Quality of Life for Immunodeficiencies via Newborn Screening

Joie Davis APRN, BC, APNG

National Institutes of Health, National Human Genome Research Institute

International Patients Organization for Primary Immunodeficiencies

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Justification for Newborn Screening

- SCID is asymptomatic at birth, but becomes fatal without intervention
- Effective treatment is available (BMT)
- Early intervention improves outcome
- BMT before severe infection->better survival at lower cost compared to those receiving delayed treatment
- Transplantation in 1st 3 mos = 95% survival rate (Buckley, US) vs. 70% survival rate after 3 mos.

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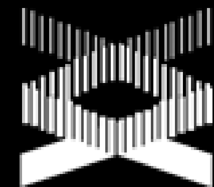

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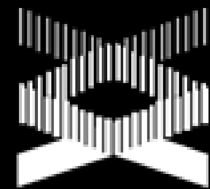

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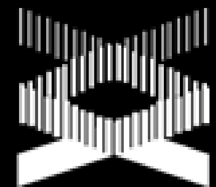

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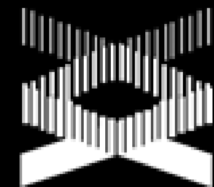
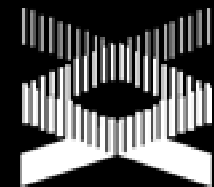


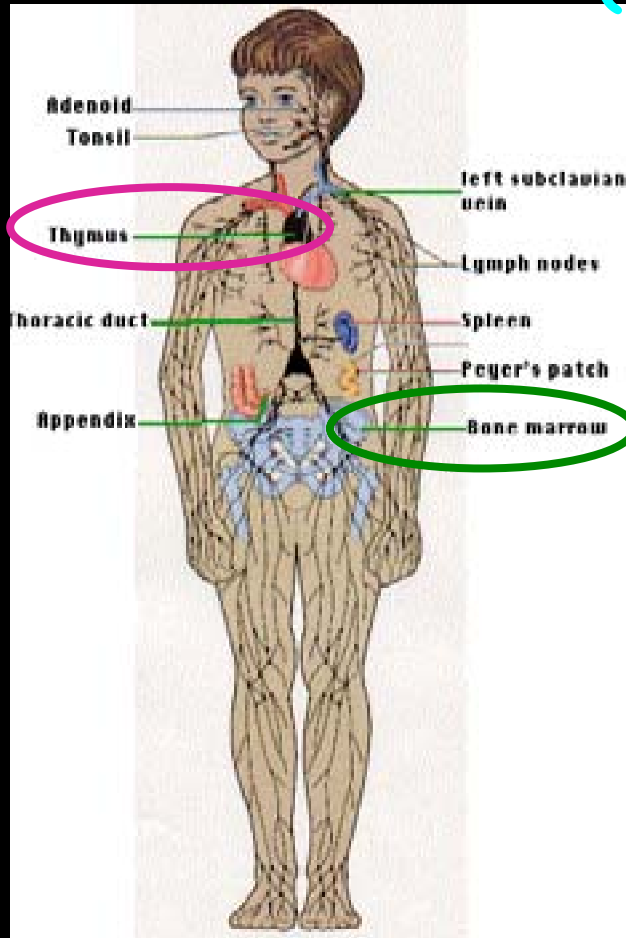
Newborn Screening Development Process

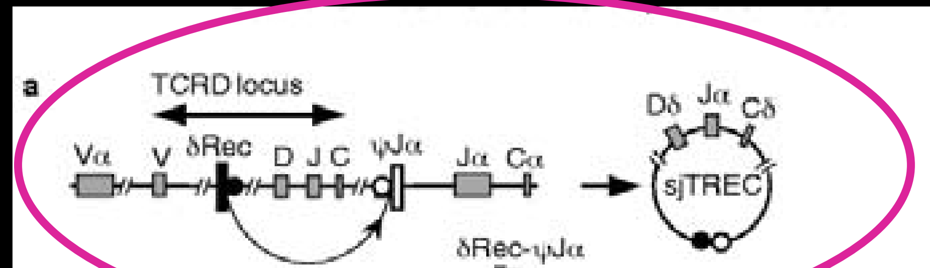
T Cell receptor excision circles (TRECS)

Family experiences



Screening test measures T cell Receptor Excision Circles (TRECs)





Is there an association?

Early Diagnosis

Favorable Outcome

- Survival
- Fewer days in hospital
- Fewer infection related doctor visits

Family Interviews

Posted invitation on *http://www.scid.net/* with the help of Barbara Ballard

Enrolled parents of SCID child born between Jan 1, 2000 and Jan 1 2005

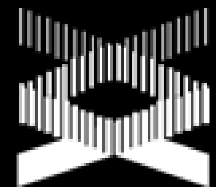
How many visits to the doctor for infections? (out patient visits & hospital stays)

Age at which diagnosis of SCID was made?

Age when transplant was received?

Course post transplant (out patient visits, and hospital days)

Did the child survive?



1. CIRCLE THE AGE AT WHICH SCID WAS DIAGNOSED. 005P

2. Under each 1-month age interval, mark the box most closely matching the medical care your child received.

3. Where indicated, add under the X a letter (A, B, or C) that most closely represents the level of care or nights in the hospital that your child required during the month indicated.

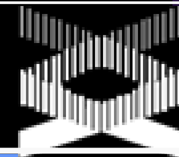
4. Put additional X's in boxes corresponding to ages when your child received IVIG or PEG-ADA.

Your child's condition	Your child's age in months																	
	0-1 mo	1-2 mo	2-3 mo	3-4 mo	4-5 mo	5-6 mo	6-7 mo	7-8 mo	8-9 mo	9-10 mo	10-11 mo	11-12 mo	12-13 mo	13-14 mo	14-15 mo	15-16 mo	16-17 mo	17-18 mo
Presymptomatic. No SCID-related medical visits (for infection, poor weight gain, rash). Visits for well-baby care, trauma, etc. don't count.																		
Outpatient. Doctor visits, but no SCID-related hospital nights (pre-BMT)				X	X	X												
A = 1-2 visits to clinic or ER during the month for problems that could be SCID-related B = 3 or more visits				B	B	B												
Any hospitalization(s) related to SCID (pre-BMT)							X	X										
A = 1 - 3 nights in hospital B = 4 -14 nights C = more than 14 nights							C	C										
BMT									X									
Post-BMT, with some nights in the hospital										X	X	X						
A = 1 - 7 nights in hospital B = 8 - 14 C = more than 14										C	C	C						
Post-BMT completely outpatient. No nights in hospital													X	X	X	X	X	X
A = no SCID medical visits during the month B = 1-2 doctor or ER visits C = 3 or more visits													C	C	B	B	B	B
Receiving IVIG								X	X	X	X	X	X	X	X	X	X	X
Receiving PEG-ADA																		
Deceased																		

Outcome vs. Age at Diagnosis

Based on family interviews for 40 SCID cases, including X-linked, JAK3, ADA, IL7R, RAG, unknown

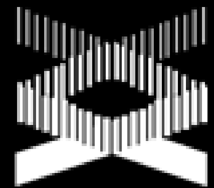
	Known affected relative (20% of the 40 cases)	**Negative family history (80% of the 40 cases)**
Number of infants	**7**	**33**
Mean age of SCID diagnosis	***2 mo***	***9 mo (1 at autopsy)***
Received BMT or ADA treatment (*mean age*)	***7* (mean age = 3 mo)*** *all had BMT	***25* (mean age = 9.5 mo)*** *8 died without BMT or ADA
Survival after BMT or ADA treatment	**7/7**	**14**/25 (56%)** **2 died with ADA, 9 after BMT
Overall survival to 2.5 years or more	**100%**	**42%**



Common presentation before diagnosis

Respiratory

- persistent cough
- rapid breathing
- fussy, difficult to console, difficulty sleeping (? Air hunger)
- wheezing
- low percentage oxygen saturation
- pneumonia(s) ->PCP->ventilator (CF testing often done)

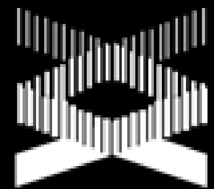


Common presentation before diagnosis

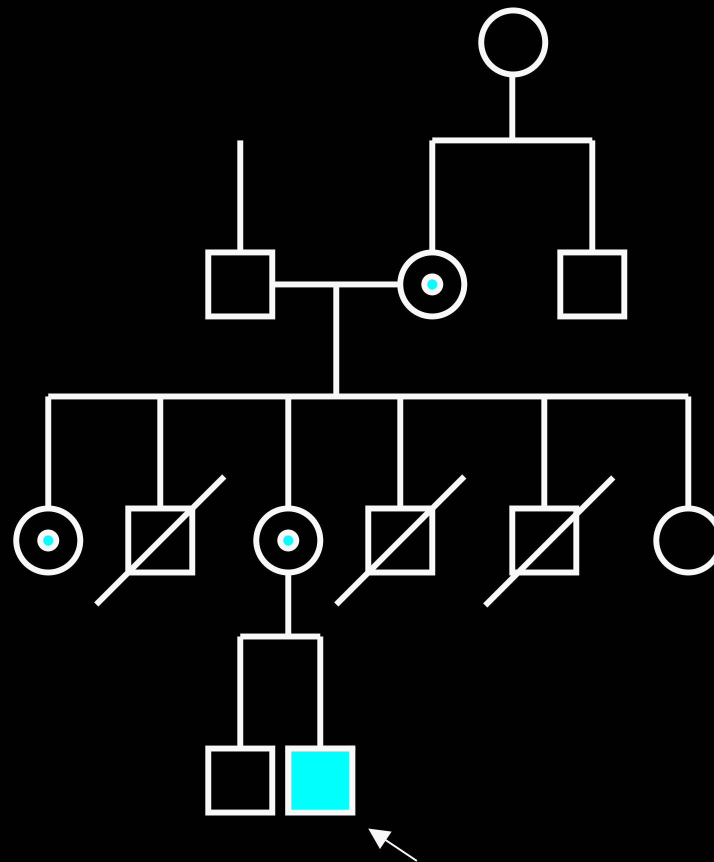
Other persistent infections

- oral & diaper monilia, present at 1 mo
- Eye infections between 6 wks - 3 mo
- Ear infections
- Skin rashes at 2 mos

Failure to thrive

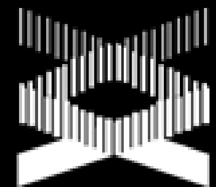


X-Linked Severe Combined Immune Deficiency (XSCID)



Lessons Hoped For

Newborn Screening can identify Primary Immune Deficiencies earlier, allow earlier intervention and hopefully decrease mortality.

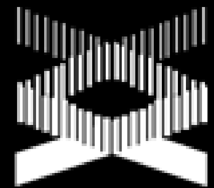


Lessons learned

NOTICE TO THE HEALTH CARE TEAM!

Before all else fails

LISTEN to the Mom



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Rebecca Buckley Duke

Made possible by
SCID families who told us their stories