Homogentisic acid

General:

A defect of homogentisic acid dioxygenase leads to a block in the tyrosine metabolism with accumulation of homogentisic acid (earlier alkapton), which is excreted into urine (alkaptonuria), but is also deposited in the articular cartilage. It leads to alkaptonuria/ochronosis. The disorder is autosomal-recessively inherited with a frequency of 1:250,000. Clinical indications are black coloration of urine with alkaline pH. The treatment consists in restricted protein diet.

Indication:	Alkaptonuria/ochronosis
Material:	10 ml urine
Preanalytics:	spontaneous urine
TAT:	2 weeks*
Method:	LCMS
Units:	umol/L
Ref.- range:	<200

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit http://www.fml-dubai.com/parameter-listings/

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