



**More than 100,000 Germans
and 12.5 million people
worldwide suffer from PKD.**



Living with PKD

(What every family needs to know)

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Uwe K.H. Korst

Version 1.2

Living with PKD



- About me
- What is PKD?
- What is it like to live with PKD?
- Anxiety and concerns of parents



About me (before PKD effects)

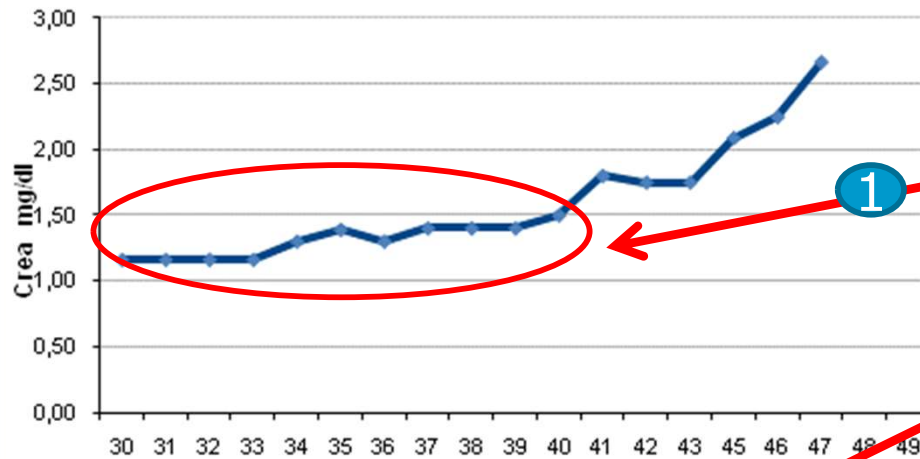
- 47 years old
- Married / two children (19 and 8)
- 4000 km/year with racing bike (endurance sport)
- motorbike
- Senior project manager and security consultant for IT-security projects
- Today: PKD stage IV



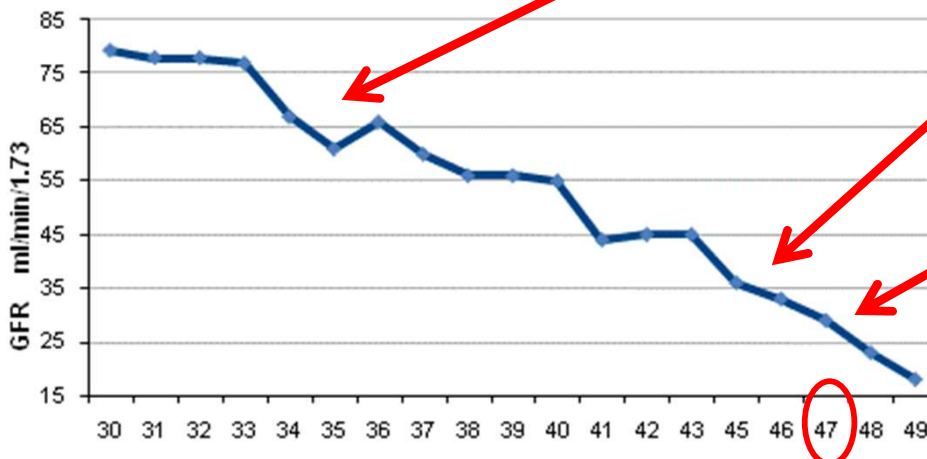
My disease progression

- 15 – Grandmother died from PKD (age: 63)
- 18 – High blood pressure diagnosed
- 27 – PKD first diagnosed
- 33 – Mother died from PKD (53 years old)
- 42 – Several episodes of bleedings into the cysts / blood in urine
- 45 – Participation with a PKD therapy study
- 46 – Stop of endurance sports (cycling, jogging) due to performance problems.
- 49 – ESRD with dialysis (date estimated)
- ???

My disease progression



- Creatinine was stable for many years (norm <1.5 mg/dl), **but....**



- Kidney function (GFR) decreased year by year (stage 3).
- Performance-oriented sports no longer possible (stage 4).
- Pain and fatigue, reduced performance (stage 4)
- ESRD (stage 5)

Effects on my life (stage 4)

- No more endurance sports possible
- Increased Fatigue
- Increasing problems with blood pressure
- Depression
- Reduced performance at work, at home, ...



Living with PKD

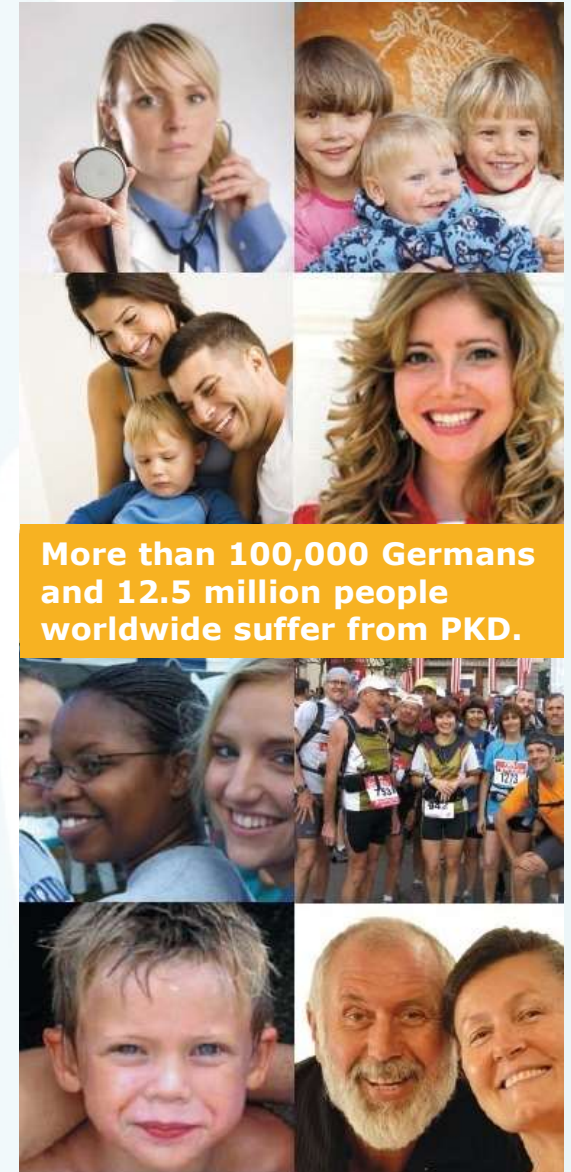
- About me



- What is PKD?

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- Polycystic Kidney Disease (Abbreviation PKD)
 - German: Zystennieren
 - French: Maladie Polykystique des Reins
 - Italian: Malattia Renale Policistica (MRP)

- The disease appears mainly in two genetic forms:
 - ADPKD
 - ARPKD



■ Autosomal Dominant Polycystic Kidney Disease (ADPKD):

ADPKD affects 1 in 500 worldwide. Parents with the dominant form of PKD have a 50% chance of passing the disease on to each of their children.

■ Autosomal Rezessive Polycystic Kidney Disease (ARPKD):

ARPKD is a relatively rare form of PKD that affects 1 in 20,000 babies and often leads to death in the first month of life. Parents who carry the ARPKD gene have a 25% chance of passing the disease on to each of their children.

This is PKD!

- Polycystic Kidney Disease is one of the most common life-threatening genetic diseases.
- Dominantly inherited & doesn't skip generations
- More common than the **combined** number of Europeans with cystic fibrosis, sickle cell anemia, hemophilia, muscular dystrophy, Down's syndrome, and Huntington's disease.
- Estimations are more than 100,000 Germans and 12.5 million people worldwide suffer from PKD.

This is PKD!

■ H₂O Balloons



- Cysts are like genetic “water balloons”, which multiply and grow bigger and bigger.
- And there are many of them.
- Dramatically varied in size.
- A cyst in the kidney begins as an outpouching of the nephron, similar to a blister, and can occur anywhere along the length of the nephron.
- Only 5 % of the nephrons form cysts.

This is PKD!

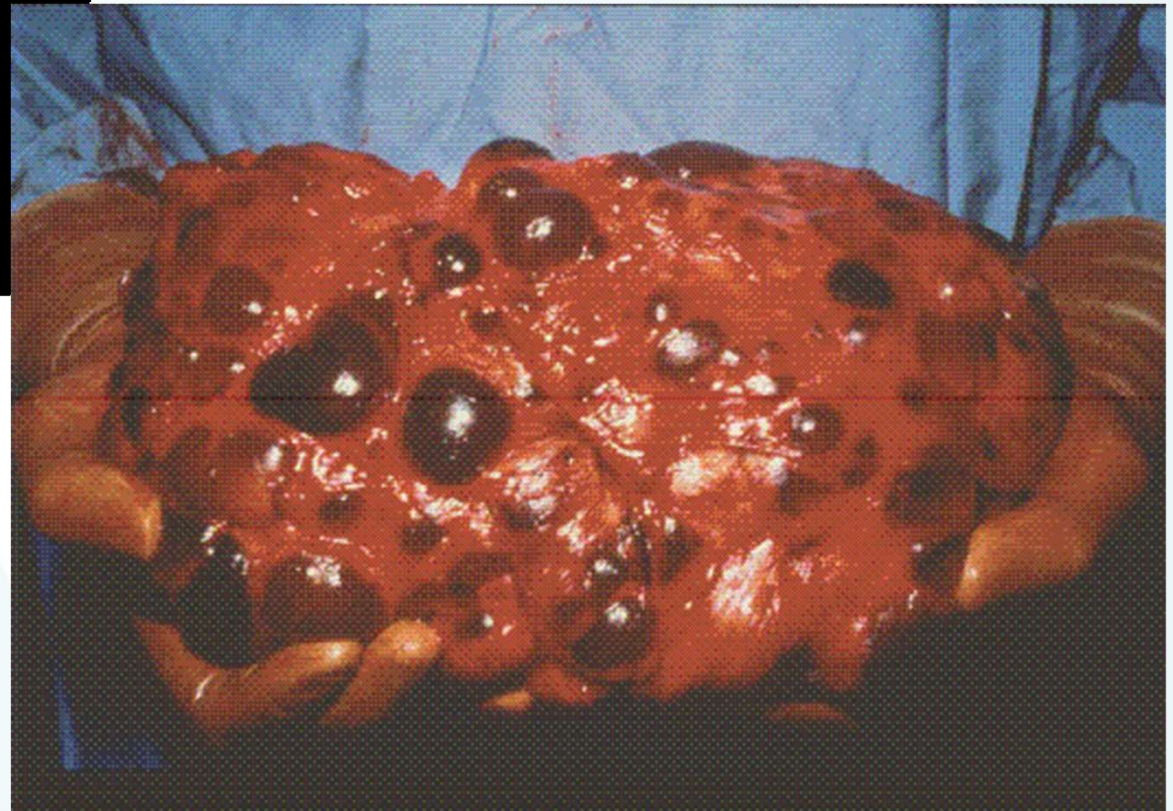
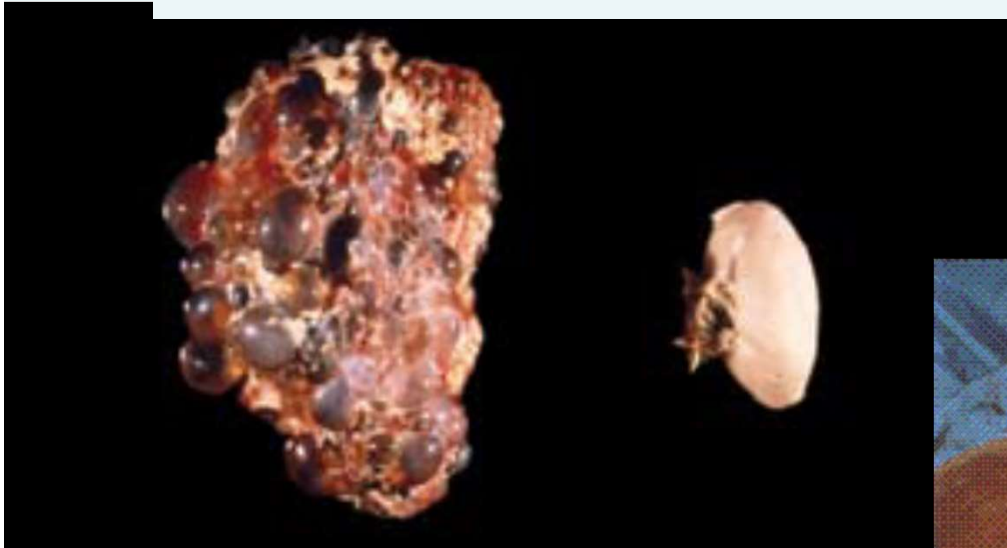
- The **#1** cause of kidney failure from a genetic disease
- The **#4** leading cause of kidney failure overall
- A '**systemic disease**' causing:
 - Heart failure/stroke
 - Life-threatening brain aneurysms
 - Cystic kidneys, livers and pancreas
 - Chronic hypertension, kidney infections & kidney stones
 - Persistent pain, anxiety and depression
 - Premature death

This ist PKD!



- There is no know treatment or cure for PKD.
- Until one is found, PKD will threaten the lives of every generation of every family living with the disease.
- PKD affects men, women and children
 - regardless of age, race, geography or ethnic origin.
- It does not skip a generation.

A picture says more than thousand of words



Development of the disease:

- More than 60 percent of people having PKD will develop ESRD (**E**nd **S**tage **R**enal **D**isease).
- Only dialysis or transplantation can help.

■ Five stages of PKD

analogous to KDIGO (Kidney Disease: Improving Outcome)



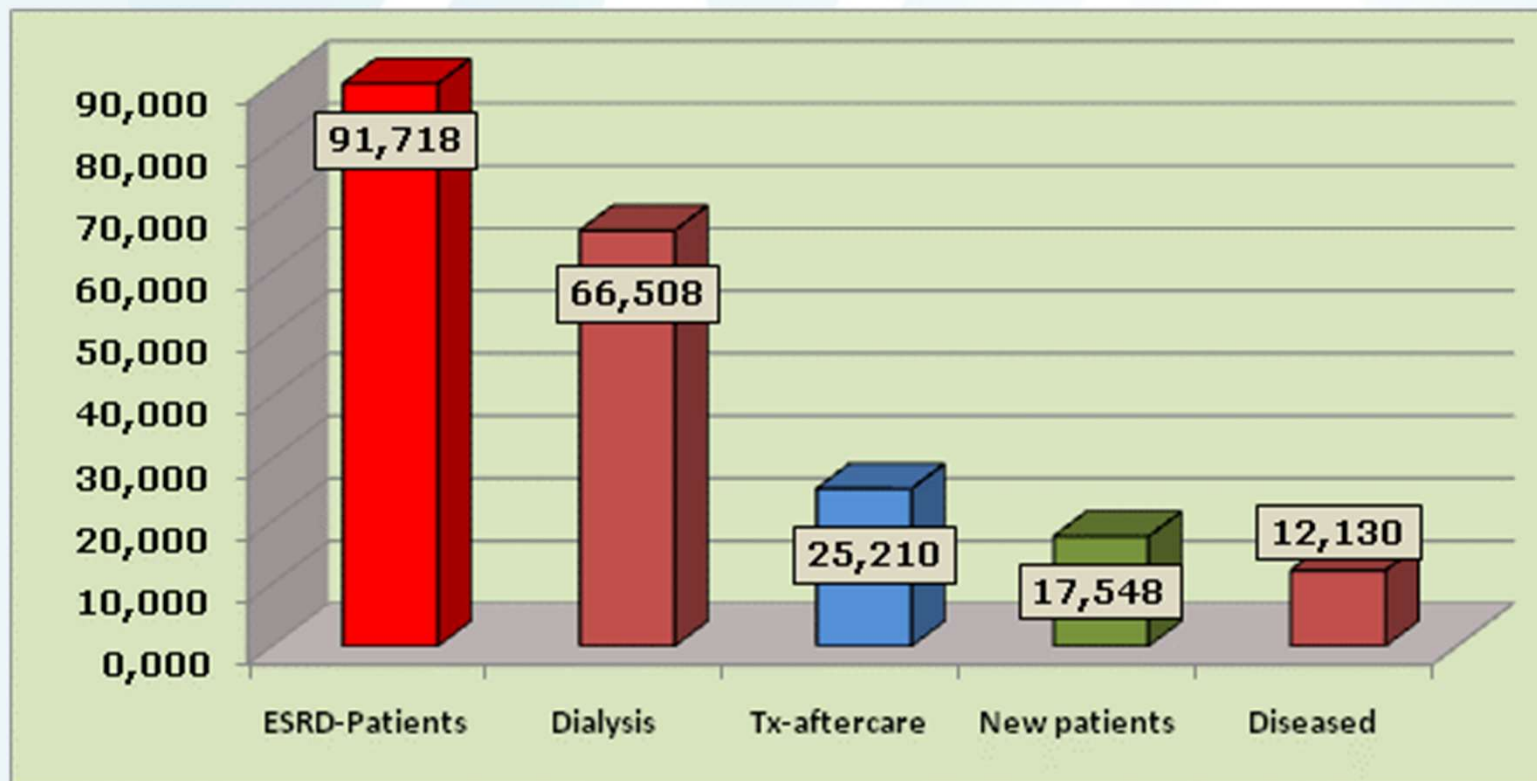
Stage	Description	GFR (ml/min)/ 1.73m ²	Measures	Prevalence (%)
1	Kidney disease with normal GFR	>90	diagnosis, no special therapy	3,3
2	Mild PKD	89 - 60	medication and lifestyle changes may help slow its progress	3.0
3	Moderate PKD	59 - 30	Diagnosis and therapy of secondary complications, such as high blood pressure, anemia, bone and cardiovascular disease.	4.3
4	Severe PKD	29 - 15	preparation for dialysis or a kidney transplant, Kidney size greater than 1500 ml	0,2
5	End stage renal disease (ESRD)	<15	loss of nearly entire kidney function, and eventually dialysis or a kidney transplant is needed.	0,1

Source: Am J Kidney Dis. 2003 Jan;41(1):1-12.

This is PKD ! (in Germany)

91,718 people with **End Stage Renal Disease (ESRD)** - (Dialysis /Transplantation)

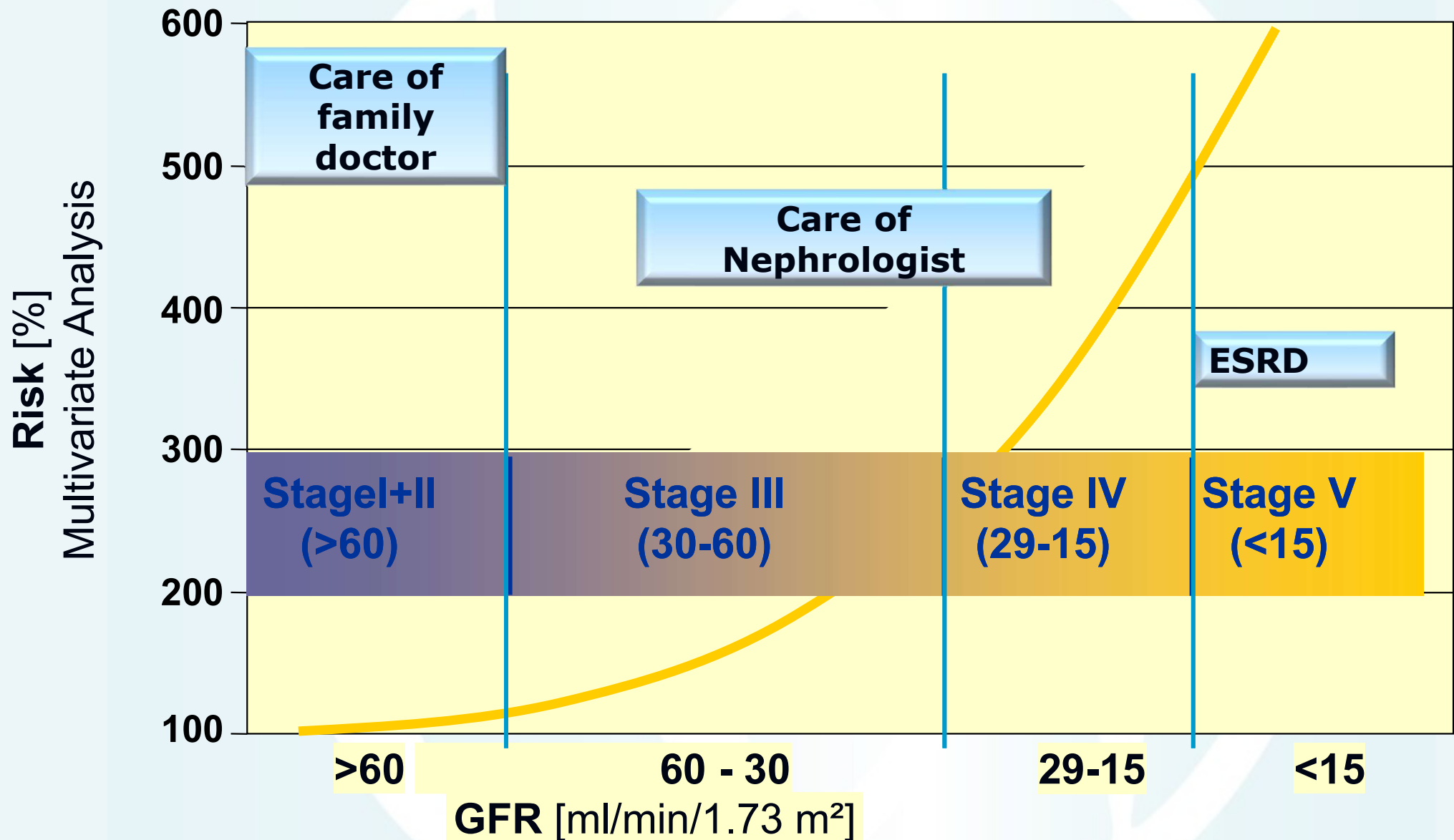
8,000 waiting for an kidney



7 % PKD

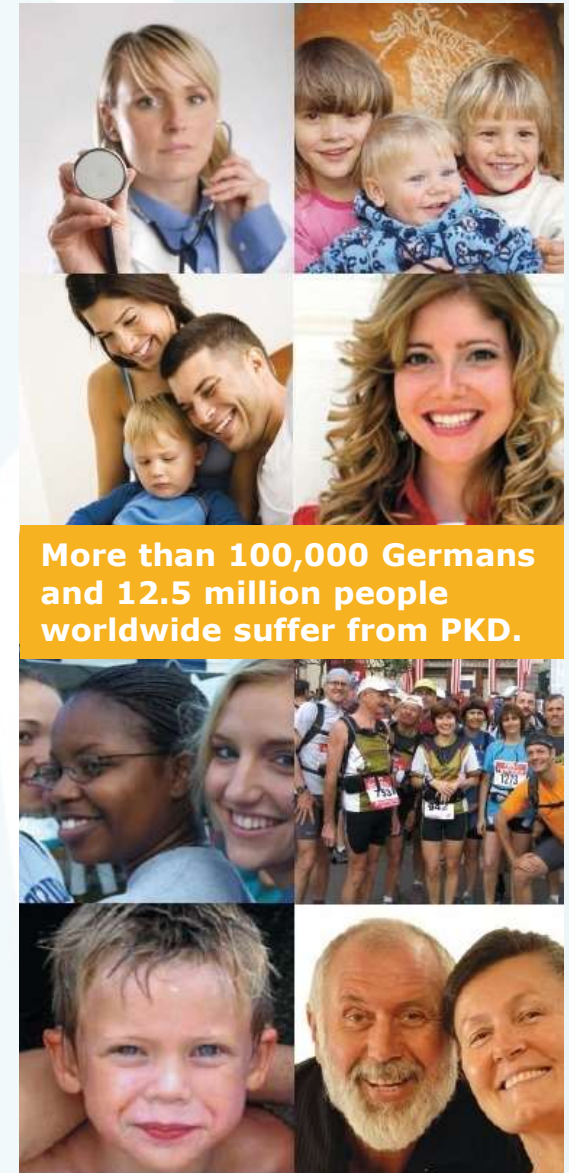
Mortality rate on
dialysis:
17%/ year

Source: Quasi-Niere (12/31/2006)



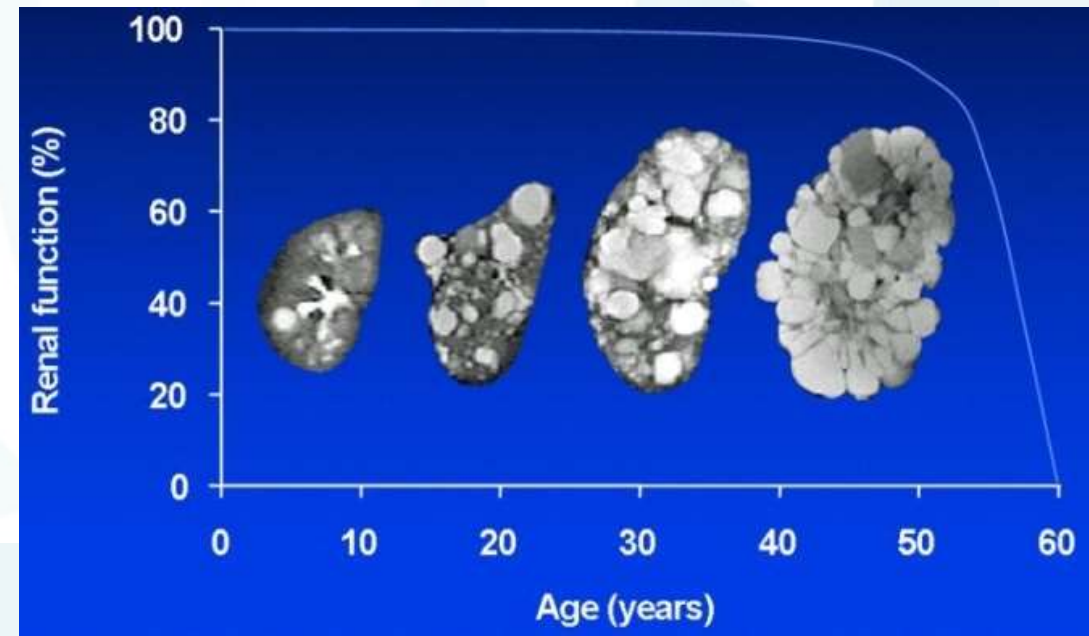
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- ➡ ■ What is it like to live with PKD?
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- At first, PKD develops very gradually.
- In the early stages Creatinine increases only minimally, but GFR shows increasing function loss.
- In the beginning there were no negative effects on my life.
- The blood pressure increase over the years made a step by step adjustment of medication **continually** necessary.

- Increasing abdominal girth followed the growth of the cysts.
- Occasionally blood in the urine and cysts infection → very painful
- With a kidney size of 1500 ml volume there is a progressive loss of kidney function (GFR)
- 5ml/min/year !
(see CRISP study)



- Problems keeping psychic balance
- Negative effects on every day work
- Necessity of increased regular medications (Compliance)
- Power sports and some team sports are not recommended
- Something to learn to accept:
Good health is not a matter of course.



Coping with the disease !

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Anxiety and concerns of parents

- Make parents guilty because of my PKD.
- Family planning: children or no children?
- What will the life of my children be like?
- Reduced life expectancy?
- Will there be a treatment or cure in the future?



Concerns of parents

Examples of children with PKD:



Shortly after birth, Eva Maria Hoffmann was diagnosed with autosomal dominant PKD. Her family came to Children's Hospital to see Ellis Avner, MD, for treatment of this disease, which will impact Hoffmann for her entire life.

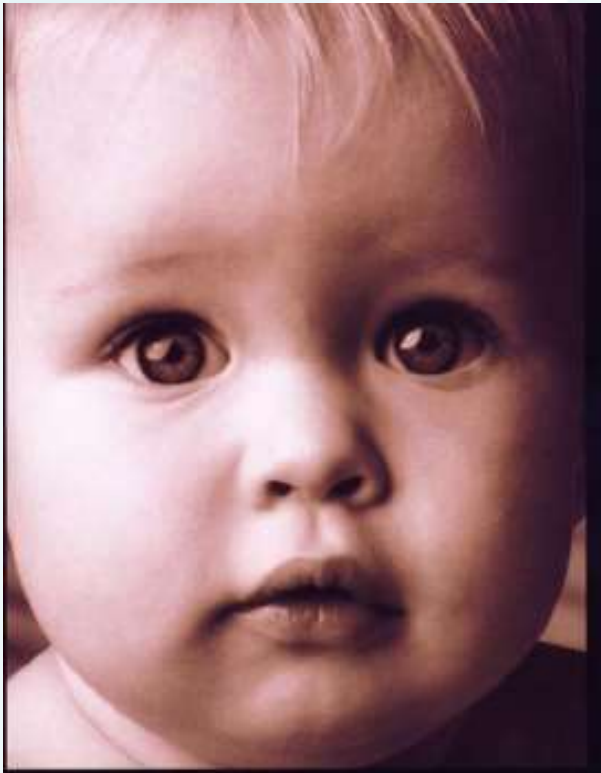


While her family knows that there is no cure for PKD, they know that Dr. Avner and his team have invested in the research to discover treatments for Hoffmann and others who suffer from PKD.



Urgent need for a PKD cure!

This is PKD!



**Apart from PKD
treatment and cure,
the future looks
perilously daunting
for millions of children
who inherit PKD.**



Urgent need for a PKD cure!

Our Vision

No one should have to suffer the full effects of PKD.



Our Mission

Promote research to find a cure for PKD and improve the care and treatment of those afflicted by it.

This is PKD!

**„The most valuable of all
capital is that invested in
human beings.“**

Alfred Marschall, " Principles of Economics" (1892)

- Contact:
 - Uwe Korst
 - PKD Familiäre Zystennieren e.V.
 - + 49 6251 983 683
 - www.pkdcure.de

- Questions?

- **Selbsthilfe** findet statt durch Informationsaustausch und ein Forum für Patienten.
- **Prävention** fördern wir durch Aufklärung bei Patienten und Ärzten und die Förderung eines öffentlichen Bewusstseins.
- **Forschung** unterstützen wir und informieren über den aktuellen Stand durch Zusammenarbeit mit unserem wissenschaftlichen Beirat.