nephroprotection, compensation.
The heavy load of treatments on personal life.

The hope for improvement of treatments, and for innovative treatments.
Quality of life (all aspects)........
Paediatric, Adult and the (g)old age.
Education (Constraints, support, prospects & achievements when already under RRT)......

<table>
<thead>
<tr>
<th>Education for adult transplanted in their childhood (308 patients)</th>
<th>Reported rate %</th>
<th>General population %</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>No diplôme</td>
<td>20.5</td>
<td>12.3</td>
<td>&lt; 0.01*</td>
</tr>
<tr>
<td>CEP, BEPC</td>
<td>7.5</td>
<td>5.7</td>
<td>0.22</td>
</tr>
<tr>
<td>BEP, CAP</td>
<td>20.1</td>
<td>20.1</td>
<td>0.97</td>
</tr>
<tr>
<td>Bac ou équivalent</td>
<td>21.4</td>
<td>22.2</td>
<td>0.81</td>
</tr>
<tr>
<td>Bac+2</td>
<td>16.6</td>
<td>18.2</td>
<td>0.53</td>
</tr>
<tr>
<td>≥ Bac+3 (High education BSc +)</td>
<td>14</td>
<td>21.6</td>
<td>&lt; 0.01#</td>
</tr>
</tbody>
</table>
Quality of life (all aspects)........ WORK

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

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