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## Vsodržavno testiranje novorojenčkov za hudo T-celično limfopenijo

**Izhodišče:** Presejalno testiranje krvi novorojenčkov (NBS), s katerim bi lahko zgodaj ugotovili hudo pomanjkanje T-celic in tako zmanjšali umrljivost novorojenčkov.

**Cilji:** Ugotoviti, ali določanje količine izrezanih krožnih delov T-celičnih receptorjev (TREC) s kvantitativno verižno reakcijo s polimerazo v realnem času na DNA, pridobljene iz posušenih otčokov krvi na kartici NBS, lahko odkrije novorojenčka s T-celično limfopenijo v vse državnem presejalnem programu.

**Oblika, nastavitve in udeleženci:** Med 1. januarjem in 31. decembrom 2008 je Wisconsin State Laboratory of Hygiene pregledal vso kri novorojenčkov, rojenih v državi Wisconsin, za T-celično limfopenijo z določanjem vrednosti TREC v 3,2 mm velikih otčokih krvi (približno 3  $\mu$  celotne krvi) na kartici NBS. S pretočno citometrijo so nato pri donošenih in nedonošenih novorojenčkih z vrednostjo TREC manj kot 25/ $\mu$ l določili število T-celic. Stari so morali biti najmanj 37 gestacijskih tednov. Novorojenčke s T-celično limfopenijo je nato pregledal klinični imunolog.

**Glavni izsledki:** Število novorojenčkov z vrednostjo TREC manj kot 25 / $\mu$ l in s pretočno citometrijo potrjeno T-celično limfopenijo.

**Rezultati:** Natanko 71 000 novorojenčkov je bilo testiranih z določanjem vrednosti TREC. Sedemnajst novorojenčkov, starih vsaj 37 gestacijskih tednov, je imelo vsaj eno spremenjeno vrednost TREC (TREC < 25/ $\mu$ l), in pri 11 so nato opravili še pretočno citometrijo ter določili število T-celic. 8 novorojenčkov je imelo T-celično limfopenijo. Vzroki za T-celično limfopenijo so bili DiGeorgov sindrom (n = 2), idiopatska T-celična limfopenija (n = 2), ekstravaskularna na ekstrasvazacija limfocitov (n = 3) in mutacija *Rac2* (n = 1). Novorojenčki z mutacijo *Rac2* so uspešno prestali transplantacijo popkovnične krvi.

**Zaključek:** V državnem presejalnem testiranju so z določanjem vrednosti TREC na karticah NBS uspešno odkrili novorojenčke s T-celično limfopenijo.

John M. Routes, William J. Grossman et al. Statewide Newborn Screening for Severe T-Cell Lymphopenia. *JAMA.* 2009;302(22):2465-2470.

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