

NORMAL T CELL RECEPTOR EXCISION CIRCLES LEVEL IN HEALTHY FULLTERM AND PRETERM NEONATES AT ALEXANDRIA UNIVERSITY CHILDREN'S HOSPITAL

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Introduction

Severe combined immunodeficiency disease (SCID) is one of the most severe forms of PIDS, the incidence is found to be 1 in 58,000 live births in the United States with higher incidence of autosomal recessive (AR) SCID in communities in which consanguineous marriage is more frequent.

SCID is characterized by absence of T lymphocytes, and/or deficiencies of B cells, and may also affect natural killer (NK) cells. Infants with SCID appear normal at birth, however, they develop severe infections in the first months of life.

Without curative treatment, in the form of allogeneic hematopoietic stem cell transplantation (HSCT) or in some specific forms of SCID, gene therapy, affected infants die within the first two years of life.

Newborn screening for SCID is particularly important in Middle Eastern countries due to the relatively higher incidence of consanguinity.

T-cell receptor excision circles (TRECs) are small circular DNA fragments formed during development of T-lymphocytes in the thymus and do not multiply during cell division. There fore, TREC is a biomarker of naïve T cells.

Aim of the work

The aim of this study was to determine the normal TRECs level in apparently healthy full-term and preterm neonates at Alexandria University Children's Hospital (AUCH).

PATIENTS AND METHODS

Patients: The study included a sample size of 191 apparently healthy Neonates (95 preterm and 96 full term) delivered in Alexandria University Children's Hospital in addition to Eleven (11) confirmed severe combined immunodeficiency disease (SCID) patients as controls to validate the TRECs measurements .

Methods:

Data collection: History taking. Routine clinical examination of the newborn.

Sample collection: Dried blood spot (DBS). DNA extraction. Real time quantitative polymerase chain reaction (PCR) on extracted DNA ,Follow up complete blood picture was done to all included newborns . Determination of the cutoff point for normal full term and preterm neonates.

Results

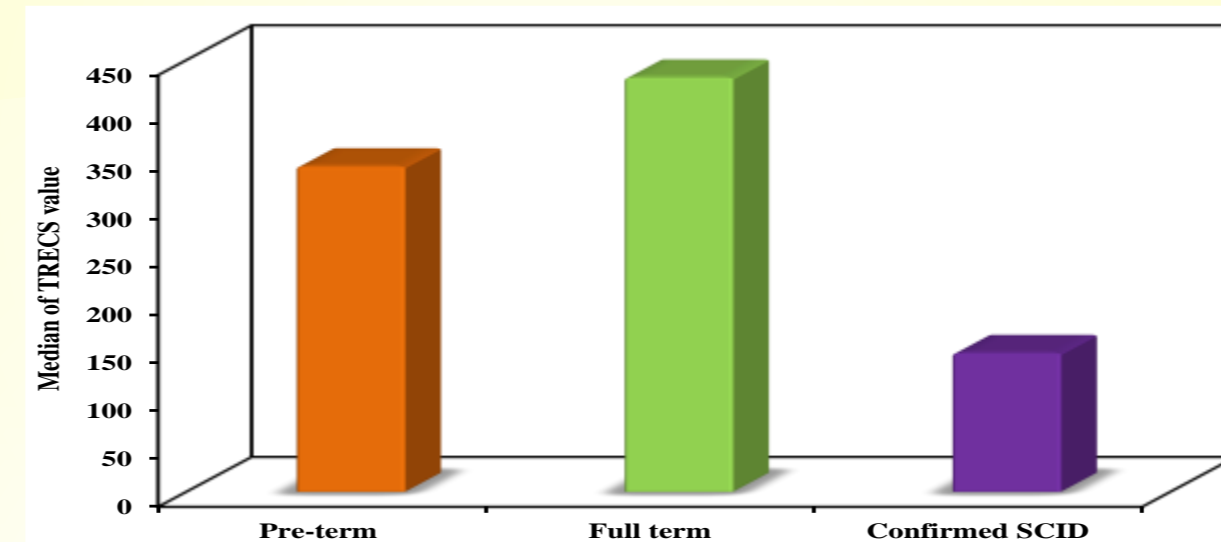


Fig 1: Comparison between the three studied groups according to median TRECS value

Comparison between the three studied groups according to median TRECs value

The median TRECs value is significantly higher in the full-term neonates group comparing to both that preterm and confirmed SCID group, the median TRECs value is higher in preterm neonates group than confirmed SCID group but the difference between them is non-significant.

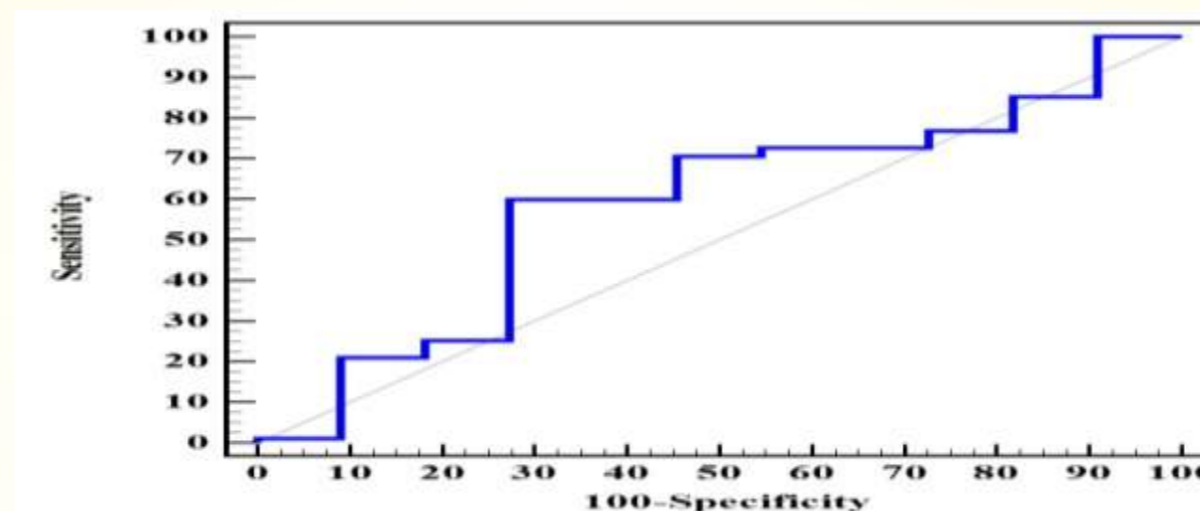


Fig 2: Diagnostic performance for TRECs to discriminate between Pre-term and confirmed SCID patients.

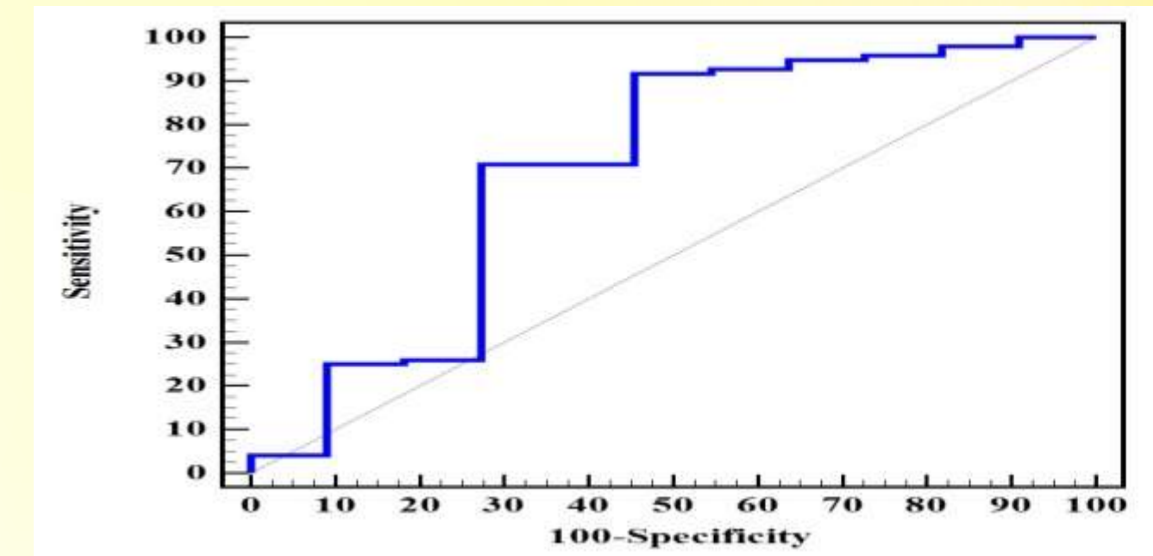


Fig 3: Diagnostic performance for TRECs to discriminate between Full term and confirmed SCID patients.

Conclusion

- Normal cut off point of TRECs level among full-term newborns was determined according to our laboratory.
- There is no defined cut off point of TRECs level in preterm newborns at birth so it must be retested again when the newborn reaches the week 37 of corrected gestational age if SCID is suspected.
- The level of TRECs can be affected by several factors as gestational age, maternal illness and birth weight.