# Thrombophilia Due to Protein C Deficiency, Autosomal Recessive

## Alternative Names
THPH4
Protein C Deficiency, Autosomal Recessive
PROC Deficiency, Autosomal Recessive

## Record Category
Disease phenotype

## WHO-ICD
Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism > Coagulation defects, purpura and other haemorrhagic conditions

## Incidence per 100,000 Live Births
0-1

## OMIM Number
612304

## Mode of Inheritance
Autosomal recessive

## Gene Map Locus
2q14.3

## Description
Congenital protein C deficiency is an autosomal recessive inherited coagulation condition. Patients with deficiency of protein C present with purpura fulminans or massive venous thrombosis few days or hours after birth. Patients with low levels of protein C show milder symptoms. Usually, heterozygous individuals for protein C deficiency are asymptomatic until adulthood when other risk factors such as surgery and/or pregnancy are encountered.

Diagnosis is based on the measurement of protein C levels. In severe deficiency, the protein C activity levels range from 0 to 30% and in case of partial deficiency, the level ranges from 30 to 70%. Treatment approaches include the administration of protein C concentrates or fresh frozen plasma to patients along with anticoagulant therapy and surgery when required.

## Molecular Genetics
Protein C deficiency is caused by mutations in the PROC gene. This gene controls the production of protein C, which plays an important role in the blood clotting cascade. It blocks the activity of two components of the clotting pathway, factor V and Factor VII, thereby preventing clot formation. In addition, PROC plays a major role in controlling inflammation.

## Epidemiology in the Arab World

### Saudi Arabia
Abu-Amero et al. (2003) described a 9-month-old Saudi boy who developed purpura fulminans shortly after birth. He went on to develop scrotal hematoma, following which a diagnosis of protein C deficiency was suspected. The level of protein C activity was below 0.01 U/ml (normal level 0.70–1.40 U/ml) confirming the diagnosis. The patient had hemorrhage in different tissues and organs such as in both testicles and both eyes. Magnetic resonance imaging (MRI) showed extensive bleeding on temporal occipital and parietal lobes. The patient improved on warfarin and protein C concentrate (500 units intravenously/weekly). Parents were consanguineous and the family history was negative for similar illness. A novel homozygous frame-shift mutation in the PROC gene was identified in the proband.

## References
Abu-Amero KK, Al-Hamed MH, Al-Batniji FS. Homozygous protein C deficiency with purpura fulminans: report of a new case and a description of a novel mutation. Blood Coagul Fibrinolysis. 2003; 14(3):303-6. PMID: 12695756.

## Related CTGA Records
Protein C

## External Links
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=745
https://ghr.nlm.nih.gov/condition/protein-c-deficiency

## Contributors

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