Tyrosine Screening Fact Sheet for Health Care Providers
Newborn Screening Program of the Oklahoma State Department of Health

What is the differential Diagnosis?
Tyrosinemia I (hepatorenal); Tyrosinemia II (oculocutaneous); Tyrosinemia III; Transient Hypertyrosinemia; Liver disease.

What are the characteristics of tyrosine disorders?
- Autosomal recessive genetic conditions.
- Most infants are born to parents who are both unknowingly asymptomatic carriers and have NO known history of a tyrosine disorder in their family.
- There are three types of Tyrosinemias, each has distinctive symptoms and is caused by a deficiency of different enzymes. Tyrosinemia Type I is the most severe form of this disorder.
- The worldwide incidence of Tyrosinemia Type 1 is estimated to be about 1:100,000 live births. Tyrosinemia Type II is about 1:250,000. Tyrosinemia type III is very rare, the incidence is unknown.
- Newborns with these disorders often appear normal. Symptoms usually appear in the first few months of life and include diarrhea, vomiting, jaundice, and increased tendency to bleed (particularly nosebleeds). Lifelong treatment includes a special diet, and close monitoring by a metabolic specialist. The medication, Nitisinone (Orfadin), may also be indicated in the treatment of Tyrosinemia Type I.

What is the screening methodology for tyrosine?
1. An amino acid profile by Tandem Mass Spectrometry (MS/MS) is performed on each filter paper. Tyrosine **may be** detected through the newborn screen. There is a lower probability of detecting this disorder during the newborn period.
2. Tyrosine is the primary analyte.

What is an in-range (normal) screen result for tyrosine?
Tyrosine < 400 µmol/L is NOT consistent with a tyrosine disorder.
See Table 1.

TABLE 1.
In-range Tyrosine Newborn Screening Results

Primary Analyte		In-Range (µmol/L)
Tyrosine	<	400

What is an out-of-range (abnormal) screen for tyrosine?
Tyrosine ≥ 400 µmol/L requires further testing.

What screen results will require a repeat filter paper?
Tyrosine ≥ 400 µmol/L in an infant less than 14 days old at time of screening requires a repeat filter. Consultation with a metabolic specialist will be left to the provider's discretion.

What screen results will require diagnostic testing?
Tyrosine ≥ 400 µmol/L in an infant at least 14 days old at time of screening requires **immediate** action and confirmatory testing. The follow-up program will provide detailed guidance on required actions.

What are the follow-up needs?
The follow-up program will provide detailed guidance on needed actions.

What is my role in screening?
If you are listed as the infant's planned health care provider on the filter paper requisition, you are required by the *Newborn Screening Program Regulations* to initiate follow-up activities.

Resources
- **ACMG Newborn Screening ACT Sheets:** https://www.ncbi.nlm.nih.gov/books/NBK55827/
- **Integris Pediatric Specialty Clinic, Inborn Error of Metabolism (IEM) Clinic**
  Geneticist pager: (405) 630-3794
- **OU Children's Physicians – Genetics Clinic**
  Page Operator: (405) 271-3636
- **Newborn Screening Follow-Up Program**
  (405) 271-6617 option 2 or (800) 766-2223; www.nsp.health.ok.gov