

Gene expression disorders/Questions and case studies

Questions

1. **Translation of mRNA with a size of 2200 bp resulted in a product of size:**
 - A – 1100 bp
 - B – 494 amino acid residues
 - C – 196 kDa
 - D – 985 amino acid residues
2. **Tissue-specific mRNA editing is mainly provided by:**
 - A – mitochondrial specific tRNAs
 - B – eRNA
 - C – gRNA
 - D – ribosomal RNA (rRNA)
3. **Moving the reading frame cannot normally result in this change:**
 - A – to create a new cap – placing the cap in a new place
 - B – to extend the protein
 - C – to change the biological half-life of the protein
 - D – to create a new stop codon
4. **Which answer is incorrect? Nuclear gene expression is regulated by:**
 - A – using Tf
 - B – in the promoter region
 - C – using nuclear receptors
 - D – by feedback according to the amount of mRNA in the mitochondria

Answers

Case reports

Newborn with focal convulsions

The patient, aged 4 days, was left in the neonatal unit because of the occurrence of focal convulsions. Biochemical examination was repeatedly normal. Epileptic activity was detected during the EEG. The pediatric neurologist evaluated the findings as benign focal neonatal epilepsy. An extensive family history revealed a frequent occurrence of epilepsy in the family. A 283insGT mutation in KCNQ2 was demonstrated in the patient.

Questions:

1. Which biochemical examination is meant?
2. What is the biochemical basis of hereditary epilepsy?
3. What does the abbreviation 283insGT mean and what does such a mutation lead to?

Answers

Patient with hypotension and in metabolic disorder

A 4-year-old patient was admitted to the pediatric ward because he had lost consciousness. The examination revealed hypotension, S-K 2.6 mmol/l, pH 7.8 and HCO₃ 52. The Nordin index was 1.4. A P124L mutation was detected in CLC-Kb.

Questions:

1. What disease could it be? And what would be the other laboratory findings to confirm the diagnosis?
2. What is the Nordin index?
3. What is the cause of this syndrome?
4. What is CLC-Kb and what does the abbreviation P124L stand for?

Answers

Patient with colorectal cancer

The patient, age 52, was examined on an outpatient basis for fatigue, low fever, gastrointestinal problems and repeated findings of admixture of fresh blood and sometimes mucus in the stool. During rectoscopy, a biopsy was performed from the suspected tumor site. The biopsy sample was examined histologically (adenocarcinoma) and molecularly genetically for the presence of mutations in the K-ras gene (substitution in position 2 of codon 12, GGT→GCT).

Questions:

1. Which other laboratory tests would be suitable for monitoring the patient?
2. What is the K-ras gene and what is its significance?
3. What is the consequence of the given point mutation?

Answers

Patient with liver cirrhosis

A 55-year-old female patient visited her family physician for persistent weakness, lethargy, loss of libido, and joint pain. Six months ago, she was diagnosed with diabetes mellitus. Physical examination revealed hepatomegaly and hyperpigmentation of the skin. The EKG showed signs of cardiomyopathy. Biochemical tests and a liver biopsy were performed. DNA was isolated from peripheral leukocytes and examined for the presence of the C282Y mutation in the HLA-H (HFE) gene.

Questions:

1. What biochemical tests would be appropriate to investigate? What histological examination was performed on the biopsy specimen?
2. What is the disease and how is it treated?
3. What is the cause of this disease?
4. What does the abbreviation C282Y mean?

Answers

Links

Related articles

- Gene Expression
- Control of gene expression and proteosynthesis in eukaryotes
- Transcript
- Translation

Source

- MASOPUST, Jaroslav – PRŮŠA, Richard. *Patobiochemie metabolických drah*. 1. edition. Praha : Univerzita Karlova, 1999. 182 pp. pp. 214–218. ISBN 80-238-4589-6.