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G6pd blood test

G6PD Deficiency: A Genetic Disorder That Affects Red Blood Cells G6PD Deficiency: Causes and Symptoms G6PD deficiency is a condition that affects the G6PD enzyme in red blood cells, making them more susceptible to oxidative stress. This can lead to episodes of hemolysis, which can be triggered by various factors such as infection or certain medications. People at risk for G6PD deficiency may benefit from screening tests, which can help identify those who need supportive care. Treatment is often limited to avoiding triggers and taking supportive measures, but severe cases may require hospitalization and treatment with oxygen, fluids, and blood transfusions. The condition is more common in certain populations, particularly black males in the US, where it affects around 10% of the population. Interestingly, G6PD deficiency has been linked to partial protection against malaria, as carriers of the enzyme are thought to have a reduced risk of infection. Diagnosis is typically made through tests that detect the generation of NADPH from NADP. The most common mutations affecting G6PD occur on the X chromosome and can lead to different classes of disease severity. #### Key Points * **G6PD deficiency** affects red blood cells, making them more susceptible to oxidative stress. * **Symptoms**: Episodes of hemolysis, which can be triggered by infection or certain medications. * **Risk factors**: Certain populations, particularly black males in the US. * **Link to malaria**: Carriers of G6PD deficiency may have partial protection against malarial infection. * **Diagnosis**: Tests that detect the generation of NADPH from NADP. Fluorescence under ultraviolet light is a characteristic that can be observed in various medical tests. In field research settings where rapid screening of large patient groups is required, alternative methods are employed; however, these often necessitate more definitive testing to confirm abnormal results. Polymerase chain reaction-based tests detect specific mutations and are utilized for population screenings, family studies, or prenatal diagnoses. Nonetheless, some patients with acute hemolysis may experience falsely negative G6PD deficiency test results due to the presence of older erythrocytes with a higher enzyme deficiency that have been destroyed. Diagnosing G6PD deficiency can be challenging in female heterozygotes, as X-chromosome mosaicism often leads to partial deficiencies that are not reliably detected by screening tests. G6PD deficiency is classified as one of the congenital hemolytic anemias and should be considered in children with a family history of jaundice, anemia, splenomegaly, or cholelithiasis, particularly those of Mediterranean or African ancestry. Infection, exposure to oxidative drugs, or ingestion of fava beans can trigger acute hemolytic reactions in individuals with G6PD deficiency. This condition should be ruled out as a cause of chronic nonspherocytic hemolytic anemia across all populations. Although rare, G6PD deficiency is also considered a potential cause of neonatal hyperbilirubinemia. The prevalence of neonatal hyperbilirubinemia is higher in males who carry the defective gene or homozygous females, but it rarely occurs in heterozygous females. The exact mechanism behind G6PD deficiency-caused neonatal hyperbilirubinemia is not fully understood, and hemolysis may be observed in affected infants with jaundice. However, other mechanisms appear to play a more significant role in the development of hyperbilirubinemia. Impaired bilirubin conjugation and clearance by the liver leads to indirect hyperbilirubinemia, which is likely secondary to G6PD deficiency. Infants with specific mutations affecting UDPGT-1 are particularly susceptible to this condition. Given text: this has not been observed.18 This may reflect genetic mutations specific to different ethnic groups.18,19 Acute hemolysis is caused by infection, ingestion of fava beans, or exposure to an oxidative drug.3 Medications that should be avoided in patients with G6PD deficiency are listed in Table 3,6 and drugs that can be used safely in these patients are listed in Table 4.6 Hemolysis occurs after exposure to the stressor but does not continue despite continued infection or ingestion. This is thought to be a result of older erythrocytes having the greatest enzyme deficiency and undergoing hemolysis first. Once the population of deficient erythrocytes has been hemolyzed, younger erythrocytes and reticulocytes that typically have higher levels of enzyme activity are able to sustain the oxidative damage without hemolysis.7 Clinically, acute hemolysis can cause back or abdominal pain and jaundice secondary to a rise in unconjugated bilirubin (Table 521). Jaundice, in the setting of normal liver function, typically does not occur until greater than 50 percent of the erythrocytes have been hemolyzed.21 The rightsholder did not grant rights to reproduce this item in electronic media. For the missing item, see the original print version of this publication. Drugs that cause hemolysis in G6PD-deficient persons inf lict oxidative damage to erythrocytes leading to erythrocyte destruction. Hemolysis typically occurs 24 to 72 hours after ingestion, with resolution within four to seven days.21 Oxidative drugs ingested by a woman who is breast-feeding may be transmitted in breast milk and can cause acute hemolysis in a G6PD-deficient child.16,28 Although persons who experience hemolysis after the ingestion of fava beans can be presumed to have G6PD deficiency, not all of them will exhibit hemolysis.6,7 Favism is most common in persons with G6PD class II variants, but rarely it can occur in patients with the G6PD A–variant.5 Fava beans (Table 6) are presumed to cause oxidative damage by an unknown component, possibly vicine, convicine, or isouramil.6,7 Infection is the most common cause of acute hemolysis in G6PD-deficient persons,6 although the exact mechanism by which this occurs is unknown. Leukocytes may release oxidants during phagocytosis that cause oxidative stress to the erythrocytes; however, this explanation alone would not account for the variety of infections associated with hemolysis in G6PD-deficient persons. The most common infectious agents causing hemolysis include Salmonella,Escherichia coli, beta-hemolytic streptococci, rickettsial infections, viral hepatitis, and influenza A. A peripheral smear taken during an acute hemolytic reaction in a G6PD-deficient person may demonstrate Heinz bodies, although this rarely is seen in clinical practice.7 In chronic nonspherocytic hemolytic anemia, which usually is caused by a sporadic gene mutation, hemolysis occurs during normal erythrocyte metabolism.5,6 The severity of the hemolysis varies, causing mild hemolysis to transfusion-dependent anemia. Exposure to oxidative stress can cause acute hemolysis in these persons. Other Clinical Considerations G6PD-deficient persons are predisposed to the Of sepsis and complications linked to severe injuries.29 Research has struggled to consistently show a significant risk for patients receiving G6PD-deficient blood, leading blood banks to reject such donors.30 Treatment for G6PD deficiency primarily involves avoiding oxidative stressors. In rare cases, anemia may be severe enough to warrant a blood transfusion, although splenectomy is generally not recommended. Folic acid and iron might help with hemolysis, as the condition often presents without symptoms and associated hemolysis is usually short-lived. Antioxidants like vitamin E and selenium have no proven benefit for treating G6PD deficiency.6,31 Research is ongoing to identify medications that may prevent oxidative-induced hemolysis in G6PD-deficient red blood cells.32 Page 2 Am Fam Physician. 2005;72(7):1285-1292 Author disclosure: Nothing to disclose. Milk thistle has been used as a cytoprotectant for liver disease, cancer treatment and prevention, and supportive treatment of Amanita phalloides poisoning. Clinical studies are largely heterogeneous and contradictory. Mild gastrointestinal distress and allergic reactions aside, side effects are rare, and serious toxicity is rarely reported. Standardized oral milk thistle appears to be safe for up to 41 months of use, with no significant drug reactions reported. Ongoing clinical trials in oncology and infectious disease will help determine the efficacy and effectiveness of milk thistle. Milk thistle (Silybum marianum) has been used since classical Greece to treat liver and gallbladder diseases, as well as protect the liver against toxins. It has recently been investigated for use as a cytoprotectant, anticarcinogen, and supportive treatment for liver damage from Amanita phalloides poisoning. Silymarin is its active ingredient, primarily found in seeds. Silymarin undergoes enterohepatic recirculation, resulting in higher concentrations in liver cells than serum. It is composed of components called flavonolignans, with silybin being the most common. Several studies suggest that silymarin is anti-inflammatory, regulating inflammatory mediators like tumor necrosis factor (TNF), TNF-alpha, nitrous oxide, interleukin-6, and interleukin-1 receptor antagonist. Silymarin also increases lymphocyte proliferation, interferon gamma, interleukin-4, and interleukin-10 cytokines in a dose-dependent manner. Studies have shown that tetrachloride can be reduced by milk thistle through lower lipid peroxidation levels. Another study found that silymarin slowed down liver fibrosis in baboons caused by excessive drinking. Laboratory and animal studies suggest that milk thistle may have anticancer properties, potentially preventing cancers of the prostate, breast, skin, colon, tongue, and bladder. In the United States, milk thistle is commonly used to treat viral infections and liver cirrhosis. However, clinical trials have yielded mixed results. A study on patients with cirrhosis found that those treated with Legalon, a product containing 70-80% silymarin, had lower mortality rates compared to those given a placebo. The benefits were most pronounced in individuals whose cirrhosis was caused by alcoholism and those who had less severe liver disease at the start of treatment. Another study on patients with chronic alcoholic liver disease found that Legalon normalized their liver enzyme levels and improved histology. In contrast, a different study failed to replicate these positive effects. A smaller trial involving 20 patients with active hepatitis found that silybin combined with phosphatidylcholine resulted in significantly lower liver enzyme levels compared to the placebo group. However, other studies have not produced similar results, and two meta-analyses concluded that the existing data are insufficient to support the use of milk thistle for liver disease due to limitations such as inconsistent inclusion criteria, non-comparable doses, and failure to account for confounding factors like hepatitis infections and ongoing alcohol consumption. Silymarin's efficacy varies greatly from one week to a prolonged period of 41 months without any consensus on minimum duration. Research suggests its effects are dose-dependent, but the bioavailability of different formulations is unclear due to diverse study products and dosages. This ambiguity complicates comparison between studies. Efforts are underway to isolate and characterize silymarin's components for more accurate testing. Studies on milk thistle as a cytoprotectant in humans have been limited. A notable study found that 49 out of 200 workers exposed to toluene or xylene for extended periods developed persistent elevations in transaminase levels; 30 of these patients were treated with Legalon, which showed improvement over the untreated group. Researchers are investigating milk thistle's active ingredients for cancer prevention and treatment. Phase trials are under way for human prostate cancer patients and those with acute lymphoblastic leukemia receiving hepatotoxic chemotherapy. Silymarin has been used to treat Amanita phalloides poisoning by inhibiting toxin binding to liver cells and interrupting the enterohepatic circulation of toxins. However, its effectiveness in combination treatments is unknown due to small patient numbers and varying treatment regimens. Side effects from milk thistle are rare, with most complaints being gastrointestinal disturbances similar to placebo incidence. Allergic reactions, while possible, are uncommon, and drug interactions do not appear significant. Given text appears to be a section of a medical research article discussing the effects of various treatments on postnatal depression and exploring their efficacy, safety, tolerability, dosage, and cost. The treatments mentioned include antidepressants (such as fluoxetine), milk thistle, and nondrug interventions such as counseling, cognitive behavior therapy, interpersonal psychotherapy, and psychodynamic therapy. We examined various treatments for postpartum depression but found no studies on light therapy. Cognitive Behavioral Therapy (Group) was shown to improve symptoms in women with high depressive levels after six months compared to routine care. Psychoeducation with partners reduced patients' and partners' psychiatric issues at 10 weeks. Mother-Infant Interaction Coaching had limited effect on maternal depression, but it improved responsiveness towards the infant within 10 weeks. Telephone-Based Peer Support (Mother-to-Mother) also showed reduced depression scores at four months. Postnatal depression is generally defined as nonpsychotic depression occurring in the first six postpartum months. Puerperal mental disorders have only been separately categorized recently, but ICD-10 and DSM-IV require specific qualifications limiting their use. Clinically, a broader definition is often used due to its implications for nursing mothers and developing infants. Symptoms of postnatal depression resemble those at other times in life but include guilt about caring for the new baby. The Edinburgh Postnatal Depression Scale identifies depressive symptoms in many countries. Incidence rates are similar to general depression in women, but triple that of nonchildbearing women in the first month after childbirth. Studies across cultures show consistent 10-15% incidence rates, with higher rates among teenage mothers. Four systematic reviews identified risk factors for postpartum depression: past history of psychopathology, low social support, poor marital relationships, and recent life events. From various studies, including those from India and China, it has been observed that spousal disappointment over the sex of a newborn child can be linked to postnatal depression, particularly in cases where the child is a girl. The mother's reaction to the baby's sex may also pose a risk within certain cultural groups. Interestingly, research suggests that postnatal depression tends to resolve on its own within three to six months; however, approximately one out of four mothers continues to experience depression by their child's first birthday. Moreover, studies have shown that postnatal depression can lead to several negative consequences for the child and mother, including reduced attachment security, impaired cognitive development, and increased risk of suicidal behavior in the postpartum period. These associations persist even after controlling for subsequent episodes of depression in the mother. Additionally, a case study presented an adolescent boy who experienced skinfolds on his scalp, prompting him to seek answers about their cause. The patient's physical examination revealed obesity but no other family members with similar findings. To induce uniform changes throughout the scalp, Nevus of Ota is a congenital melanin patch typically located in the ophthalmomaxillary region. Cerebriform nevus appears at birth and doesn't cover the entire scalp. Breastfeeding with mothers taking combined oral contraceptives has raised concerns about infant safety. While there's no conclusive evidence of harm, few studies have been conducted, and those that exist are limited by their quality. Combined oral contraceptives may reduce breast milk volume but don't seem to affect infant growth. These pills are more effective than progestin-only options for most patients, but concerns remain about their impact on breastfeeding infants. A Cochrane review examined three trials involving 371 participants, comparing combined oral contraceptives to controls. The reviewed studies had limitations due to unclear randomization methods, high loss-to-follow-up rates, and inadequate sample size calculations. Two of the trials published in 1966 and 1970 reported conflicting results regarding milk volume and lactation duration. However, these studies used outdated estrogen doses that are no longer used. A more recent trial from 1984 compared combined oral contraceptives to progestin-only pills, finding statistically significant declines in breast milk volume but no impact on infant growth or milk composition. The data from this study should be interpreted cautiously due to high loss-to-follow-up rates and the fact that many participants were using supplemental feedings by the 12th week postpartum. The Cochrane reviewers concluded that existing evidence is insufficient to make recommendations about the effects of hormonal contraceptives on breastfeeding infants. The postpartum period is a critical time for breastfeeding mothers considering birth control options. The American College of Obstetricians and Gynecologists recommends waiting six weeks postpartum to initiate combined oral contraceptives, when lactation is well established and infant nutritional status can be closely monitored.5 In contrast, the World Health Organization advises against using low-dose combined oral contraceptives in the first six weeks postpartum for breastfeeding women, instead opting for alternative methods unless other options are unavailable or unacceptable.6 The La Leche League International also recommends avoiding combined oral contraceptives due to available alternatives.7 However, after six months postpartum, the use of low-dose combined oral contraceptives is generally recommended by many health organizations.8 If breastfeeding mothers do consider using combined oral contraceptives, they should be aware of potential decreased breast milk volume and need for infant growth monitoring. Abstinence, barrier methods like condoms, and progesterone-only contraception such as Depo-Provera are viable options immediately postpartum. In a different context, sleepwalking in children can be concerning for parents. It occurs when a child partially wakes up during the night and may perform actions without remembering them later.9 Children who sleepwalk may appear dazed and have clumsy movements.10 When encountering a sleepwalking child, it's crucial to keep objects out of reach, lock doors and windows, and gently guide them back to bed without yelling or making loud noises. Fortunately, most children outgrow sleepwalking, but if the issue persists, consulting a doctor is advisable, as sometimes medication can help treat this condition.11 People who struggle with insomnia may find it challenging to fall asleep or stay asleep due to various factors like stress, caffeine intake, depression, and pain.12 In addition to these causes, changes in work shifts can also contribute to insomnia issues. Is insomnia a major health issue? No, it's not serious. But it can cause fatigue, depression, and irritability. It might also make concentration during the day challenging. How much sleep do I need? Most adults require seven to eight hours of sleep each night. You're getting enough rest if you don't feel tired during the day. Your sleep needs remain relatively constant throughout adulthood, but patterns may change with age. Older people may sleep less at night and take daytime naps. What can my doctor do to identify the cause of my insomnia? Your doctor will ask about your sleep habits, including when you go to bed and wake up, as well as any medications you're taking and your caffeine and alcohol consumption. They might also inquire about whether you smoke or have any pain or snoring issues. If the underlying reason for your insomnia is unclear, your doctor may request a sleep diary to track your bedtime, falling asleep time, nighttime wakings, morning wake-up time, and overall sleep quality. Insomnia can be easily treated once the underlying cause is addressed. The key is to identify what's causing it. Making a few simple changes to your sleep habits can often resolve the issue for many people. What can I do to improve my sleep habits? Here are some suggestions: Go to bed and wake up at the same time every day, including weekends, even if you didn't get enough rest. This helps train your body for night-time sleep. Establish a bedtime routine by doing the same things before going to bed each night, such as taking a warm bath or reading for 10 minutes. Use your bedroom only for sleeping and sex; avoid eating, talking on the phone, or watching TV while in bed. Ensure your bedroom is quiet and dark, using earplugs or a fan if necessary. If you're still awake after 30 minutes of trying to fall asleep, get up and sit quietly in another room for about 20 minutes before returning to bed. Limit your use of caffeine, cold medicines, alcohol, and tobacco. Exercise regularly but not within a few hours of bedtime. Learn stress-reduction techniques to manage life's challenges. Worrying can be overwhelming, so it's a good idea to set aside dedicated time for worrying, such as 30 minutes after dinner to jot down concerns and potential solutions. When preparing for bed, opt for a light snack like warm milk or cheese and crackers instead of a heavy meal. Avoid napping during the day if it tends to disrupt your nighttime sleep. Older adults typically require eight hours of sleep to feel fully alert. However, as people age, they may experience difficulties sleeping due to various factors. Common sleep changes include falling asleep earlier in the evening, struggling to fall asleep at bedtime, or waking up too early and having trouble going back to sleep. These issues can lead to excessive daytime sleepiness. Many factors can cause sleep problems. As adults age, their natural sleep-wake cycle may become less effective. Habits like smoking, consuming alcohol or caffeine, and being sick or in pain can also disrupt sleep. Certain medications can interfere with a person's ability to fall asleep. Additionally, sleep disorders such as sleep apnea, restless legs syndrome, and periodic limb movement disorder can cause insomnia. Sleep apnea involves pauses in breathing during sleep, which can occur hundreds of times per night. This condition is often accompanied by loud snoring and can lead to daytime fatigue. It may also worsen high blood pressure and heart disease. Losing weight, sleeping on one's side, avoiding alcohol and sleep medications, and using a CPAP machine can help alleviate symptoms. Restless legs syndrome is characterized by a creepy-crawly feeling in the legs that may be worse at night. This sensation can keep individuals awake, especially older adults who are more prone to this condition. Applying heat or cold packs, exercising, or rubbing one's legs and feet may provide relief. Medication may also be an effective treatment option. Periodic limb movement disorder is a condition characterized by repetitive movements of the legs during sleep. This issue can disrupt nighttime sleep patterns, leading to daytime fatigue. with this disorder, one or both legs may move uncontrollably during sleep. The person may not even realize they're kicking unless a bed partner brings it up. This can disrupt sleep and cause daytime drowsiness. Some people with restless legs syndrome also experience periodic limb movements during sleep. To improve sleep quality, establish a consistent bedtime routine and wake-up time. Avoid long naps and drinking caffeine in the afternoon or evening. Stop consuming alcohol altogether, as it may initially help you fall asleep but disrupt your sleep patterns later on. Don't lie awake for extended periods trying to fall asleep; instead, get up and engage in quiet activities like reading or listening to soft music before trying again. Consult with your doctor about whether any medications are affecting your ability to sleep at night. Discuss pain or health concerns that may be keeping you awake with your doctor as well. Try exercising daily, as this can help improve sleep quality for many older adults. People with sleep apnea stop breathing for 10-30 seconds while asleep due to airway blockage. This can occur hundreds of times per night, waking the individual from deep sleep and depriving them of adequate rest. Sleep apnea may also cause health problems if left untreated. There are two types: central sleep apnea and obstructive sleep apnea (OSA). OSA is more common, with nine out of 10 people affected having this type. To determine if you have sleep apnea, your bed partner or doctor may first notice the condition. You may be unaware of your nocturnal breathing disruptions but feel tired during the day. Additional symptoms include daytime sleepiness, forgetfulness, and morning headaches. If you suspect you have sleep apnea, consult with your doctor about undergoing tests at a sleep center. Sleep apnea is considered dangerous if left untreated, increasing one's risk of heart disease, stroke, and car accidents caused by drowsy driving. It is essential to seek treatment for this condition. To help manage sleep apnea, make lifestyle changes such as avoiding alcohol and medications that promote relaxation, which can worsen the condition. Given article text here are overweight, lose weight. Sleep on your side instead of on your back. If you still have problems after making these changes, your doctor may have you wear a special mask over your nose and mouth while you are sleeping. This treatment is called “continuous positive airway pressure,” (CPAP, for short). The mask will keep your airway open by adding pressure to the air you breathe. CPAP helps most people with sleep apnea. Sometimes surgery is needed to remove tonsils or extra tissue from the throat. Doctors think that about 12 million Americans have sleep apnea. People who are older than 40 years, especially men, are more likely to have sleep apnea, but it can affect anyone at any age. Will this problem change my life? If you have sleep apnea, it already may have affected you more than you know. Things probably will get better after your doctor diagnoses your sleep apnea. If your sleep problem can be solved by not using alcohol or sleep medicine, losing weight, or sleeping on your side, you’ll start feeling much more rested and have more energy very soon. If you use CPAP, you should feel better soon. If you need surgery, you’ll be able to sleep better afterward. Where can I get more information? American Sleep Apnea Association Telephone: 1–202–293–3650 Page 10 Nightmares are scary dreams. Most children have them from time to time. One out of every four children has nightmares more than once a week. Most nightmares happen between 4 a.m. and 6 a.m. Your child may wake up and come to you for comfort. He or she might be able to tell you what happened in the dream and why it was scary. Your child may have trouble going back to sleep and might have the same dream again. Some children have a different kind of scary dream called a “night terror.” Night terrors usually happen between 1 a.m. and 3 a.a.m.. Your child might wake up screaming, and he or she may be sweating and breathing fast. When this happens, your child is still asleep, but his or her eyes are open. Your child might not answer you if you ask what’s wrong, and it might be hard to wake him or her up. Your child usually does not remember what happened. Will my child keep having nightmares or night terrors? Nightmares and night terrors don’t happen as much as children get older. Nightmares and night terrors probably will stop when your child is a teenager. Some people, especially those who are creative and have active imaginations, may keep having nightmares when they are adults. When should I worry about nightmares or night terrors? Nightmares and night terrors do not mean your child is sick or has mental problems. Nightmares often happen for a few months after a child has a physical or emotional stress. If nightmares keep happening, your child might have trouble with normal activity during the day. Ask your doctor if treatment can help. Make sure your child is safe during the night. Use toddler gates on staircases, and don’t use bunk beds for children who have nightmares or night terrors often. Talk with your doctor if your child gets hurt while he or she is asleep. Your doctor may want to test your child. Page 11 Clinical Question: Is intensive rehabilitation as effective as fusion of the Given study evaluated the effectiveness of spinal fusion surgery versus intensive rehabilitation in improving function and reducing disability in patients with chronic low back pain. In this randomized controlled trial, 349 patients from UK hospitals aged 18-55 years were enrolled after being uncertain about whether to undergo surgery or rehabilitation. The patients underwent either spinal fusion surgery or an outpatient rehabilitation program consisting of exercises, cognitive behavior therapy, and hydrotherapy. Review Summary: The authors comprehensively searched databases and excluded non-English publications, focusing on randomized studies addressing quality of life. After applying exclusion criteria, they identified 34 trials with varied interventions, primarily medications and behavior modifications. Only a few showed improvement in general health-related quality of life, while depression was unaffected. The majority of included trials were plagued by methodological shortcomings, leading to biased findings favoring the intervention. The studies spanned six weeks to two years and evaluated different treatments. Notably, only nine out of 34 trials demonstrated a positive impact on general health-related quality of life. A critical analysis of existing research concludes that weight loss interventions have limited effects on health-related quality of life. Furthermore, the overall quality of the evidence is deemed poor. Glucose-6-phosphate dehydrogenase (G6PD) is an essential enzyme protecting cells from oxidative damage caused by reactive oxygen species. A deficiency in G6PD can lead to hemolytic anemia due to accelerated red blood cell breakdown. Individuals with this condition may experience increased susceptibility to certain foods, such as fava beans, which can trigger hemolytic crises. ### G6PD deficiency is more common in men than women, particularly in areas such as Africa, Asia, and the Mediterranean. In the US, African-American men are most frequently affected. If you experience symptoms like severe tiredness, pale skin, shortness of breath, or jaundice, your doctor may order a G6PD test.