Urine Homogentisic acid (HGA)

Description	An intermediary metabolite in tyrosine degradation
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. Diagnosis requires demonstration of increased plasma or urinary HGA and decreased homogentisate-1,2-dioxygenase activity in liver or kidney cells. Genetic testing can also be undertaken, although it is usually a research procedure.
Concurrent Tests	None
Dietary Requirements	The patient need not be fasting 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	Urine samples should be preserved by acidification to < pH 2.5 by addition of concentrated sulphuric acid. A 24 hour sample is preferred. All samples must be stored and shipped frozen.
Frequency of testing	As required