Autosomal dominant polycystic kidney disease (ADPKD)

- ADPKD is a progressive and chronic genetic disease, primarily characterised by the development and enlargement of multiple fluid-filled cysts in the kidneys[1,2]

- ADPKD is the most common inherited kidney disease[3]
    - ADPKD is caused by faulty genes that can affect both sexes of all age, racial and ethnic groups[3]
    - ADPKD is thought to affect between 3-4 people per 10,000 – as many as 205,000 people in Europe[4,5]
    - ADPKD is a disease of genetic variability and therefore the disease course and progression can vary for each individual
    - In certain cases, patients in early stage ADPKD often do not experience any symptoms and as a result are diagnosed late (30-55 years).[6-8] By the time a decline in kidney function is identified, the destruction in kidney anatomy is already quite advanced[9,10]

The physical burden of ADPKD

- The formation and growth of kidney cysts leads to an increase in kidney size, or totally kidney volume (TKV), which is a continuous and quantifiable indication of disease progression[1,10]
    - ADPKD patients experience a variety of complications including hypertension, chronic and acute pain, repeated urinary tract infections (UTIs) and blood in the urine (haematuria)[1]
    - Approximately half of patients reach end-stage renal disease (ESRD) and require renal replacement therapy (RRT) in the form of dialysis or a kidney transplant by age 54[11,12]
    - ADPKD is the fourth leading cause of ESRD[13] and accounts for around 10% of patients with ESRD requiring RRT[6]

The emotional burden

- In addition to the physical symptoms of the disease, ADPKD causes a profound psychological and emotional burden for those living with the disease, their families and loved ones[14-16]
    - Patients with ADPKD report significantly impaired health-related quality of life issues due to pain, anxiety, depression and consequent difficulty with activities of daily living[14-16]
    - ADPKD patients are more likely to experience anxiety and depression compared to the general population[17,18]
    - The findings of the largest ever survey of people with ADPKD in Europe highlighted the negative impact the disease has on families (77% of respondents), relationships (41%), sexual relationships (42%), social lives (33%) and the decision to have children (35%)[19]

The financial burden for society:

- ADPKD exacts a high financial and societal cost on healthcare systems.[20,21] This is due to the increased risk of adverse outcomes such as cardiovascular disease events, hospitalisations, resource utilisation and mortality[20]
- Annual direct healthcare costs for ADPKD are estimated at €2 billion in the EU alone[22]

Autosomal dominant polycystic kidney disease (ADPKD)

References

1. Torres VE, Harris PC et al. Tolvaptan in Patients with Autosomal Dominant Polycystic Kidney Disease. *The New England Journal of Medicine*. 2012;367 (25): 2407-2418
2. Patel V, Chowdhury R et al. Advances in the pathogenesis and treatment of polycystic kidney disease. *Current Opinion in Nephrology and Hypertension*. 2009;18:99-106
3. Torres VE, Harris PC. Autosomal dominant polycystic kidney disease: the last 3 years. *Kidney International*. 2009;76:149–168
4. Neumann H, Jilg C et al. Epidemiology of autosomal-dominant polycystic kidney disease: an in-depth clinical study for south-western Germany. Nephrology Dialysis Transplantation. 2013;28:1472-1487
5. Patch C, Charlton J et al. Use of antihypertensive medications and mortality of patients with autosomal dominant polycystic kidney disease: a population-based study. American Journal of Kidney Disease. 2011;57(6):856-862
6. Thong KM, Ong ACM. The natural history of autosomal dominant polycystic kidney disease: 30-year experience from a single centre. *QJ Med.* 2013;2-8
7. Ozkok A et al. *Clin Exp Nephrol.* 2013 Jun;17(3):345-51
8. Cornec-Le Gall et al. ERA-EDTA poster 2013 "Clinical factors predicting renal outcomes in ADPKD: results of the GENKYST registry"
9. Grantham JJ. Autosomal Dominant Polycystic Kidney Disease. *The New England Journal of Medicine*. 2008;359:1477-85
10. Grantham JJ, Chapman AB et al. Volume progression in Autosomal Dominant Polycystic Kidney Disease: The Major Factor Determining Clinical Outcomes. *Clinical Journal of the American Society of Nephrology*. 2006;1:148-157
11. Takiar V & Caplan MJ. Polycystic kidney disease: pathogenesis and potential therapies. *Biochimica et Biophysica Acta*. 2011;1812(10):1337-43
12. Alam A, Perrone RD. Management of ESRD in Patients With Autosomal Dominant Polycystic Kidney Disease. Advances in Chronic Kidney Disease, Vol 17, No 2. March 2010: pp 164-172
13. Masoumi A, Reed-Gitomer B et al. Developments in the Management of Autosomal Dominant Polycystic Kidney Disease. *Therapeutics and Clinical Risk Management.* 2008;4(2):393–407
14. Christophe JL, van Ypersele de Strihou C et al. Complications of autosomal dominant polycystic kidney disease in 50 haemodialysed patients. A case-control study. *Nephrology Dialysis Transplantation*. 1996;11(7):1271-1276
15. Rizk D, Jurkovitz C et al. Quality of life in Autosomal Dominant Polycystic Kidney Disease patients not yet on dialysis. *Clinical Journal of the American Society of Nephrology*. 2009;4:560-6
16. Perlman RL, Finkelstein FO et al. Quality of life in chronic kidney disease (CKD): a cross-sectional analysis in the Renal Research Institute-CKD study. *American Journal of Kidney Diseases*. 2005;45:658-66
17. de Barros BP, Nishiura JL et al. Anxiety, depression, and quality of life in patients with familial glomerulonephritis or autosomal dominant polycystic kidney disease. *Jornal Brasileiro de Nefrologia*. 2011;33(2):120-128
18. Pérez-Dominguez T, et al. Progression of chronic kidney disease. Prevalence of anxiety and depression in autosomal dominant polycystic kidney disease. *Nefrologia* 2012;32:397v9
19. Otsuka Pharmaceutical Europe Ltd. ADPKD Patient Semi-Quantitative Survey: Overall report for Europe and the Nordics. 2013. Data on file
20. Lentine KL et al. Renal function and healthcare costs in patients with polycystic kidney disease. *Clinical Journal of the American Society of* Nephrology. 2010 Aug;5(8):1471-9
21. Knight T, Schaefer C, Krasa H et al. Poster "Economic Burden of Autosomal Dominant Polycystic Kidney Disease in the United States" ISPOR May 2013; ERA-EDTA May 2013
22. KDIGO. KDIGO controversies conference on ADPKD [online] 2014. Available from: http://www.kdigo.org/ControConf/ADPKD/Presentations/KDIGO%20ADPKD%20Conference%20Overview%20&%20Objectives.pdf [Last accessed: April 2015]