What is Alkaptonuria (AKU)?
- An autosomal recessive disorder with a prevalence of 1 in 500,000
- Characterised by many features but dominated by premature disabling arthritis (Fig 1)
- Due to accumulation of homogentisic acid (HGA)
- A new drug, Nitisinone, decreases production of HGA (Fig 2)
- Optimal use of Nitisinone requires effective identification of people with AKU

Problem of AKU in the UK
- AKU is one of the orphan diseases due to its rarity
- There is no UK database of all diagnosed AKU patients
- There is no National register for disease (rare or common)

Resources available to Identify people with AKU
AKU Society of the UK (established in 2003)
AKU Website (established 2003) (Fig 3)
National Lottery Grant Award (awarded 2007)
Manned AKU Information Centre in Royal Liverpool University Hospital (since 2007) (staffed by 2 people)

Methodology used to Identify people with AKU
Active Manned Interactive Website
General Practitioner Postal Questionnaire Survey in UK (Fig 4)
Targeted Sibling Screening
Targeting Medical Professionals in Conferences

Results (Table 1)
GP’s survey: Over 9668 GP’s were mailed (15% response rate to questionnaires) (Fig 5)
Total number of patients in UK with AKU (78)
(Fig 6. UK Map of AKU Patients)
Total Number of patients with AKU worldwide (512)
(Fig 7. World Map of AKU patients)
Numbers of Patients with AKU identified by different approaches
Website (including patient networks) 44
GP Survey 23
Targeted family screening 11
Medical Conference Targeting (GPs, Rheumatology, Biochemists, Orthopaedic) 429

Conclusion
In the absence of a national disease register, various strategies may be needed to identify individuals with a rare disease, some clearly more cost-effective than others

Continuing Barriers
The full identification of an individual with AKU requires two things:
1. Knowledge of Location of Patient
2. Contact with Patient
Due to patient confidentiality concerns, contact was often incomplete
A limitation of the study is that people with access to electronic resources like the internet are more likely to be identified

Recommendations
Our experience suggests that a dedicated manned website may be the most cost-effective way to approach the issue of identifying people with a rare disease and supporting them