# Screening of the deficiency of glucose-6-phosphate dehydrogenase of newborn babies

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**Key words:** reproductive period, hyperandrogenia, hypothyroidism

Screening programs are considered important method for primary prophylactics of genetic pathology of children. Newborn screening or neonatal screening are selection programs directed at revealing the tests, express methods and examinations of which results are obtained in a short period of time.

The disease of deficiency of glucose-6-phosphate dehydrogenase of the newborn babies is prevalently spread in Azerbaijan, so the screening programs related to such deficiency is of great importance. This indicator is 29-36% in the endemic zones.

Deficiency of deficiency of glucose-6-phosphate dehydrogenase (G-6PD) is a genetic disease that leads to hemolytic anemia. The disease appears as a result of substitution of amino acid by another one in enzyme. Unlike full disruption of protein synthesis, the disease is milder during partial disruption. Full disruption of G-6PD enzyme synthesis leads to severe hemolytic anemia.

Chronic form of hemolytic anemia is related to G-6PD enzyme deficiency and is much similar to the clinical properties of micro spherical hemolytic anemia. The patients mostly complain about fatigue and the increase of jaundice on their skin as a result of infectious diseases. Anemia and spleen enlargement are observed, erythrocytes keep their normal size in blood, yet the activity of G-6PD enzyme reduces. Severe hemolytic anemia is related to deficiency of enzyme and appears as a result of use of preparations against malaria that produce hemolytic erythrocytes, medications against tuberculosis, sulfonamides, a number of nitro furan preparations and salicylic acid. The disease may progress without any medications during the infectious diseases. Protein deficiency in the organism may lead to severe clinical complications. The newborn babies with G-6PD enzyme deficiency may have anemia and hyperbilirubinemia.

When positive results are observed, withdrawal from some medications and removal of legumes from the food ration of patients reduce the emergence of severe hemolytic anemia and other aggravations.

G-6PD enzyme of the 1782 newborn babies was examined in the Scientific and Research Institute of Obstetrics and Gynecology, 178 of which were known to have 9.9% enzyme deficiency.

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## Yenidoğulmuş uşaqlarda qlükoza 6 – fosfatdehidrohenazanın defisitinin skrininqi

### SUMMARY

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***Açar sözlər:*** *reproduktiv dövr, hiperandrogeniya, hipotireoz*

Qlukoza 6 fosfat dehidrogenaza fermentinin çatışmazlığı X ilişikli xəstəlik olmaqla, kəskin infeksiya və yaxud oksidant qəbulundan (salisilat və sulfanilamid) sonra hemoliz verməklə biruzə verir. Diaqnostikası qanda Q6-FD-nin miqdarının müəyyən edilməsinə əsaslanır, hərçənd ki, kəskin hemoliz zamanı analizin cavabı yanlış ola bilər. Müalicəsi dəstəkləyicidir. Patologiya X ilişikli olduğuna görə xəstəliyin tam kliniki təzahürü kişilərdə və homoziqot qadınlarda müşahidə olunur, heteroziqot qadınlarda isə dəyişkən xarakter daşıyır.

## Скрининг дефицита глюкозе – 6 - фосфатдегидрогеназы новорожденных детей

### РЕЗЮМЕ

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***Ключевые слова:*** *репродуктивный период, гиперандрогения, гипотиреоз.*

Дефицит глюкозо – 6 - фосфатдегидрогеназы (Г6ФД) – Х - сцепленный ферментный дефект, который может проявляться гемолизом после перенесенных острых инфекций или приема оксидантов (салицилаты и сульфаниламиды). Диагностика основана на определении уровня Г6ФД, хотя анализ часто дает ложноотрицательный результат при наличии острого гемолиза. Лечение поддерживающее. Поскольку данная патология является Х-сцепленной, полная клиническая картина наблюдается у мужчин и гомозиготных женщин, у гетерозиготных женщин клинические проявления вариабельны.