

Wilson Disease

Patient Clinical Journey



DELAYED DIAGNOSIS

PRE-DIAGNOSIS

- Typical Accompanying Diagnoses**
 - Anemia
 - Anxiety
 - Ascites
 - Behavioral issues
 - Cirrhosis
 - Dizziness
 - Dysphagia
 - Edema
 - Elevated serum aminotransferase activity (AST, ALT)
 - Esophageal varices
 - Fatty liver
 - GERD
 - Hepatitis
 - Infertility
 - Jaundice
 - Kayser-Fleischer rings
 - Kidney stones
 - Liver failure
 - Migraines
 - Muscle weakness
 - Neuropsychology-related disorders
 - Portal hypertension
 - Vomiting
 - Misdiagnoses – *including but not limited to Parkinson's, Huntington's disease*
- Testing & Procedures**
 - Ceruloplasmin level
 - Liver transplant
 - MRI
 - Transjugular intrahepatic portosystemic shunt (TIPS)
 - Urinary copper level
- Specialists**
 - Behavioral therapist
 - Gastroenterologist
 - Hepatologist
 - Neurologist
 - Ophthalmologist
 - Pathologist
 - Pediatrician/Primary care provider
 - Psychiatrist
 - Radiologist
- Events**
 - Emergency department visits
 - Hospitalizations
- Without appropriate treatment, Wilson Disease may be fatal.*

TIME OF DIAGNOSIS

- Typical Accompanying Diagnoses**
 - All previously listed diagnoses may remain as accompanying diagnoses as treatment begins.*
- Testing & Procedures**
 - Echocardiogram
 - Electrocardiogram (ECG/EKG)
 - Gene sequencing of ATP7B gene
 - Liver biopsy
 - Liver function tests
 - Liver transplant
 - Metabolic panel
 - Radiography
 - Slit-lamp eye exam
 - Transjugular intrahepatic portosystemic shunt (TIPS)
 - 24-hour urine collection
- Treatments & Supportive Therapies**
 - Copper chelating – *penicillamine, trientine dihydrochloride*
 - Zinc acetate
 - Various medications – *specific to needs of each patient*
- Specialists**
 - Gastroenterologist
 - Genetic counselor
 - Geneticist
 - Hepatologist
 - Neurologist
 - Nutritionist
 - Ophthalmologist
 - Pathologist
 - Psychiatrist
 - Radiologist
- Events**
 - Emergency department visits
 - Hospitalizations

POST DIAGNOSIS

- Testing & Procedures**
 - Liver transplant
 - Long-term monitoring of liver function
 - Transjugular intrahepatic portosystemic shunt (TIPS)
 - Slit-lamp eye exam
- Treatments & Supportive Therapies**
 - Copper chelating – *penicillamine, trientine dihydrochloride*
 - Zinc acetate
 - Nutrition adjustments
 - Physical Therapy (PT)/Occupational Therapy (OT)/Speech Pathology (SP)
 - Various medications – *specific to needs of each patient*
- Specialists**
 - Counseling Support Services
 - Gastroenterologist
 - Genetic counselor
 - Geneticist
 - Hepatologist
 - Neurologist
 - Nutritionist
 - Ophthalmologist
 - Pathologist
 - Psychiatrist
 - Radiologist

TIMELY DIAGNOSIS

Items in green are typically only seen with a Timely Diagnosis

PRE-DIAGNOSIS

- Typical Accompanying Diagnoses**
 - Elevated serum aminotransferase activity (AST, ALT)
 - Hepatitis
 - Fatty liver
 - GERD
 - Hepatitis
 - Kayser-Fleischer rings
- Testing & Procedures**
 - Prenatal screening**
 - Ceruloplasmin level
 - Urinary copper level
- Specialists**
 - Gastroenterologist
 - Hepatologist
 - Nutritionist
 - Ophthalmologist
 - Pathologist
- Events**
 - Emergency department visits
 - Hospitalizations

TIME OF DIAGNOSIS (by age 2)

- Typical Accompanying Diagnoses**
 - All previously listed diagnoses may remain as accompanying diagnoses as treatment begins.*
- Treatments & Supportive Therapies**
 - Copper chelating – *penicillamine, trientine dihydrochloride*
 - Zinc acetate
 - Nutrition adjustments
- Testing & Procedures**
 - Gene sequencing of ATP7B gene
 - Liver biopsy
 - Liver function tests
 - Metabolic panel
 - Slit-lamp eye exam
 - 24-hour urine collection
- Specialists**
 - Counseling Support Services
 - Gastroenterologist
 - Genetic counselor, Geneticist
 - Hepatologist
 - Nutritionist
 - Ophthalmologist

POST DIAGNOSIS

- Treatments & Supportive Therapies**
 - Copper chelating – *penicillamine, trientine dihydrochloride*
 - Zinc acetate
 - Nutrition adjustments
- Testing & Procedures**
 - Long-term monitoring of liver function
 - 24-hour urine collection
- Specialists**
 - Counseling Support Services
 - Gastroenterologist
 - Genetic counselor
 - Geneticist
 - Hepatologist
 - Nutritionist
 - Ophthalmologist

*This guide is not intended as a clinical decision-making tool. Please consult your specialists for any medical decisions.