Clinical Challenge 3:

A 15 year old Caucasian boy was referred to you by his GP with complaints of worsening abdominal distension over the last 6 months. The boy complained of increased tiredness in the last few months; however he has attributed this to the school work. There are no other reported symptoms. He and his parents were in Egypt for three years and they have moved back to the UK 6 months ago because of the political unrest in the region. His father is an engineer in the oil industry and mother is a teacher. 12 year old sister is fit and well. There is no family history of any significant illness. There is no history of any significant illness in the past and the boy has received hepatitis B vaccination prior to the relocation to Egypt.

On physical examination, he has mild pallor without any clubbing or jaundice. Abdomen was distended. Firm hepatomegaly (up to the level of umbilicus) was noted on palpation, spleen was enlarged and palpable up to the level of umbilicus. No feature of portal hypertension was evident on examination. Other system examination was normal.

Blood tests showed normochromic normocytic anaemia (Hb 92) with platelets 68, WBC 10.7, N 2.5, L 2.8 and eosinophils 5.2. Liver function tests were normal including clotting profile and albumin. US abdomen showed enlarged liver with coarse echo texture, splenomegaly and small mesenteric and hilar lymphadenopathy. Doppler studies showed flow changes consistent with mild portal hypertension. Hepatic veins and IVC, SVC are reported as normal. CXR was normal. Viral serology including hepatitis viruses and HIV were negative. Blood films for malaria were negative three times. Tuberculin test was negative. Blood tests and urine tests for storage disorders and Wilson's disease were negative. Genetic analysis was negative for alfa1 anti trypsin deficiency.

<u>Some more information becomes available:</u>

A bone marrow examination was done which did not show any features of leukaemia or myelodysplasia.
On specific questioning, the boy said that he gets intermittent diarrhoea, but he has not noticed any blood or mucus in the stools.
OGD showed one grade 1 oesophageal varix, stomach was normal. Diffuse patchy redness was noted in duodenum. Duodenal biopsies showed preserved villous architecture, but nonspecific chronic inflammatory changes were noted in the lamina propria. No granulomas were seen.
Liver biopsy showed peri-portal fibrosis, neo vascularisation of portal venous system and granulomas.
Repeat Mantoux test was negative; T- spot result is awaited.

1. What is the diagnosis?
2. What is the treatment of the condition?

Answer to Clinical Challenge 3

This patient has **schistosomiasis infection**. Treatment with **Praziquantel** is usually effective. Schistosomiasis is more commonly found in endemic areas such as Africa and Far East. Humans are infected initially when cercariae found in fresh water bore through the skin, usually the feet. Transmission of the disease cannot occur in the UK, because of the lack of suitable snail hosts. Initial infection may not be noticeable like in this patient. Diagnosis is made by identification of ova in stool or ELISA. However ELISA cannot differentiate between new or old infection.