



Are Their Genes Compatible?

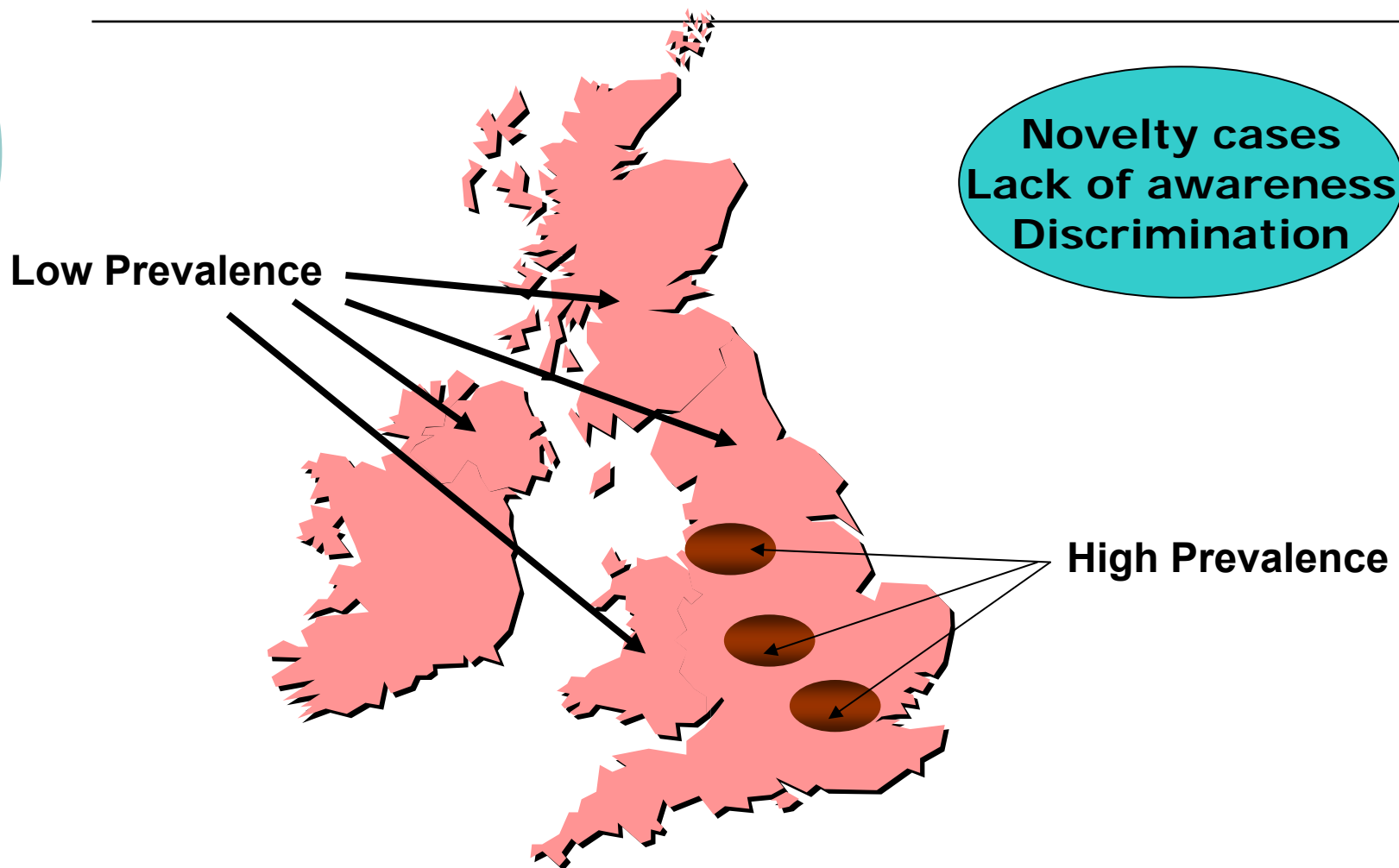
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DISTRIBUTION, EPIDEMIOLOGY, PREVALENCE OF SICKLE CELL & THALASSAEMIA

- **World Population** >4.7% (birth rate 20 per 10 000) ¼ million affected births / year
- **Distribution**-Malarial belt
- SC carrier frequency** : African- Caribbean's (1 in 10), West Africans (1 in 4), and Asians (1 in 50), the risk is still significant for Northern Greeks, Italians and Mediterranean's (1 in 100) (UK Sickle Cell Society, 2004)
- βThal carrier frequency** : Greek Cypriots (1 in 7), Mediterranean (1 in 10), Asian/Middle Eastern (1 in 10-30), Far Eastern (1 in 30), African Caribbean (1 in 50), N European (1 in 1000)
- **Europe & UK** >(12,500 SCD/ 800 β Thal). Prevalence in England ↑ by 45-60% in past 10 yrs (Streetly 2005)
- **Wales** 1.8% population (52,715) are at risk of carrying a sickle cell gene. 86% residing in S.Wales alone.

UK Sickle Cell & Thalassaemia Distribution



Inherited blood disorders are called ***haemoglobinopathies***

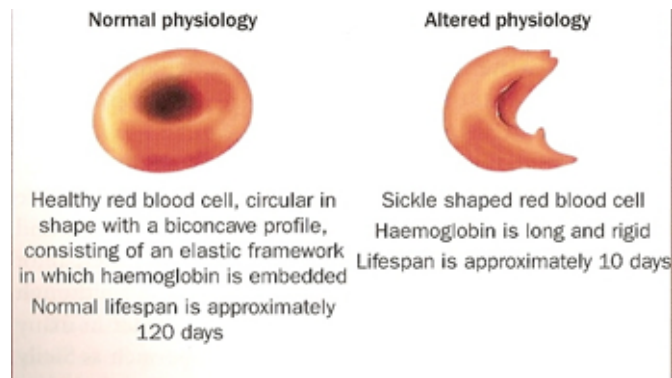
Inherited abnormalities of haemoglobin (Hb) production fall into 2 categories:

- **1. Production of an abnormal Hb**
- **2. Diminished production of Hb**

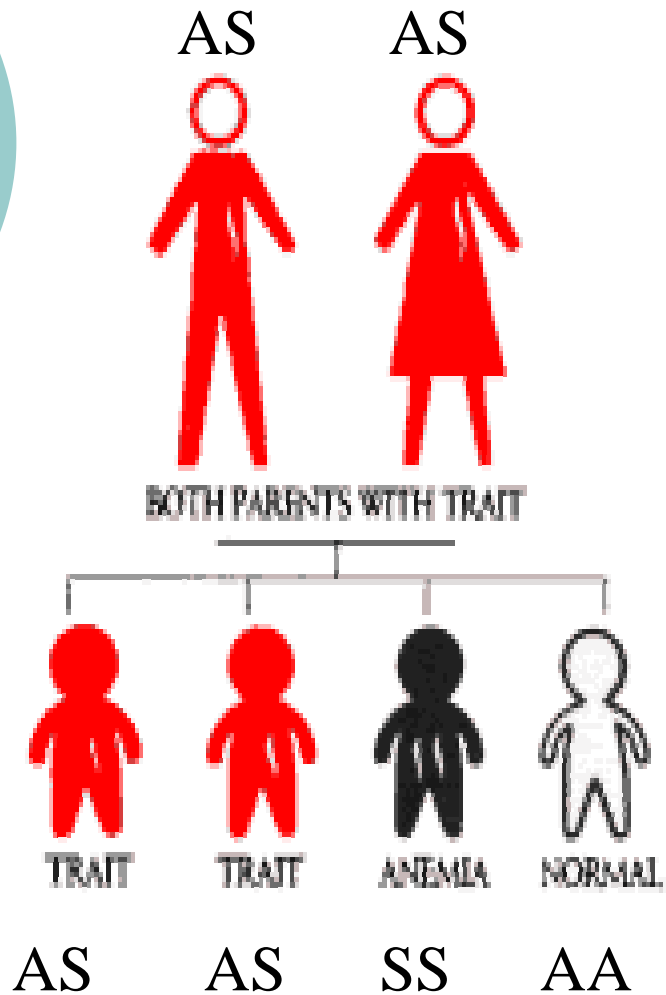
*The common feature is inheritance of at least one abnormal haemoglobin B chain gene designated **HbS***

What are they? Aetiology

- 'Normal' Haemoglobin HBAA
- Sickle Cell anaemia HBSS
- Sickle Cell disease HBSC
- Sickle Beta Thalassaemia HBS β
- Sickle cell trait HBAS



GENETICS/INHERITANCE



	A	S
A	AA	AS
S	AS	SS

Developments

- The NSC NHS Plan England
- Welsh Assembly Govt

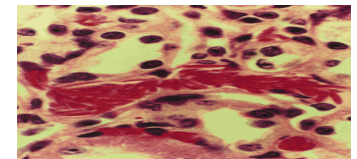
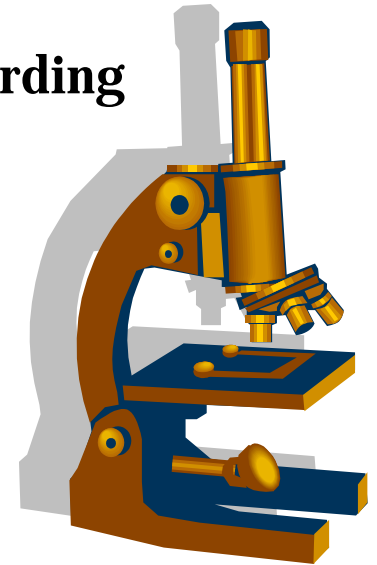
Wales Antenatal Screening Question?

Do

- **You, or your partner have a family history of sickle cell or thalassaemia?**
- **You have unexplained anaemia?**
- **You, your partner, anyone in your family, or anyone in your partner's family, no matter how many generations back, come from anywhere in the world apart from Northern Europe?**
- **You not know your family history – for example, you were adopted?**

Benefits of Antenatal Screening

- **Detects carriers**
- **Allows women at risk to make informed decisions regarding reproduction**
- **Offers couples the choice of prenatal diagnosis**
- **Provides genetic counselling**
- **Selective abortion**
- **If a child goes undiagnosed, they are 600-900 times more likely to die of an infection by the time they are six months old (Augustine 2004)**
- **Identifies high risk pregnancies**



Women who suffer from sickle cell anaemia during reproductive age are likely to...

- Bear less children
- Have higher risks of miscarriage
- 23% give birth prematurely
- Have more complications during delivery 38% suffer severe sickling crises
- Have more still births
- Have more frequent urinary infections

High risk pregnancies!

(Maternal mortality 3 X that of those with HBAA)



Sickle Cell Disorder & Special Care Required in Pregnancy



▪ **Fetal Problems**

- Spontaneous abortion
- Intra uterine growth restriction/retardation
- Stillbirth
- Neonatal death
- Barts hydrops fetalis

▪ **Maternal Problems**

- Bone pain
- Haemolysis
- Acute chest syndrome
- Pre-eclampsia
- Urinary tract infection
- Mortality/morbidity

Eboh and van den Akker (1994)

RESEARCH STRANDS

Novelty cases
Lack of awareness
Discrimination

- Weatherall & Clegg identified in 1981 that serious inherited blood disorders were back then one of the most important health issues facing society and this still applies to today.
- There is no universal cure at present for sickle cell disorders; therefore appropriate information is essential in order for individuals to then go on to make well informed reproductive choices (Asgharian, Anie and Berger 2003).
- It has been recommended that further research should be commissioned to examine the role of the midwife in haemoglobinopathy screening and such research should place the views of those living with haemoglobinopathies and their carers at its core (Sutton et al 2003).
- DoH (2003) drive to improve genetics in the nursing & midwifery curriculum

My Studies

○ ARE THEIR GENES COMPATIBLE?-

1 yr research training fellowship from Health Professions Wales looking into midwifery antenatal screening knowledge and attitudes of sickle cell and thalassaemia based on the work of Simon Dyson in England in the mid 1990's.

○ LIVING AND WORKING WITH SICKLE CELL AND THALASSAEMIA (S.C.A.T) –

3 yr qualitative study aiming to promote patient empowerment, awareness, quality of client care & genetic competence.
(best practice visits UK & overseas, focus groups, oral histories)

ARE THEIR GENES COMPATIBLE?

- **Study Aims**
- **Hypothesis**
- **Literature Review**
- **Study Design**- method, pilot, samples
- **Ethical Considerations and procedure**
- **Data Analysis**
- **Results**- questionnaire breakdown, ethnic groups, inheritance & trait, prevalence, education & training
- **Study Limitations**
- **Discussion**
- **Conclusion**
- **Feedback**

Discussion

1. How or what is the best way to train midwives (or any other health care professional in low prevalence areas) to ensure that they retain adequate knowledge on conditions that they don't often come across in practice?

2. Which option do you think would be the most effective (investment of resources) in low prevalence areas?

- a) *more timetable space be allocated to sickle cell and thalassaemia training within pre-registration midwifery programmes?*
- b) *more training emphasising the importance of general screening for midwives instead?*
- c) *make post registration midwifery sickle cell and thalassaemia training updates mandatory for qualified staff?*
- d) *or instead of more training for midwives, invest in more sickle cell and thalassaemia specialist nurses to support them?*
- e) *Continue with current input?*



Conclusion

- An understanding of sickle cell and thalassaemia disorders is crucial if midwifery and antenatal care is to meet the needs of minority ethnic groups and if genetic competence in the midwifery profession is to be improved

Genome Policy Unit et al 2003

My Future work:

Publications, presentations, further research: Training review, An all Wales study with focus groups, future funding ++£, MPhil, Advisory network

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Further Reading:

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