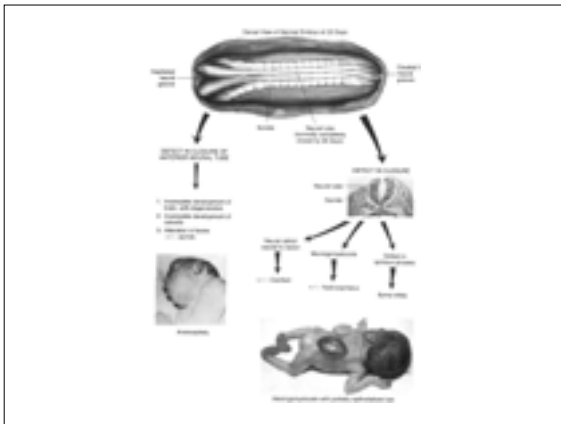


Neural Tube Defect



Definition

- Failure of closure of the neural tube.
- Can occur at various levels.
- Can have widely varying severity.
- Most severe is anencephaly.
- Least severe is spina bifida occulta.
- Most clinically challenging is lumbo-sacral myelomeningocele.

Basic Epidemiology

- Varies widely depending on ethnicity and socio-economic status
- ~1% in some places in UK
- 1:1000 in US
- High in Mexican/Amer-Indian populations.
- Virtually all cases are sporadic, isolated malformations.

Folate

- Low serum folate correlated with NTDs
- Folate supplementation with 400-800 ug per day reduces incidence by ~75%.
- Clearly, a sub-set of NTDs are not “folate dependent”.

MHTFR

- 5,10-methylenetetrahydrofolate reductase is in a pathway for recycling folate
- Catalyses a critical step in methionine biosynthesis.
- Several common variants of this enzyme exist.
- Between 5 and 15% of caucasians are homozygous for the variant form.

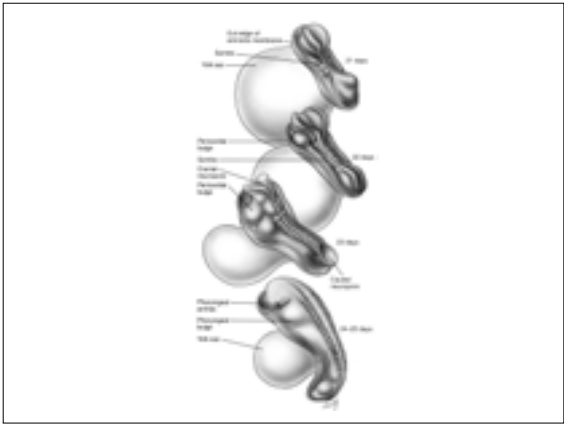
- Homozygotes are thought to be at risk for NTD and higher than normal folate supplementation has been recommended.

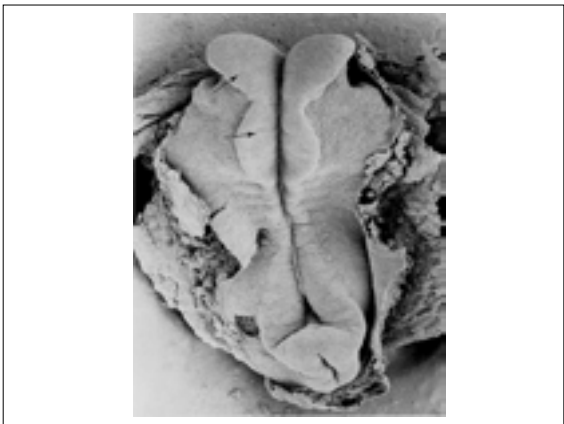
Prenatal Detection

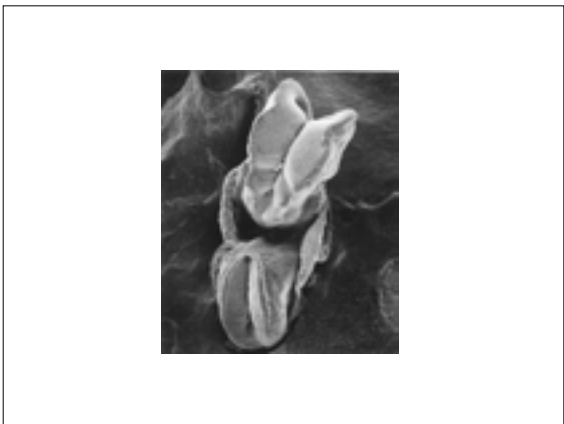
- In developed countries, prenatal detection of severe NTD should approach 100%.
- Maternal serum AFP screening is efficient as a screening tool.
- Sonography is efficient at both screening and diagnosis.

Counseling

- Outcome depends a lot on level.
- Lower lesions may result in relatively mild deficits.
- Another issue is the related hydrocephalus, cerebellar malformation and long-term cognitive dysfunction.











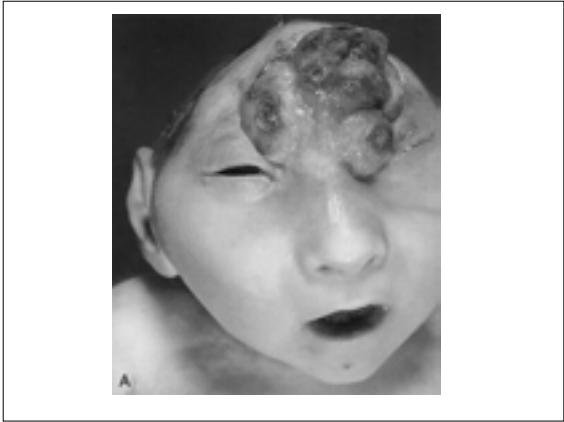
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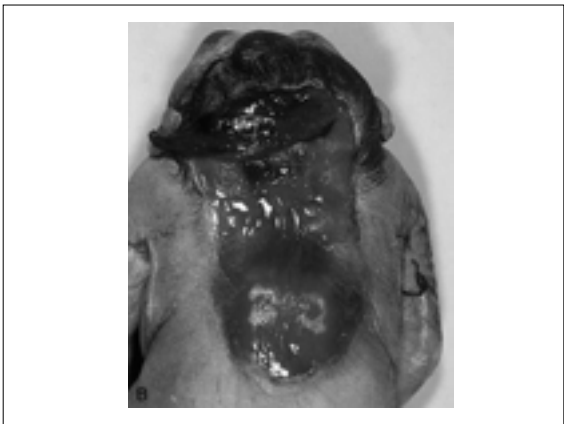
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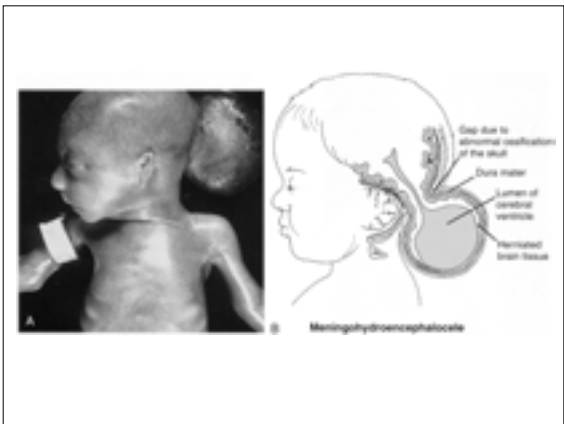


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Summary

- Generally not genetic
- Recurrence risk is elevated over background but still low
- Prenatal detection is excellent but some cases can present a big counseling challenge.
