Title: Tyrosinemia Type I *GeneReview* — Table 4
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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 4. Guidelines for Monitoring Children with Tyrosinemia Type I Diagnosed by Clinical Presentation

Evaluation		Initiation of Therapy (Baseline)	First 6 Mos:		After 1 Yr of Rx: every 6-12 mos	As Clinically Indicated
			Monthly	Every 3 mos		
Tyrosinemia type I markers	Plasma concentration of methionine, phenylalanine, tyrosine	X	X		X	X
	Blood / urine succinylacetone	X	X (urine)		X	X
	Blood nitisinone concentration		X		X	or X
CBC	Hemoglobin, hematocrit, WBC, platelet count	X	X		X	or X
Liver evaluation	Serum AFP concentration	X	X		X	or X
	PT/PTT	X	X (until nml)			X
	Bilirubin	X				X
	ALT/AST/GGT	X		X (until nml)	X	or X
	Alkaline phosphatase	X		X (until nml)	X	
	CT or MRI [1]	X			X	or X
Renal studies	BUN / creatinine	X	X			X
	Urine: PO_4, Ca, Prot/Cr ratio	X				X
Skeletal evaluation	X-ray of wrist (for rickets)	X				X

AFP = alpha-fetoprotein

ALT/AST = alanine transaminase/ aspartate transaminase

BUN = blood urea nitrogen

CBC = complete blood count

GGT = gamma-glutamyl transferase

PT/PTT = prothrombin time/partial thromboplastin time

1. MRI with contrast to evaluate for liver adenomas or nodules and for kidney size