

## Environmental Toxin-induced Porphyrinurias

Analytes	Heavy Metals				Organotoxins							
	Aluminum	Arsenic	Mercury	Lead	Hexachlorobenzene	Methyl chloride	Dioxin	Polymethylchloride	Polybrominated biphenyl	Alcohol (chronic hepatic porphyria - early)	Alcohol (chronic hepatic porphyria - late)	
Porphyrinurias	+	+			+		+		+		+++	
Heptacarboxyporphyrin		(+)			(+)		(+)		(+)		+	
Hexacarboxyporphyrin		(+)			(+)		(+)		(+)		+	
Pentacarboxyporphyrin		(+)	+									
Precoproporphyrin			++									
Coproporphyrin I		+		+		+		+	+	+		
Coproporphyrin III	+		+	+		+		+	+	++		
Precopro/Uro I & III†			+									
Copro I/Copro III‡		+										

**Table 1A: Effects of toxins on urinary porphyrin levels**

The green pluses (+) show the typical pattern of elevated porphyrins. The double pluses (++) show dominant elevations. Pluses enclosed in parentheses indicate alternate patterns that may appear. Calculated total porphyrins levels are generally elevated in the severe types of any condition that produce porphyrias. Aminolevulinic acid (ALA) and zinc protoporphyrin are other porphyrin pathway intermediates elevated in lead toxicity and iron deficiency, respectively.

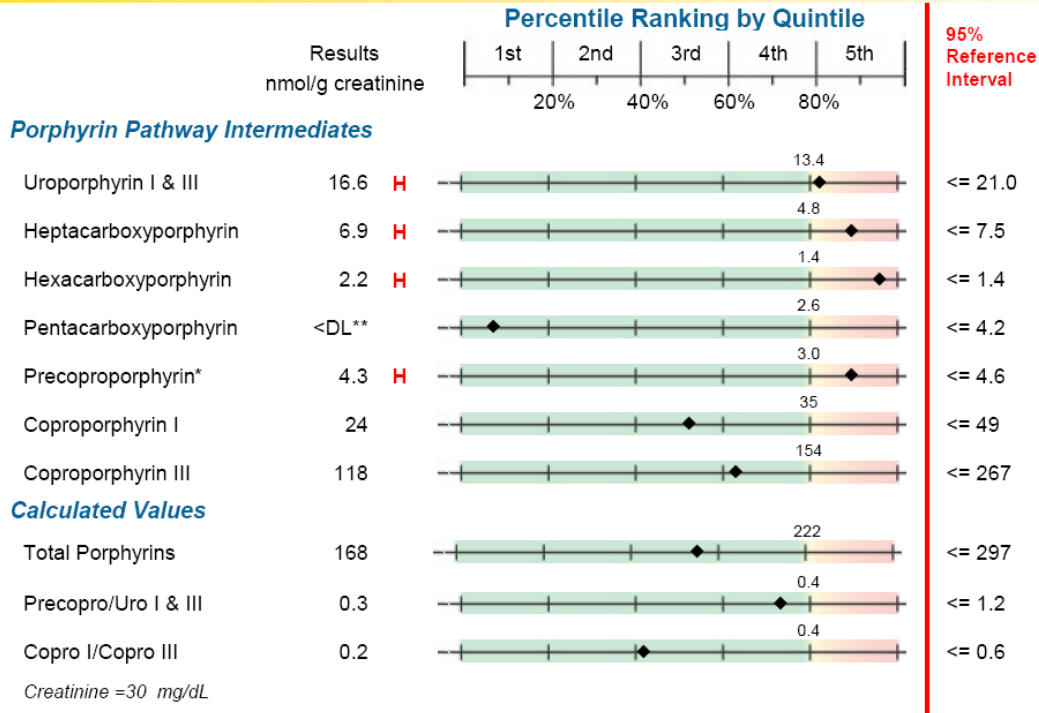
\* Hepatic degeneration due to porphyrin accumulation in the liver is found in these conditions.

Overview: Toxicants can interfere with enzymes in the heme synthesis pathway. When toxicants interrupt healthy enzyme function, the intermediates, called porphyrins, spill into the urine. In the cases below, the Porphyrins profile shows that the child may have toxicant interference in heme synthesis. If clinical history supports toxic exposure, a detoxification protocol may be effective in the child's recovery. To identify the specific toxin at fault, further testing is recommended, such as toxic elements in hair, urine, or blood and/or organotoxin testing.

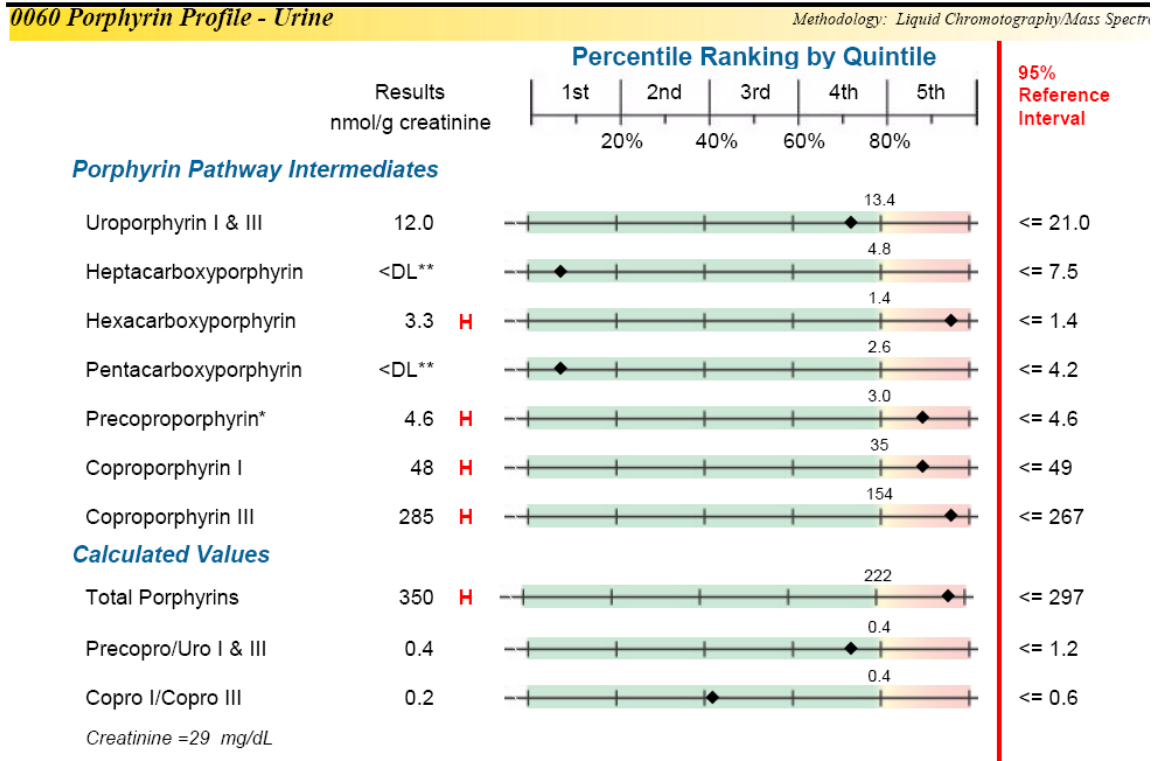
Case AG. This 4-year-old male shows up-regulation of heme synthesis, due to the elevations in Uro I & III, Hepta, and Hexa. This can sometimes be caused by iron deficiency anemia or other demands for increased heme synthesis. Hepta and Hexa may be elevated due to arsenic exposure or organotoxin exposure. Precoproporphyrin is high, however the other porphyrins that confirm mercury toxicity are not elevated (Penta and Copro III).

**0060 Porphyrin Profile - Urine**

Methodology: Liquid Chromatography/Mass Spectroscopy



Case OM. A 2-year-old female with ASD has porphyrins spilling in urine which indicates toxicant interference in heme synthesis. This patient shows 2/3 porphyrins elevated from the mercury toxicity pattern (elevated pentacarboxyporphyrin, precoproporphyrin, and coproporphyrin III). Hexacarboxyporphyrin can elevate with arsenic, hexachlorobenzene, dioxin, polybrominated biphenyl, and alcohol (see Table 1A). Coproporphyrin I can elevate due to arsenic, lead or other organotoxins.



\* Atypical porphyrin consistent with precoproporphyrin reported in the literature.<sup>1,2</sup> Precoproporphyrin is reported as a ratio of peak area to internal standard peak area in units of response/g creatinine.

1. J.S. Woods, M.A. Bowers, H.A. Davis, Toxicology and Applied Pharmacology 110, 464-476 (1991).
2. D. Echeverria et.al., Neurotoxicology and Teratology 28 (2006) 39-48.

\*\*Result Less than detection limits

\*\*\*Unable to calculate due to indeterminate value