AKU Profile (serum)

Description	Profile used to monitor AKU patients on nitisinone
Indication	Investigation and monitoring of alkaptonuria
Additional Info	Alkaptonuria is an autosomal recessive inborn error of metabolism caused by deficient activity of hepatic homogentisate-1,2-dioxygenase, which metabolises homogentisic acid (HGA) to maleylacetoacetic acid. The resulting block in phenylalanine metabolism causes accumulation of HGA, 4-hydroxyphenylpyruvic acid, tyrosine and other intermediates. The AKU profile encompasses tyrosine, phenylalanine, HGA, hydroxyphenyllactate, hydroxyphenylpyruvate and nitisinone.
Concurrent Tests	None
Dietary Requirements	The patient need not be fasting, although this preferable and no special preparations are necessary.
Interpretation	The metabolic consequence of alkaptonuria is elevated HGA in plasma (typically 40 umol/L; normally undetectable in health) and urine (300-fold to millimolar excretion; again undetectable in non-AKU).
Collection Conditions	Serum samples need to be deproteinised with perchloric acid. Please contact laboratory for protocol and information. If procedure is not possible, then serum should be separated and frozen immediately. Samples should be transported frozen.
Frequency of testing	As required