
به نام
خدایی
که دانندهٔ رازهاست

انجمن بیماری های متابولیک و دیابت

انجمن بیماری های متابولیک و دیابت

- شروع به کار:

تیر ماه

1399

انجمن بیماری های متابولیک و دیابت

- اعضا موسس:
- دکتر محمدحسین انباردار (پاتولوژیست)
- دکتر محمدهادی ایمانیه (گوارش کودکان)
- دکتر سرور اینالو (مغز و اعصاب کودکان)
- دکتر زهرا بیضایی (دکترای سلولی و مولکولی)
- دکتر سید علیرضا دستغیب (ژنتیک) (پاتولوژیست)
- دکتر بیتا گرامی زاده
- دکتر حسین مروج (غدد و متابولیسم کودکان)
- دکتر نیما مهدی زادگان (قلب کودکان)
- دکتر سید جلیل معصومی

انجمن بیماری های متابولیک و دیابت

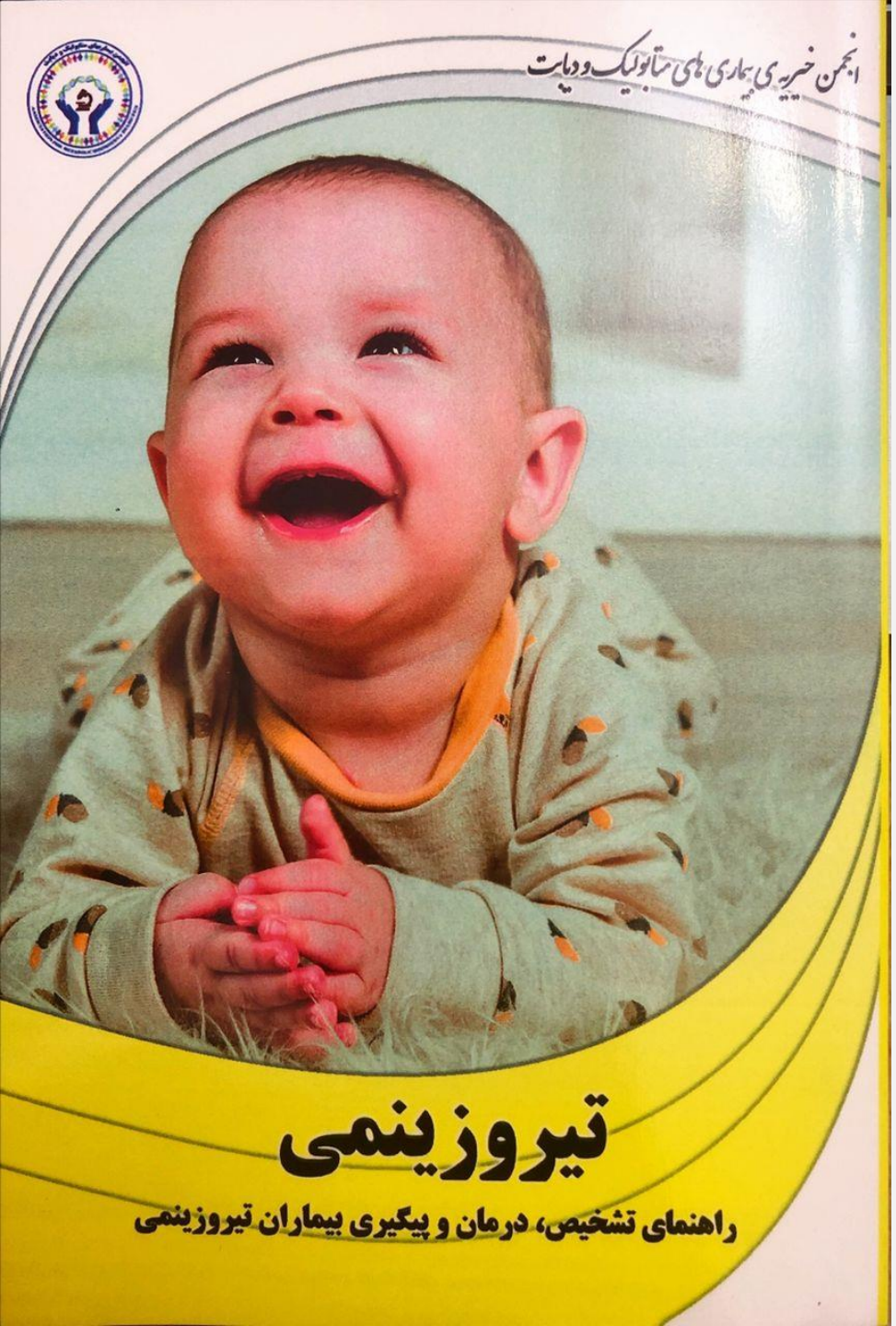
- **هدف:**

تلاش در جهت بهبود شرایط تشخیص و درمان کودکان مبتلا به بیماری های متابولیک

و دیابت

اقدامات

- تهیه راهنمای تشخیص، درمان و پیگیری بیماران گالاکتوزمی متناسب با امکانات کشور

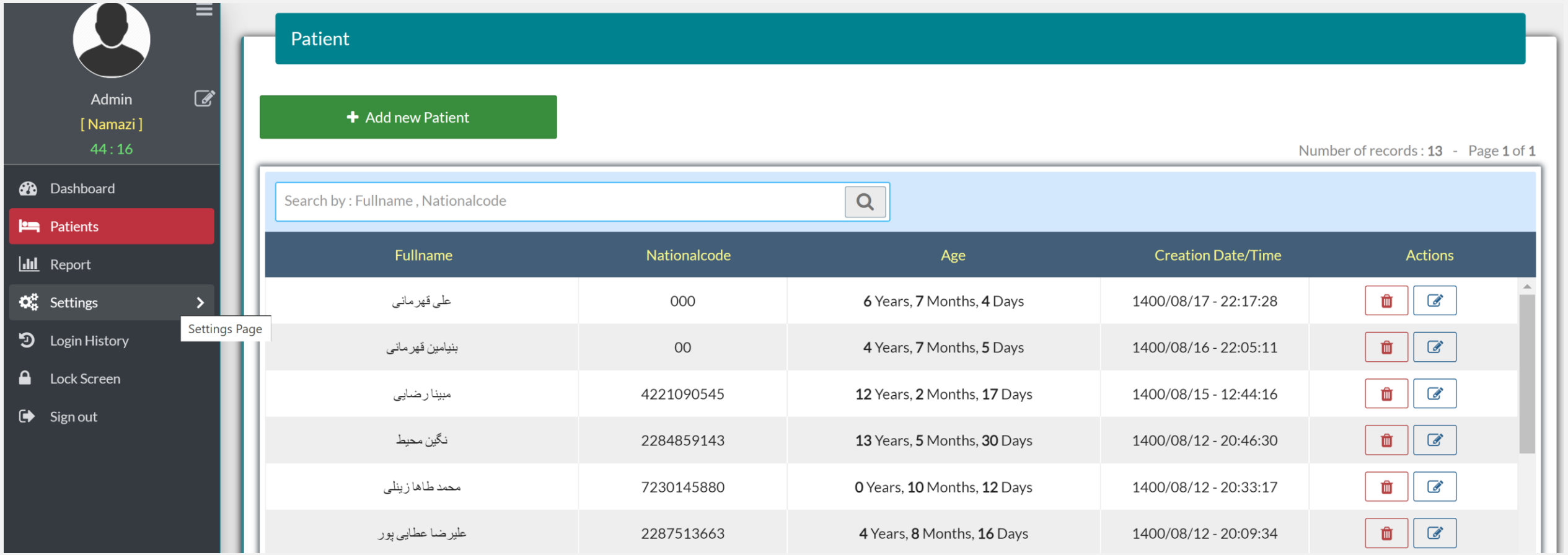


اقدامات

- تهیه راهنمای تشخیص، درمان و پیگیری بیماران تیروزینمی نوع یک، متناسب با امکانات کشور

اقدامات

- ایجاد سیستم رجیستری جهت ثبت بیماران متابولیک و دیابت، با همکاری مرکز تحقیقات نوزادان



اقدامات

- شناسایی بیماران نیازمند و تلاش برای کاهش مشکلات آنها
- همکاری با بهزیستی و مراکز کاردرمانی و گفتار درمانی سطح استان

اقدامات

- آموزش بیماران با استفاده از واتس اپ و اینستاگرام و سایت
- **www.metabolicda.com**

انجمن بیماری های متابولیک و دیابت

- شما هم تمایل به همکاری دارید؟

👇 لینک گروه واتساپ

https://chat.whatsapp.com/CKXCk1ObwYTHLdLOAzlvuY

لینک اینستا
👇

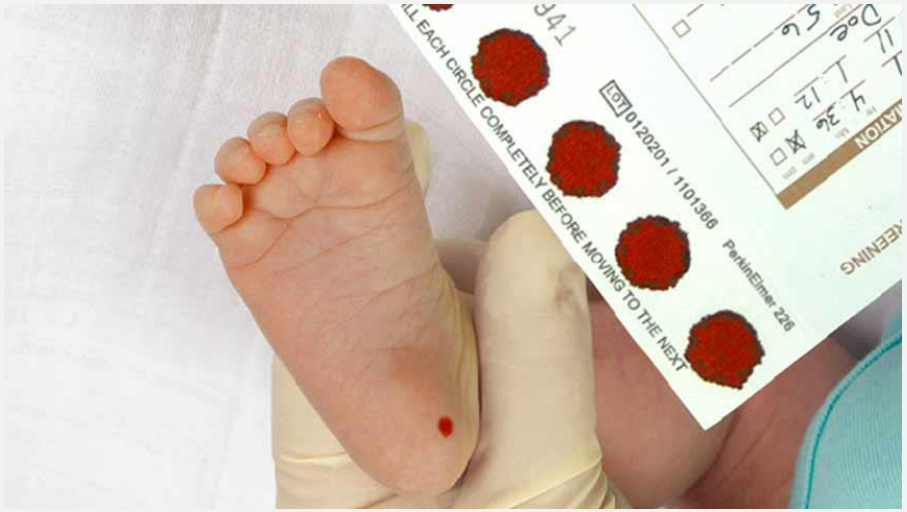
https://www.instagram.com/metabolic_diabetes_foundation

مرکز بیماری های متابولیک و دیابت :
👇
09174845891
071_32337031

GALACTOSEMIA, DIAGNOSIS & MONITORING

WHEN DO WE SUSPECT CLASSIC GALACTOSEMIA (CG)?

- Neonatal screening
- Symptomatic patient



SCREENING

- Neonatal screening for CG in Fars province:
- Total galactose level in DBS at 3-5 days
- Galactose levels above 4 mg/dl are referred.
- Galactose levels above 10 mg/dl (or 20) are usually significant.

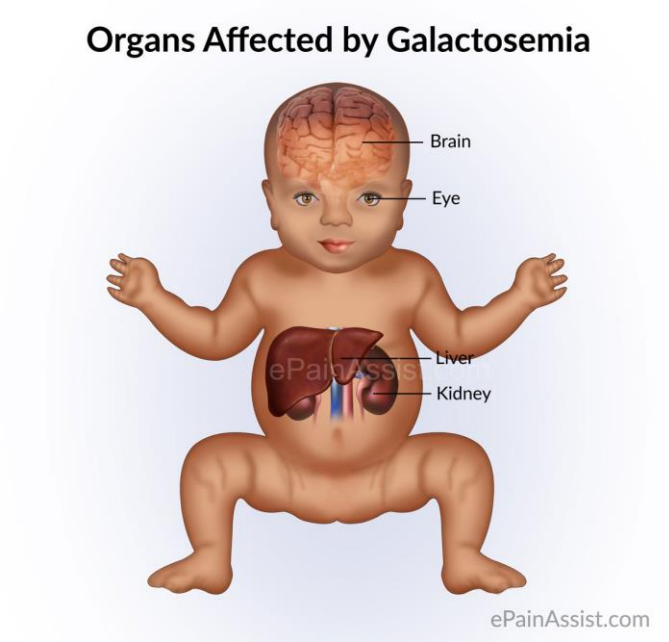
SCREENING

- Neonatal screening for CG in developed countries:
- Galactose, Galactose 1 phosphate, and RBC GALT levels are measured.

DIFFERENTIAL DIAGNOSIS OF HIGH GALACTOSE LEVEL

- Normal newborn (6-10mg/dl)
- G6PD deficiency
- Liver dysfunction, portosystemic vascular shunt, biliary atresia
- Fanconi Bieckle syndrome
- Citrullinemia type 2

- Clinical manifestations of CG (the most common and severe type)



EARLY MANIFESTATIONS : (FIRST FEW DAYS AFTER BIRTH)

- Jaundice
- Vomiting
- Diarrhea
- heptomegaly
- FTT
- poor feeding
- lethargy

EARLY MANIFESTATIONS : (FIRST FEW DAYS AFTER BIRTH)

- Sepsis (especially with Ecoli)
- Coagulopathy
- Ascites
- Edema
- Excessive bruising or bleeding

EARLY MANIFESTATIONS : (FIRST FEW DAYS AFTER BIRTH)

- Seizure
- Hypotonia
- Cataract may be present at birth but generally appear after 2 weeks

LATE MANIFESTATIONS OF CG

- Neuro developmental impairment
- Cataract
- Growth delay
- Premature ovarian failure

LABORATORY FINDINGS

- Increased plasma galactose
- Hypoglycemia; not a primary manifestation:

  Lethargy , poor feeding and liver dysfunction can result in hypoglycemia
- Liver dysfunction:

  Direct hyperbilirubinemia, Elevated transaminases, Coagulopatlny

LABORATORY FINDINGS

- Increased plasma aminoacids: phenylalanine , Tyrosine, Methionine
- Renal tubular acidosis:
- Metabolic acidosis , galactosuria (urine reducing substance), glycosuria , aminoaciduria, albuminuria
- Hemolytic anemia

GALACTOKINASE DEFICIENCY

- Cataract : bilateral, resolves with dietary therapy
- No liver, kidney or brain damage
- Pseudotumor cerebri: rare

URIDINE DIPHOSPHATE GALACTOSE 4-EPIMERASE DEFICIENCY

- 1- Deficiency confined to RBC: Asymptomatic
- 2- Generalized deficiency (very rare):

  Dysmorphic face

  Sensorineural hearing loss

  FTT

  Global developmental delay

MONITORING

MONITORING

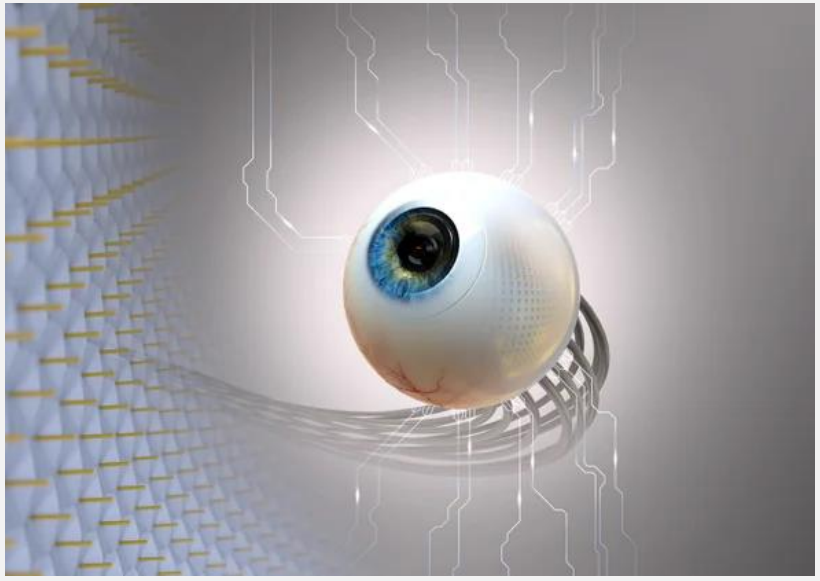
- Every 3 mo follow up until 1 yr, our suggestion include 1 month after diagnosis
- Every 4 mo until 2 yr
- Every 6 mo until 14 yr
- Then annually

- RBC galactose 1 ph: Reflects galactose intake in last 24 hr, not correlate with long term outcome
- Measure with every follow up
- Urinary galactitol: better reflect the long term galactose intake

- Overall, the role of laboratory monitoring is unclear
- Because the rate of endogenous production of galactose exceeds nondairy products, elevated galactose level likely reflects endogenous production rather than non-compliance

OPHTHALMOLOGY EXAM

- Eye exam to detect cataract: at diagnosis, every 6 mo until 3 yr, then annually



GROWTH

- Post natal growth (height and weight) is lower in CG
- Diet should be assessed by a nutritionist annually or more frequently, record of the patient's diet in last 3 days

NEURODEVELOPMENTAL ASSESSMENT

- Regular assessment of speech and cognitive function
- Speech therapy if needed

NEURODEVELOPMENTAL ASSESSMENT

- Most CG have intellectual deficit, but some have normal or above normal intelligence
- Many children have speech and language problems
- IQ and cognitive function may decrease with age.

SPEECH PROBLEMS

- Delayed acquisition of vocabulary
- Difficulty with articulation (verbal dyspraxia)

NEURODEVELOPMENTAL ASSESSMENT

- Adolescents and adults often have focal neurologic findings such as tremor, ataxia, dysmetria
- Other neurological findings: abnormalities in coordination, gait, balance
- Dietary compliance and RBC galactose 1 ph levels do not affect IQ.

OVARIAN FAILURE

- Most women with CG have hypergonadotropic hypogonadism.
- Mechanism: galactose and its metabolites may be toxic to the ovaries.
- AMH level may predict ovarian function.
- The risk of POF may be influenced by the genotype.

OVARIAN FAILURE

- Most women with CG are infertile however, spontaneous pregnancy has been reported.
- Pubertal development and fertility in males with CG are normal.

BONE HEALTH

- Bone densitometry at 8-10 yr,
- If normal: repeat after the puberty is complete.
- To prevent osteoporosis:
- Optimization of calcium intake
- Vitamin D supplementation
- Regular exercise
- Hormone replacement if needed

PROGNOSIS

- The principal cause of early mortality in CG is sepsis (esp. E coli)
- Most patients with CG are healthy and inellectually normal in childhood. However they frequently develop neuropsychological and ovarian problems in teenage years.

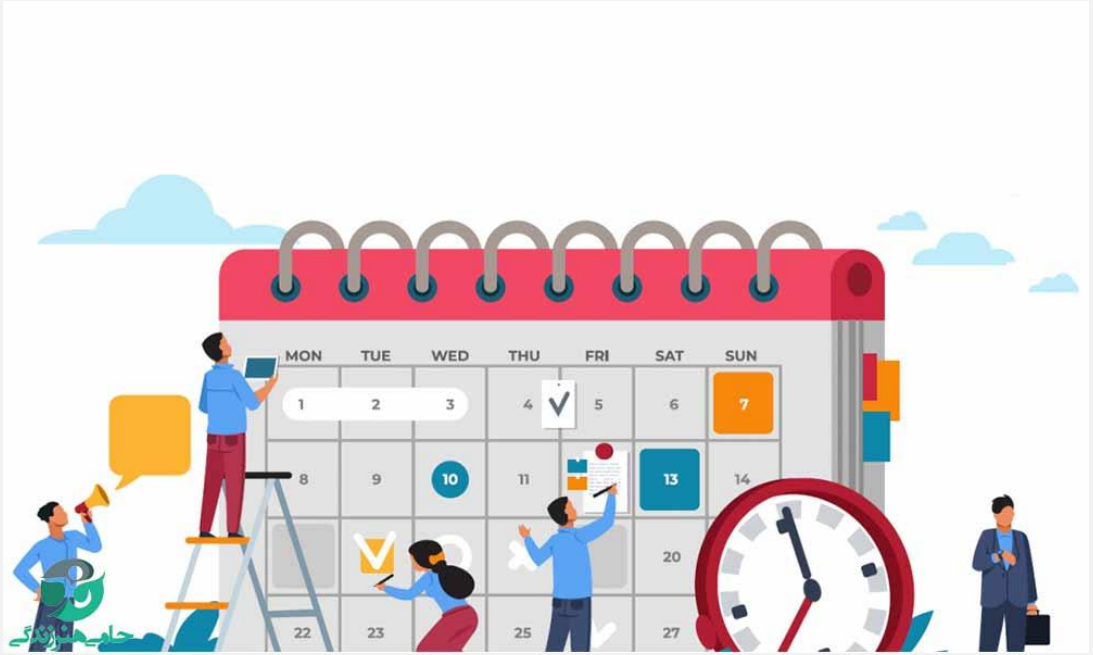
OUTCOME

OUTCOME

Early diagnosis and treatment result in Improvement of:

- Liver function
- Susceptibility to infection
- FTT
- Cataract
- Unfortunately, neuropsychiatric and ovarian problems occur in most adolescents and adults

FOLLOW UP



AT DIAGNOSIS

- General physical exam
- Neurological exam
- Development
- Wt, Ht, HC
- Ophthalmologic exam
- Education about diet

AT DIAGNOSIS

- AST, ALT, PT, INR, T.B, DB
- Blood Galactose, Galactose.1.phosphate
- Urine galactitol, urine reducing agent
- Serum ca, Vit.D

1 MONTH (OUR SUGGESTION)

- General physical exam
- Neurological exam
- Development
- Wt, Ht, HC
- Education about diet
- AST, ALT, PT, INR, T.B, DB
- Blood Galactose, Galactose.1.phosphate
- Urine galactitol, urine reducing agent

3 MONTHS

General physical exam

Neurologic exam

Development

AST, ALT, PT, INR, T.B, DB

Blood Galactose, Galactose.1.phosphate

Urine galactitol, urine reducing agent

6 MONTHS OLD, AND THEN EVERY 6 MONTHS

- General physical exam
- Neurologic exam
- Development
- AST, ALT, PT, INR, T.B, DB
- Blood Galactose, Galactose.1.phosphate
- Urine galactitol, urine reducing agent
- Ophthalmology exam


به امید دیدار