AKU-Symptoms:

- Chronic fatigue symptoms
- Sleep apnea
- Joint pain
- Skin problems
- Loss of vision
- Loss of hearing
- Kidney impairment
- Digestive problems
- Neuropathy
- Gum bleeding
- Cardiac problems
- Eyesight decline
- Eye problems
- Hair loss
- Dental problems
- Bone problems
- Deep vein thrombosis
- Easy bruising
- Many other symptoms

Abstract:

Alkaptonuria (AKU, ochronosis) is an inborn metabolic disease, resulting in the accumulation of the metabolic intermediate homogentisic acid (HGA). Oxidation of HGA by air or within connective tissue causes darkening of the urine, pigmentation of eyes and ears, kidney- and prostate-stones, aortic stenosis, but most severely an early onset of arthritis called ochronotic arthropathy (ochronosis) due to deposition in the cartilage. Ochronosis is very painful, disabling and progresses rapidly. Starting in the thirties with the spine and affecting large joints in the forties, patients frequently require joint replacements in their fifties and sixties1. Like many of the rare diseases, AKU patients undergo a long odyssey of several years until their diagnosis.

The German AKU Society "Deutsche Aknapathen Gesellschaft für Akpakunurie (DSAKU) e.V." was founded in 2012 and became subsequently registered as a non-profit patient organization. First of all, the DSAKU identified AKU-patients, set up a homepage and designed flyers with information for patients, their families, medical professionals and healthcare services. Second, it offered workshops on AKU-related topics and enabled personal exchange. Third, it raised awareness of AKU, both nationally and internationally, by information booths, presentations and posters at scientific congresses as well as rare disease days (RDD). Fourth, in response to the needs of patients, it established collaborations and built up national networks for a better healthcare accordingly. Thus, patients were encouraged to visit the centres for metabolic diseases at the Charité (Berlin), Hannover Medical School (HHU), University of Düsseldorf and Institute of Human Genetics at the University of Würzburg to bundle knowledge and expertise. The DSAKU is member of ACHSE e.V., NAKOS, EURORDIS and MetaFELN and registered in the databases SC-Athlete, ZIEF and Orphanet. Finally, the DSAKU is nationally and internationally active in health politics regarding training in drug safety and evidence-based medicine.