Mid Essex Hospital Services NHS Trust

ANTENATAL REFERRALS FOR TAY SACHS DISEASE	CLINICAL GUIDELINES Register no 08093 Status: Public

Developed in response to:	Intrapartum NICE Guidelines RCOG guideline
Contributes to CQC Core Outcomes	11, 12

Consulted With	Individual/Body	Date
Anita Rao/ Alison Cuthbertson	Clinical Director for Women's, Children's Division	February 2018
Vidya Thakur	Consultant for Obstetrics and Gynaecology	
Dr Hassan	Consultant Paediatrician	
Alison Cuthbertson	Associate Director of Midwifery/Nursing	
Paula Hollis	Lead Midwife Acute Inpatient Services	
Chris Berner	Lead Midwife Clinical Governance	
Ros Bullen-Bell	Lead Midwife Community Services	
Sarah Iskander	Antenatal Clinic Midwife	
Sarah Moon	Specialist Midwife Guidelines and Audit	
Deborah Lepley/Samantha Lasagna	Warner Library	

Professionally Approved By:		
Miss Rao	Lead Consultant for Obstetrics and Gynaecology	February 2018

Version Number	4.0
Issuing Directorate	Women's and Children's
Ratified by	DRAG Chairmans Action
Ratified on	10th April 2018
Implementation Date	23rd April 2018
EMG sign off Date	April/May 2018
Next Review Date	February 2021
Author/Contact for Information	Emma Neate, Antenatal Newborn Screening Co-ordinator
Policy to be followed by	**Midwives, Obstetricians**
Distribution Method	Intranet & Website. Notified on Staff Focus
Related Trust Policies (to be read in conjunction with)	04071 Standard Infection Prevention 08045 Amniocentesis for Antenatal Diagnosis. 08046 Interpret and Act on CVS/Amniocentesis Results. 06035 Referral to Teritiary Unit for Suspected Fetal Abnormality. 06032 Use of Mifepristone, Misoprostil, Gemprost and Prostin in a termination of Pregnancy for Fetal Abnormalities or Intrauterine Death 09062 Maternity Care 06031 Receiving and acting on Test Results in Maternity 09113 Calling Paediatric Staff and obtaining paediatric referral 08076 Clinical Audit Strategy & Policy 2017-2018

Document Review History:

Review No:	Reviewed by:	Issue Date:
1.0	Kathleen Bird	January 2009
2.0	Nicky Leslie	February 2012
2.1	Nicky Leslie – clarification to point 10.0	September 2012
3.0	Nicky Leslie, Antenatal Newborn Screening Co-ordinator	March 2015
4.0	Emma Neate – Full review	23rd April 2018

INDEX

1.0 Purpose

1.1 The purpose of this guideline is to aid medical and midwifery staff in management of women and their partners who are in a high risk population of passing on Tay Sachs disease

2.0 Equality and Diversity

2.1 Mid Essex Hospital NHS Trust is committed to the provision of a service that is fair, accessible and meets the needs of all individuals.

3.0 Background

3.1 Tay-Sachs disease is a rare inherited disorder that progressively destroys nerve cells (neurons) in the brain and spinal cord.

3.2 This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

3.3 The most common form of Tay-Sachs disease becomes apparent in infancy. Infants with this disorder typically appear normal until the age of 3 to 6 months, when their development slows and muscles used for movement weaken.

3.4 Affected infants lose motor skills such as turning over, sitting, and crawling. They also develop an exaggerated startle reaction to loud noises. As the disease progresses, children with Tay-Sachs disease experience seizures, vision and hearing loss, intellectual disability, and paralysis. An eye abnormality called a cherry-red spot, which can be identified with an eye examination, is characteristic of this disorder. Children with this severe infantile form of Tay-Sachs disease usually live only into early childhood.

3.5 Other forms of Tay-Sachs disease are very rare. Signs and symptoms can appear in childhood, adolescence, or adulthood and are usually milder than those seen with the infantile form. Characteristic features include muscle weakness, loss of muscle coordination (ataxia) and other problems with movement, speech problems, and mental illness. These signs and symptoms vary widely among people with late-onset forms of Tay-Sachs disease

4.0 The Aim of the Screening Programme

4.1 If a patient is pregnant, then her partner should also endeavour to attend for screening.

4.2 Tay Sachs is a genetic disease which is inherited in an ‘autosomal recessive’ manner. The parents are at risk of having a baby with Tay Sachs disease if both parents are carriers. If both parents are carriers there is a 1 in 4 chance, with each pregnancy, that the baby could have the disease. If one parent is a carrier, there is a 2 in 4 chance that the baby will be a carrier. If the other parent is not a carrier there is no risk of having a baby with Tay Sachs.

4.3 Being a carrier does not affect the parent’s health, and they would have no way of knowing if they are a carrier, unless they have been tested. Testing is done by a simple blood test.

5.0 Screening

5.1 Anyone can be a carrier of Tay-Sachs

5.2 Testing can be offered to the non-Jewish partners of Jewish people; or anyone from any ethnic group who have a family history of Tay Sachs

5.3 When both parents are carriers, each child has a 25% of having the disease, the carrier rate for the general population is 1:250

5.4 Ashkenazi Jews are all considered high risk with a carrier rate of 1:27

5.5 Ashkenazi Jewish

5.5.1 People of Ashkenazi Jewish descent have a **1 in 5 chance** of being a healthy carrier for **at least one of 9 serious genetic disorders** as follows:

- Bloom syndrome (1 in 100 Ashkenazi Jews are carriers)
- Canavan disease (1 in 40 - 57 Ashkenazi Jews are carriers)
- Cystic fibrosis (1 in 25 - 29 Ashkenazi Jews are carriers)
- Familial dysautonomia (1 in 30 Ashkenazi Jews are carriers)
- Fanconi anaemia type C (1 in 89 Ashkenazi Jews are carriers)
- Glycogen storage disease 1a (1 in 71 Ashkenazi Jews are carriers)
- Mucolipidosis IV (1 in 100 - 125 Ashkenazi Jews are carriers)
- Niemann-Pick disease type A (1 in 90 Ashkenazi Jews are carriers)
- Tay Sachs Disease (1 in 26 - 30 Ashkenazi Jews are carriers)

5.6 The NHS has funds for Tay Sachs screening but not any additional tests. These can be performed via the Jnetics centre see below.

6.0 How and Where to go for Screening:

6.1 Jnetics – Jewish Genetic Disorder Clinic

The clinic at Barnet Hospital provides education about the risks of these conditions and what it means to be a carrier, and gives you the opportunity to discuss the genetic testing options.

There are two tests available.

- You can give a blood sample and be tested for Tay Sachs disease alone - this test is free.
- Alternatively you can book to see a genetic counsellor in the clinic and give a saliva sample to be tested for the 9 common conditions - this test costs £190.

6.2 **Clinic Times**

The genetic counselling clinic takes place at Barnet Hospital and is open 4 days a month:

- 1st Thursday of the month from 8.30am to 11.30am
- 2nd Wednesday of the month from 3.30pm to 6.30pm
- 3rd Thursday of the month from 8.30am to 11.30am
- 4th Thursday of the month from 3.30pm to 6.30pm

Address - Barnet Hospital, Level 2, Breast Clinic, Wellhouse Lane, Barnet, EN5 3DJ

6.3 **How to make an appointment**

If you are of Ashkenazi Jewish descent and would like to make an appointment with a Genetic Counsellor for advice and screening, please book an appointment in the Jnetics Barnet clinic - https://jnetics-barnet-clinic.youcanbook.me/

To only book a free Tay Sachs blood test - https://www.swiftqueue.co.uk/pre_timescreen.php?id=10084

6.4 Pregnant women are advised to bring their partners with them when attending the screening clinic.

6.5 A genetics' counsellor will be available to discuss the screening process, answer any questions; and take blood samples.

7.0 Time Frame for Results

7.1 Routine test results are sent out within 3-4 weeks. Urgent test results (i.e. in pregnancy) are sent out within 2 weeks. Results are sent direct to the couple and also sent to the GP.

8.0 Options Available for Couples with a Known Carrier Status

8.1 Couples who are both carriers of the same disease may want to explore their options for a healthy family.

8.2 A referral will be offered to clinical genetics Great Ormond Street Hospital.

8.3 Prenatal diagnosis can be offered in pregnancy to confirm if the fetus has Tay-Sachs. At-risk couples can choose from two procedures: Chorionic villus sampling (CVS) or Amniocentesis after 16 weeks. These tests are available via the NHS. In either case, if the fetus is affected with Tay-Sachs, couples may elect to have a therapeutic abortion.

9.0 Neonatologist Review

9.1 A neonatal alert form should be completed and sent to the Antenatal and Newborn Screening Co-ordinator, located in the screening office, Antenatal Clinic. A copy of the alert form is kept in the screening office and a copy is forwarded to the named Paediatric Consultant for a plan of care post-delivery. (Refer to the 'Guideline for calling paediatric staff and for obtaining paediatric referral'; register number 09113) (Refer to Appendix A)

9.2 When the named Paediatric Consultant has completed the neonatal alert form with a care plan, a copy will be retained in the neonatal folder. A further copy will be sent to the Screening team who will place a copy in the Labour Ward folder and in the patient's lilac folder.

10.0 Staffing and Training

10.1 All midwifery and obstetric staff must attend yearly mandatory training which includes antenatal screening update.

10.2 All midwifery and obstetric staff are to ensure that their knowledge and skills are up-to-date in order to complete their portfolio for appraisal.

11.0 Infection Prevention

11.1 All staff should follow Trust guidelines on infection prevention by ensuring that they effectively 'decontaminate their hands' before and after each procedure.

11.2 All staff should ensure that they follow Trust guidelines on infection control, using Aseptic Non-Touch Technique (ANTT) when carrying out procedures i.e. obtaining blood samples.

12.0 Audit and Monitoring

12.1 Audit of compliance with this guideline will be considered on an annual audit basis in accordance with the Clinical Audit Strategy and Policy (register number 08076), the Corporate Clinical Audit and Quality Improvement Project Plan and the Maternity annual audit work plan; to encompass national and local audit and clinical governance identifying key harm themes. The Women's and Children's Clinical Audit Group will identify a lead for the audit.

12.2 The findings of the audit will be reported to and approved by the Multi-disciplinary Risk Management Group (MRMG) and an action plan with named leads and timescales will be developed to address any identified deficiencies. Performance against the action plan will be monitored by this group at subsequent meetings.

12.3 The audit report will be reported to the monthly Directorate Governance Meeting (DGM) and significant concerns relating to compliance will be entered on the local Risk Assurance Framework.

12.4 Key findings and learning points from the audit will be submitted to the Patient Safety Group within the integrated learning report.

13.0 Guideline Management

13.1 As an integral part of the knowledge, skills framework, staff are appraised annually to ensure competency in computer skills and the ability to access the current approved guidelines via the Trust's intranet site.

13.2 Quarterly memos are sent to line managers to disseminate to their staff the most currently approved guidelines available via the intranet and clinical guideline folders, located in each designated clinical area.

13.3 Guideline monitors have been nominated to each clinical area to ensure a system whereby obsolete guidelines are archived and newly approved guidelines are now downloaded from the intranet and filed appropriately in the guideline folders. 'Spot checks' are performed on all clinical guidelines quarterly.

13.4 Quarterly Clinical Practices group meetings are held to discuss 'guidelines'. During this meeting the practice development midwife can highlight any areas for further training; possibly involving 'workshops' or to be included in future 'skills and drills' mandatory training sessions

14.0 Communication

14.1 A quarterly 'maternity newsletter' is issued and available to all staff including an update on the latest 'guidelines' information such as a list of newly approved guidelines for staff to acknowledge and familiarise themselves with and practice accordingly.

14.2 Approved guidelines are published monthly in the Trust's Staff Focus that is sent via email to all staff.

14.3 Approved guidelines will be disseminated to appropriate staff quarterly via email.

14.4 Regular memos are posted on the Guideline and Audit notice boards in each clinical area to notify staff of the latest revised guidelines and how to access guidelines via the intranet or clinical guideline folders.

15.0 References

Clinical Genetics, Guy's and St Thomas' NHS Foundation Trust (2008) Tay Sachs Disease carrier screening in the Ashkenazi Jewish Population. Available at: www.guysandstthomas.nhs.uk/our-services/genetics/clinics/tay-sachs/overview.aspx

National Tay-Sachs and Allied Diseases Association. Standards for Carrier Screening [online] http://www.tay-sachs.org/screening_standards.php

Online Resources

National Tay-Sachs & Allied Diseases Association http://www.ntsad.org

Jnetics http://jnetics.org/

UK NSC Policy Database (accessed 10/2/12) Policy under review April 2012; policy accessed January 2015 (archived)

Your Genes, Your Health http://www.yourgenesyourhealth.org/tay/whatisit.htm

Climb National Information Centre for Metabolic Diseases http://www.climb.org.uk/

Appendix A

Mid Essex Hospital Services NHS Trust

Neonatal Alert Form

First Name	**Surname**	
NHS No	**Hospital No**	**Referral Date**
EDD	**Gestation**	**Consultant**

Background history & problem summary

Delivery Plans

Broomfield Hospital ☐ Not Decided ☐

Other Hospital _____

Neonatal Alert Form Criteria

Please use the neonatal alert form for the following conditions:

- Multiple pregnancy (higher order > 2 fetus)
- Hepatitis B positive mother
- HIV positive mother
- Previous baby with GBBS sepsis / meningitis
- Significant structural abnormalities diagnosedon ultrasound scan
- All cases that require referral to specialist units for treatment or advice
- Mothers with high antibody titres e.g. Anti-D, C and Kell
- Severe oligohydramnios / IUGR
- Abnormal dopplers
- Genetic / hereditary conditions in the immediate family that may affect the fetus
- Social e.g. drug abuse, alcohol abuse in this pregnancy
- Any other condition that will require paediatric input at birth

Postnatal Plan *(paediatric)*

Designation _____ Date _____

Print Name _____ Signature _____