Laboratory Monitoring of Tyrosinemia Type I with Nitisinone

Type of Evaluation	What it Measures	At Diagnosis	With Appropriate Treatment
		These results reflect average results and may vary from child to child	
CBC:			
(1) Hgb, Hct, WBC, plts	Complete Blood Count (CBC)	Possibly Low	Normal range
Tyrosinemia Markers (blood):			
(2) alpha-fetoprotein	Liver Function	Very high	Normal range
(3) succinylacetone	Diagnosis of tyosinemia	Present	Absent
(4) (delta-ALA dehydratase)*	Enzyme	Low	Normal range
Blood Chemistries:			
(5) amino acids (methionine, phenylalanine, tyrosine)	Metabolism of protein	Methionine: high Phenylalanine: high Tyrosine: high or normal	Methionine: normal range Phenylalanine: normal range Tyrosine: high (but <500 mcmol/L)
(6) prothrombin time (PT)	Blood clotting	Elevated	Normal range
(7) partial thromboplastin time (PTT)	Blood clotting	Elevated	Normal range
(8) bilirubin	Jaundice	Possibly elevated	Normal range
(9) ALT/AST	Liver Function	Possibly elevated	Normal range
(10) GGT	Liver Function	Possibly elevated	Normal range
(11) alkaline phosphatase	Bone formation	Possibly elevated	Normal range
(12) BUN, creatinine	Kidney function	Possibly elevated	Normal range
Urine Samples:			
(13) succinylacetone	Diagnosis of tyosinemia	Present	Absent
(14) (delta-aminoleuvulinic acid)*	A metabolite elevated in tyrosinemia	Elevated	Normal range
Renal Studies:			
(15) renal ultrasound (nephromegaly)	Non-invasive view of kidneys	Possible dilation of kidneys	Normal size
Skeletal Evaluation:			
(16) x-ray of wrist (rickets)	Non-invasive view of bones	Possible rickets	May resolve
CT Evaluation:			
(17) abdominal CT (hepatocarcinoma)	Non-invasive view of liver	Possible 'nodules' seen	May resolve, or remain unchanged
Developmental Evaluation:			
(18) Differential Ability Scale (DAS), WAIS-R, Stanford Binet, etc.	Intellectual development	N/A	Appropriate
(19) Growth	Height, weight	Possibly delayed	Appropriate
Nutritional Evaluation			
(20) Nutrient Intake	Overall nutritional status	N/A	N/A
Other (optional)			
(21) Mutation Analysis	The genetic changes that caused tyrosinemia	N/A	N/A

*** Those children who were diagnosed after Orfadin became commercially available do not require these laboratory measurements. They are part of a research protocol only.**