Normal neonatal Lymphocyte counts on day one of life and incidence of lymphopenia – could FBC be used to screen for Severe Combined Immune Deficiency?

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Background

- Severe Combined Immune Deficiency (SCID) is a term for a group of disorders arising from disturbed development of functional B and T cells.
- It is more frequent in the minority population of Irish Travellers (1 in 2000 vs. 1 in 58,000 in the general population)
- Gold standard for diagnosis is TREC assay (T-cell receptor excision circle) but this is expensive and not used for screening in Ireland.
- Lymphopenia (lymphocyte count <1.5 x 109/L) is a potential incidental presentation of SCID.
- We investigated the possibility of using a full blood count (FBC) as a screening test.
- We determined the incidence of lymphopenia on day 1 of life in term neonates who have had an FBC as part of their newborn care.

Methods

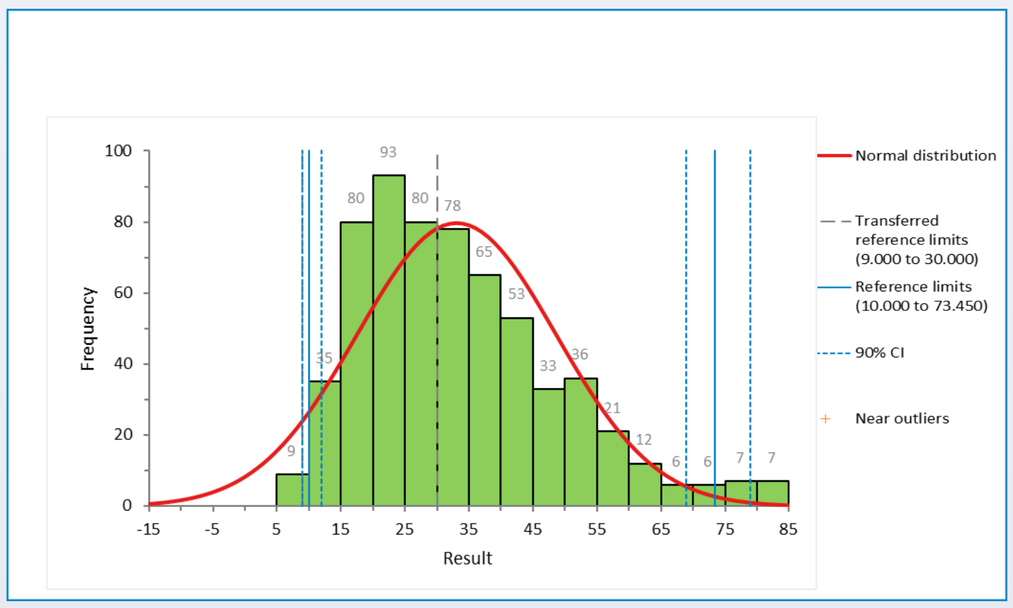
- We retrospectively examined the results of all full blood count (FBC) specimens taken in a six-month period in 2015.
- Preterm infants, babies with positive blood cultures and repeat samples were excluded
- The earliest sample taken on each baby was included for analysis.
- We defined 1.5 x 10^9/L as significant lymphopenia in order to determine how often this finding would occur on routine sampling in neonates.

Results

There were 1239 relevant blood samples taken during the period of interest. Of these, only 10 specimens had an absolute lymphocyte count less than 1.5 x 10^9/L, giving an incidence of 0.8% of this finding. If a more generous cut-off point of ≤ 2.0 x 10^9/L were used, 29 infants in our study population would have needed further investigation for SCID.

Conclusions

- Only 0.8% of normal, unaffected newborns have a lymphocyte count <1.5 x 10^9/L
- Day 1 FBC would be a cheap, pragmatic first line screening tool for children at high risk of SCID with a low likelihood of false positives.
- Early diagnosis is vital in these infants to prevent end organ damage and ensure life saving bone marrow transplantation is carried out as early as possible.



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