Transplantation of haemochromatosis liver and intestine into a normal recipient

Background—Haemochromatosis is a common genetic disease leading to iron overload. Although the gene had been identified (HFE), the pathogenesis had not been fully elucidated.3 The inadvertent transplant of a C282Y homozygous liver and intestine provided a unique opportunity to study this problem.

Methods—A 19 year old man underwent orthotopic liver and intestinal transplantation in January 1997 for the treatment of short bowel syndrome secondary to a mid gut volvulus with resection and cholestatic liver disease resulting from total parenteral nutrition. The organ donor was an 18 year old woman posthumously discovered to be a C282Y homozygote for haemochromatosis.

Results—Preoperative recipient blood tests included a serum ferritin of 34 µg/l (normal range 15–300 µg/l) and a transferrin saturation of 10% (normal 20–55%). Transplantation of the intestine and liver was performed as previously described at this medical centre and included duodenal, jejunal, and ileal.1 At 21 months after transplantation the recipient had a great increase in transferrin saturation at 94% with a normal serum ferritin of 103 µg/l. Hepatic iron concentration at four months was 20 µmol/g and at 22 months was 22.3 µmol/g (normal 0–35 µmol/g). Genetic testing for haemochromatosis on his liver graft revealed homozygosity for the C282Y mutation of the HFE gene. Genetic testing on a peripheral blood sample at 21 months was normal (wild type) for the C282Y mutation. The donor family was investigated for haemochromatosis (fig 1). A brother of the donor was an iron loaded homozygote and her mother appeared to be a non-expressing homozygote. The mother had a non-identical twin that was homozygous for the C282Y mutation with abnormal iron studies.

Conclusions—This case suggests that the genetic defect of haemochromatosis has been transplanted into the recipient with the donor intestine and that iron accumulation will probably occur with time. Within 21 months of transplantation the recipient is showing evidence of the typical biochemical abnormality seen in a young patient with haemochromatosis, namely an increase in transferrin saturation with a normal hepatic iron concentration. Although the serum ferritin is normal, it is likely that if untreated it will continue to rise with time. Therefore, we have identified a predisposition to future iron overload rather than iron overload at 21 months. The concomitant transplantation of the haemochromatosis liver is less likely to be contributing to the abnormal iron metabolism. Transplantation of a haemochromatosis liver alone into a normal recipient has previously been documented at this centre with a progressive decline in hepatic iron concentration and a normal radiourex absorption study.5 This supports the hypothesis that the fundamental defect in haemochromatosis is site specific at the level of the intestine rather than a systemic abnormality.

P C ADAMS
G JEFFREY
Department of Medicine, University of Western Ontario, London, Ontario, Canada

K ALANEN
S CHAKRABARTI
Department of Pathology, University of Western Ontario, London, Ontario, Canada

R PRESHAW
Department of Surgery, University of Calgary, Calgary, Alberta, Canada

W HOWSON
Department of Laboratory Medicine, University of Western Ontario, London, Ontario, Canada

D GRANT
Department of Surgery, University of Western Ontario, London, Ontario, Canada

Correspondence to: Paul C Adams, MD, Department of Medicine, London Health Sciences Centre, 339 Windermere Road, London, Ontario, Canada N6A 5A5. Email: padams@julian.uwo.ca


Transplantation of haemochromatosis liver and intestine into a normal recipient

P C ADAMS, G JEFFREY, K ALANEN, S CHAKRABARTI, R PRESHAW, W HOWSON and D GRANT

Gut 1999 45: 783
doi: 10.1136/gut.45.5.783

Updated information and services can be found at:
http://gut.bmj.com/content/45/5/783

These include:

References
This article cites 5 articles, 1 of which you can access for free at:
http://gut.bmj.com/content/45/5/783#BIBL

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/