



Haemochromatosis- treatment and implications of diagnosis

Frances Neville
Clinical Nurse Specialist
Regional Haematology/Oncology Dept.
Tullamore.
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HAI Nurses Group



Content

- Background
- Definitions
- Treatment
- Midland perspective
- Audit
- Nurse role
- Quality of life issues
- Diet
- The future
- Questions



Haemochromatosis- background

- Hereditary haemochromatosis is a disorder of iron metabolism
- Continued absorption from the upper small intestine
- Occurs when a person inherits a defective gene (HFE gene) from each parent
- Seen mostly in adults of northern European extraction.
- Incurable, but early treatment leads to normal life expectancy



Genetic Disease (AR)

>90% cases

HFE gene



Each person has two copies of the same gene

Allele : different form of the same gene

In HFE haemochromatosis two well known alleles

C282Y and H63D

Disease manifestations vary depending on allele combination inherited.



Potential Genetic Combinations

C282Y/C282Y
H63D/H63D
($<1\%$)

Symptomatic (80%)
Mild disease

C282Y / Normal
H63D/Normal

Unknown/Unknown



Iron in the body

- Normal adult - 3g iron
- No system for getting rid of extra iron
- Loss of 1mg/day by loss of cells mainly in skin and bowel
- Women - loss through periods & pregnancy - averages about another 1mg/day



Absorbing iron

- Normal diet 10mg/day
- Normal adult absorbs only 1-2mg ie only what's needed
- If deficient absorbs more - when there's enough the gut cells block more iron getting in
- Block doesn't work in haemochromatosis



How do we diagnose it?

- Suspected - symptoms, Family history
- Often just a “screening” test
- Ferritin - raised usually implies increased iron stores
- Transferrin saturation - more sensitive
- Genetic test



Tests for organ damage

- Liver function tests
- Glucose
- Alpha-fetoprotein
- Ultrasound of liver
- Echocardiogram
- Hormonal tests
- ?Liver biopsy



HH- signs and symptoms

- Unexplained weakness or fatigue
- Joint pain
- Weight loss
- Abdominal pain
- Impotence



Liver biopsy

- Before genetic test, was gold standard to diagnose haemochromatosis
- Now needed in some patients to diagnose cirrhosis of the liver, which is a complication
- Only needed in those who may be more severely affected



What happens after diagnosis?

- Venesections (Therapeutic phlebotomy)
- Every week until target
- Every 2 weeks in mildly affected patients
- Then every 3 months (2-4 months) to keep on target
- Treatment for any complications



HH- disease progression

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- Liver: abnormal liver function, swelling or cirrhosis
 - Pancreas: late onset diabetes
 - Pituitary gland: impotence, irregular menses, infertility and early menopause.
 - Heart: palpitations, irregular heart beat and heart enlargement
 - Brain: depression, confusion and memory loss
 - Joints: swelling of first two finger joints
 - Skin: bronzing



HH- treatment

- Treatment involves the regular removal of blood, phlebotomy, initial treatment may span 12-24 months.
- Maintenance therapy is life-long 3 -4 venesections may be required to maintain normal levels of iron.
- Regular blood testing will indicate when a venesection is necessary.

Venesection





The midland perspective

- Patient attend hospital with results of tests from GP
- May have been screened if first degree relative recently diagnosed
- Consultant will refer to nurses for venesection
- Day ward, Oncology/Haematology Unit, some GP surgeries offer phlebotomy service
- Similar service in Mullingar and Portlaoise Hospitals.



Audit of our service

- **Aim:** to check that all patients were reaching target as defined in our service
- **Methods:** 50 sets of case notes retrospectively reviewed



Audit results

- 41 male 9 female
- Youngest 28m, oldest 74 m
- 25 C282Y homozygous, 15 compound heterozygote
- 4 complained of joint pain
- 6 raised glucose, 3 diabetic
- 18 raised ALT or AST, 1 liver biopsy
- Serum ferritin ranged from 214 to 2132 μ /L
- 2 had abnormal ECHO Reports
- AFP normal for all



Conclusions and recommendations

- On the whole compliance to SOP
- Maintenance phase patients not reviewed by Doctor unless there were problems
- Follow-up
- Nurse- led service has benefit
- MDT referral
- QOL issues need addressing



The Nurses Role

- To provide continuity in care and promote a holistic approach
- Clear and concise explanation of condition and the need for treatment and investigations.
- Verbal information supplemented by written.
- Treatment plan implemented with target set, usually Ferritin <50 ug/l
- Co-ordinates investigations
- Available for patient and family to discuss impact of diagnosis.
- Nurses carry out venesection.
- Need to be actively involved in assessing QOL issues



Diet

- Low iron diet is of little benefit
- No iron supplements or multivitamins containing iron
- No bread or cereal which is fortified with iron



Tea drinking

- Kaltwasser (1998) investigated the effect of black tea on iron storage.
- 18 patients
- 2 groups, tannin rich tea
- Body iron stores measured
- Showed a one third reduction of iron absorption
- Concluded that tea drinking reduced the need for phlebotomies

Oysters.....





Haemochromatosis and raw oysters a dangerous combination....

- Stubblefield (1995) suggests that fatal septicemia may occur in individuals with HH who ingest raw oysters contaminated by *Vibrio vulnificus*
- Patients and families need to be aware
- Can be found in other shellfish

Cereals...

- Check for added iron
- Most have very high levels
- Porridge not fortified



Meat..

- Moderate intake
- Need for well-balanced diet
- Possibly best to limit the intake





Vitamin C

- Vitamin C increases the absorption of iron from gut
- Drinking orange juice at meal times should be avoided



Alcohol

- Alcohol and haemochromatosis not a good combination
- Advice a low or no alcohol particularly in treatment phase



Benefits of treatment

- Simple treatment once diagnosis has been made
- Some symptoms may improve
- Early diagnosis and treatment improve survival and people with no complications have normal life expectancy
- As it is nurse-led, greater deal of flexibility with regard to timing of venesections



What about family members?

- Brothers and sisters
 - We give letter(s) to patient
 - No need to test minors until 18 years old
- Children of patient - very low risk
 - Test partner
- Parents/Uncles/Aunts/Cousins???



Why do we throw away the blood?

- May not meet donor criteria
- Family members who have no symptoms may be
- Issues re cost and incentives
- Issues re follow-up
- Trials in some blood banks in USA, trial with IBTS and Mater



Quality of life issues

- Feeling constantly unwell, reduces confidence and self-esteem
- Treatment can interfere with normal working life
- Poor work performance and loss of concentration may be labelled 'lazy'
- Family and spouse relationships may be affected



The future

- Increase awareness, may lead to early diagnosis
- ?? Screening programme
- Use of blood for blood transfusion, currently been reviewed.



Summary

- Genetic haemochromatosis is a disease of iron overload
- Incurable, but early diagnosis will lead to normal life expectancy
- Nurses have pivotal role in treatment, health promotion and audit and research
- Continued publicity



Thank you , any
questions?