



Response Evaluation In Neurofibromatosis Schwannomatosis INTERNATIONAL COLLABORATION

- If sharing any data or information from these slides generated by the REiNS International Collaboration, please acknowledge the authors, group chairs, and specific working group.
- If using any information presented with a citation, please reference the primary source.

Perspectives of Adolescents with NF1 and Cutaneous Neurofibromas

Ashley Cannon, PhD, MS, CGC
Cutaneous Neurofibroma Working Group
REiNS Summer Meeting 2023



Response Evaluation In Neurofibromatosis Schwannomatosis
INTERNATIONAL COLLABORATION

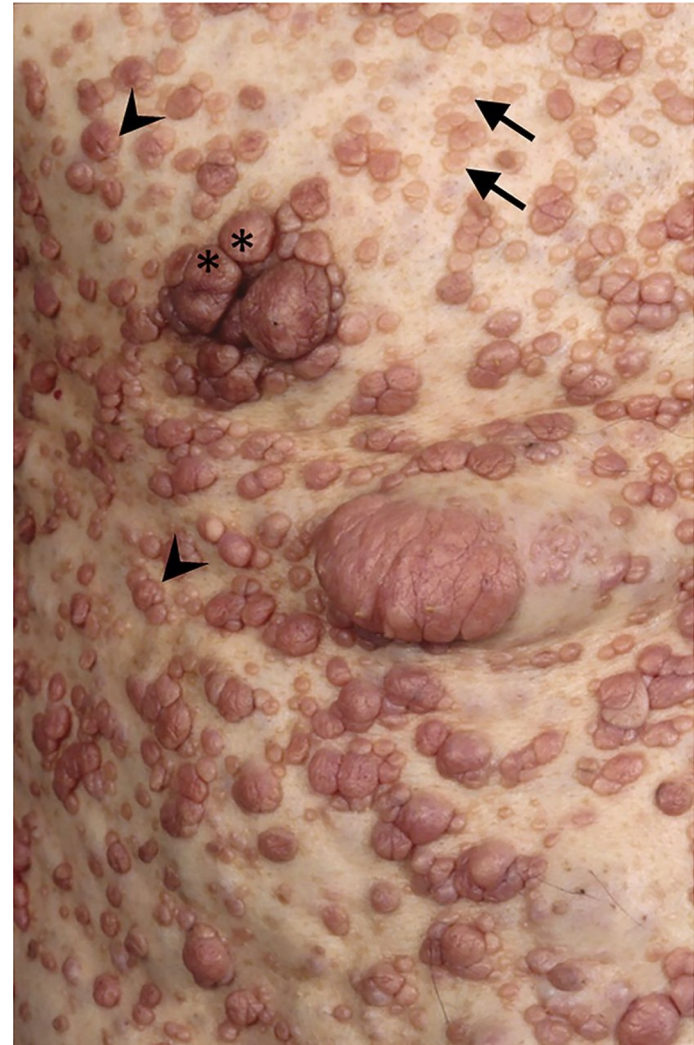
Cutaneous Neurofibromas

Benign nodules on the skin

Develop in 99% of individuals with NF1

Adversely impact quality of life

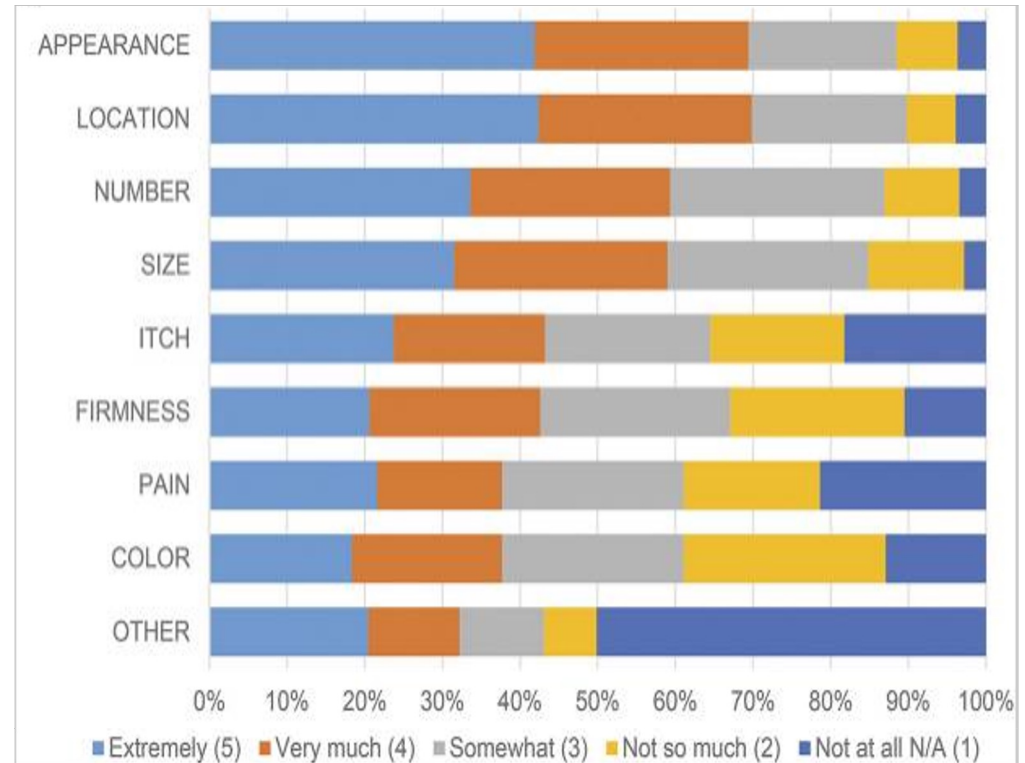
Appear in adolescents and increase throughout adulthood



Adult Perspectives on cNFs

Appearance, number, and location most bothersome features

>65% willing to try experimental treatments



KOSELUGO[®] (selumetinib) is the FIRST and ONLY FDA-approved prescription medicine that is used to treat children 2 years of age and older with neurofibromatosis type 1 (NF1) who have plexiform neurofibromas that cannot be completely removed by surgery. It is not known if Koselugo is safe and effective in children under 2 years of age.

**LESS
TUMOR SIZE**



**MORE
ME**

– Cooper, age 8, actual patient

[WATCH COOPER'S STORY](#)

Koselugo is proven to reduce the size of plexiform neurofibromas (PN).*

[Learn more](#)

*66% or 33 of 50 patients in the clinical study saw a 20% or more decrease in the size of their plexiform neurofibromas, confirmed by 3D magnetic resonance imaging scan.

Adolescent cNF Survey

Paired adolescent and caregiver surveys

Survey link sent to CTF NF Registry participants

Eligibility criteria included:

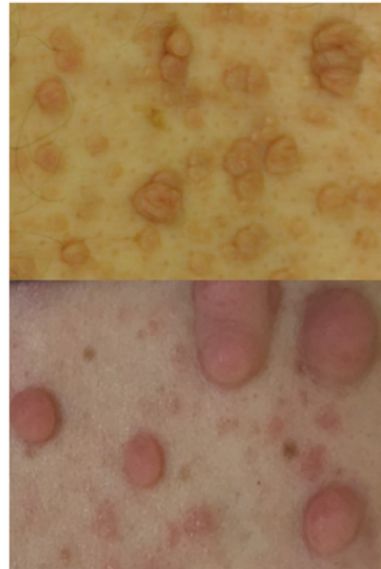
- self-reported NF1 diagnosis
- adolescent child 12-17 years
- ≥ 1 cNF
- ability to read English



1.c7. Adolescent Assent

1.1.c7. We are asking you to be in a research study. This research study will ask you questions about the bumps on your skin caused by Neurofibromatosis type 1 (NF1). This study will help doctors understand more about your NF1 and your thoughts about treatment. The information you share will be kept safe. We ask that you answer all the questions honestly. There are no right or wrong answers. You can choose not to answer every question. You can choose not to be in this study.

Flat or raised bumps on the skin in people with Neurofibromatosis type 1 (NF1) are called cutaneous neurofibromas. We will call them cNFs from now on. Here are some pictures of cNFs:



1.2.c7. Do you agree to take part in this research study?

Yes No

Survey Respondent Characteristics

ADOLESCENTS = 28

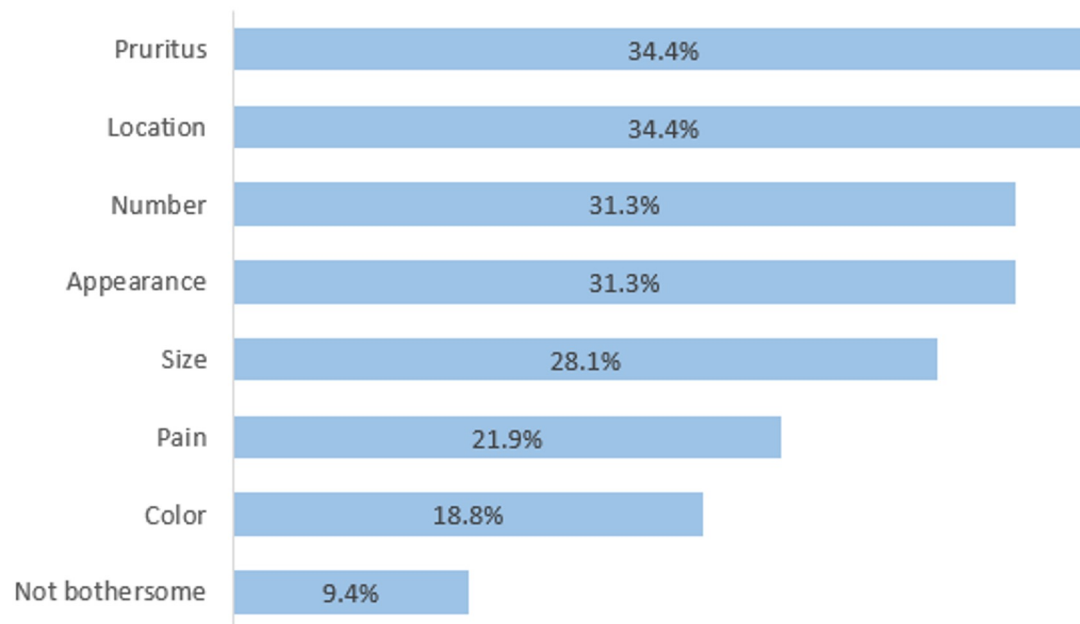
- Mean age: 14.2 years
- 92% were the first person in the family diagnosed with NF1
- 77% reported having 1-19 cNFs

CAREGIVERS = 32

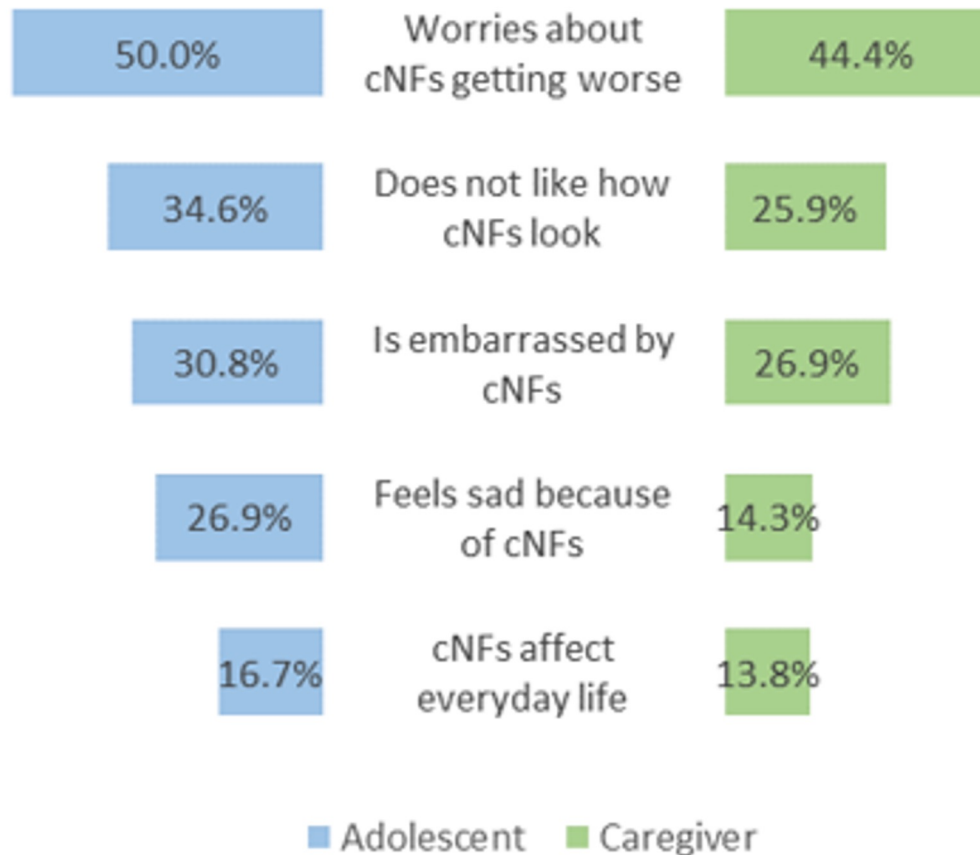
- Mean age: 46.3 years
- 94% female
- 75% White
- 84% live in the United States



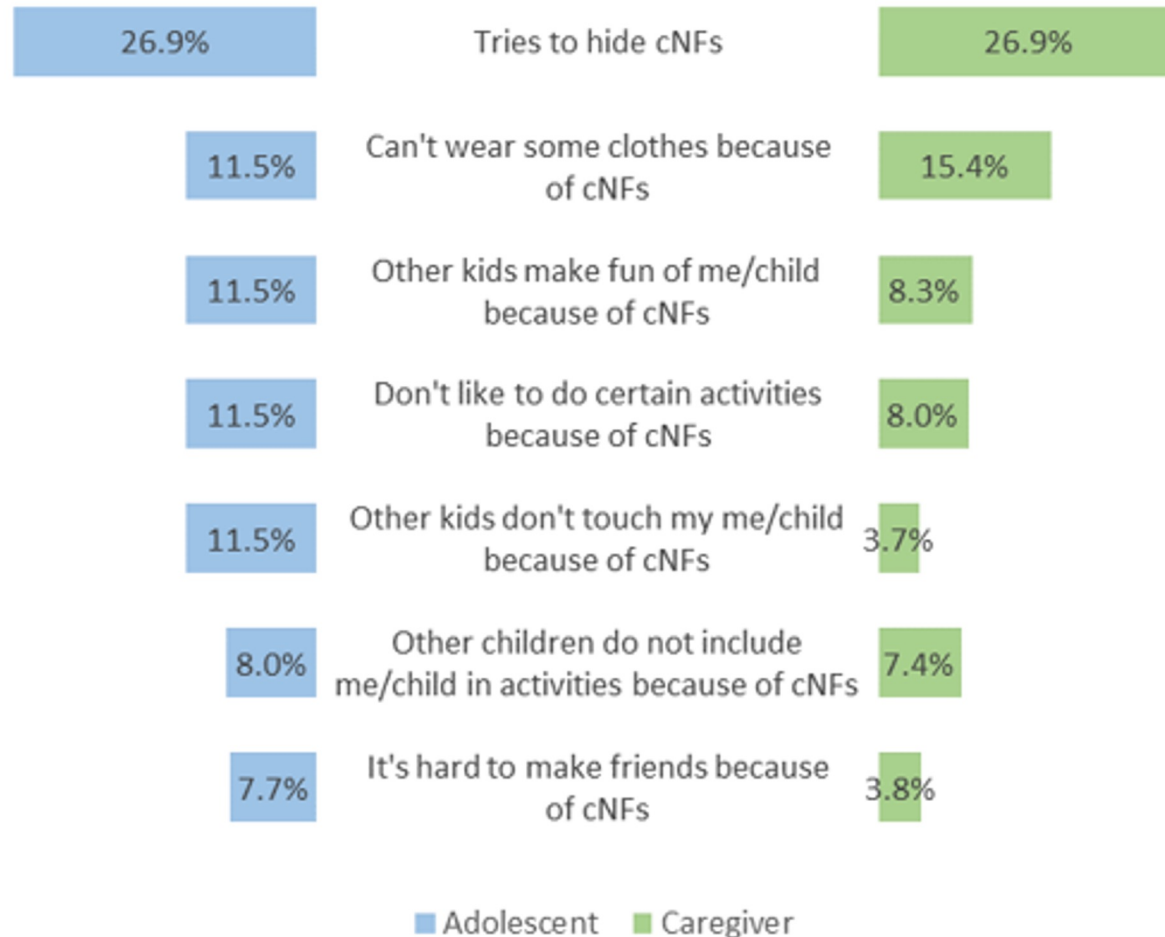
Most Bothersome cNF Features



Impact of cNFs on Adolescent Feelings



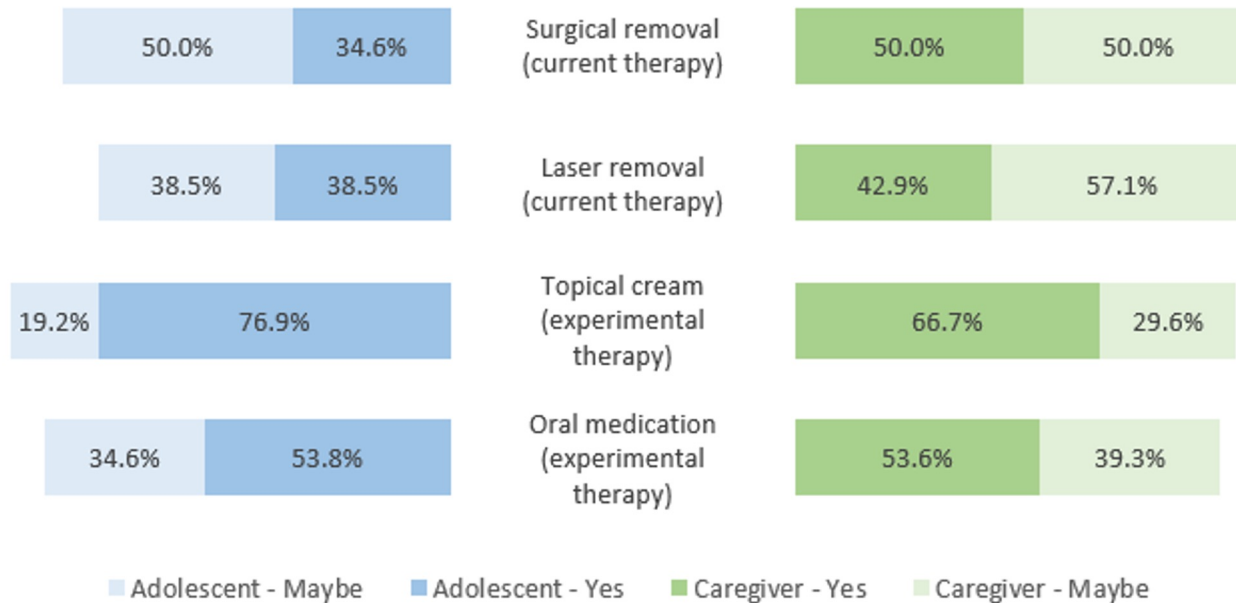
Impact of cNFs on Social Interaction



How Do cNFs Affect Everyday Life?

Major Themes	Adolescent Reported	Caregiver Perspective	Representative Adolescent Quote
Physical	6 (54.5%)	5 (41.7%)	If they get bumped, they hurt like crazy.
Emotional	4 (36.4%)	5 (41.7%)	I worry about them.
Social	1 (9.1%)	2 (16.7%)	I'm embarrassed for others to see me.

Willingness to Treat cNFs with Current and Experimental Therapies



Preferred Treatment Initiation

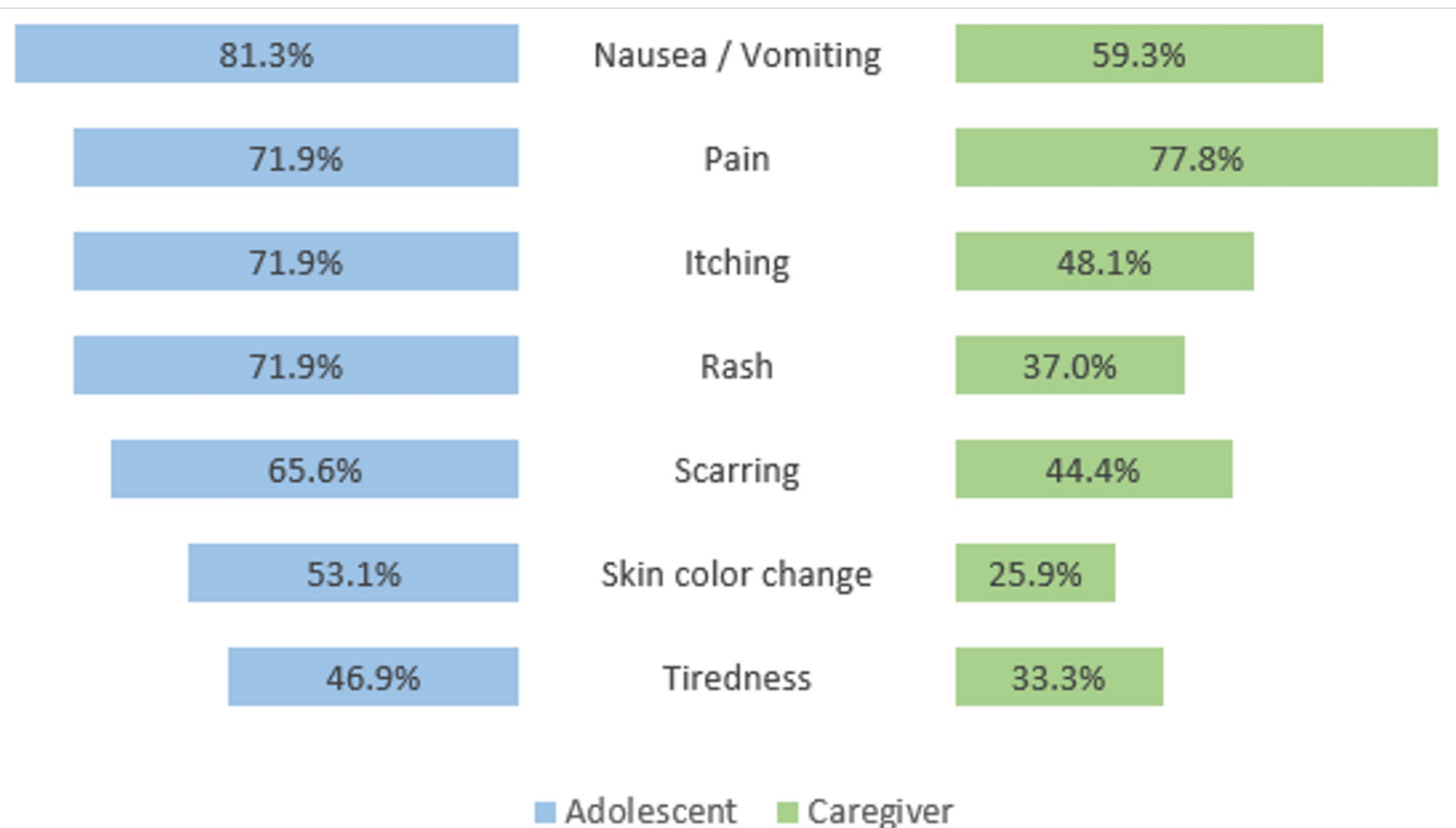
	Adolescent	Caregiver
When cNFs Become Bothersome	40%	54%
When cNFs Appear	28%	18%
Before cNFs Develop	11%	8%

Preferred Treatment Length

	Adolescent	Caregiver
Many Years	36%	57%
More than a Year	20%	0%
One Year	8%	18%
Six Months	16%	11%
One Month	20%	11%
One Week	0%	4%



cNF Treatment Side-effects Adolescents and Caregivers are Not Willing to Risk



Conclusions

cNFs can negatively impact the social and emotional wellbeing of adolescents with NF1

Adolescents and caregivers are willing to try long-term experimental treatment

Both adolescents and caregivers reported low tolerability for some side effects

Overall adolescent and caregiver views seem similar; however, agreement among adolescent and caregiver pairs was relatively low

Limitations: relatively small and homogenous study cohort



Conclusions

cNFs can negatively impact the social and emotional wellbeing of adolescents with NF1

Adolescents and caregivers are willing to try long-term experimental treatment

Both adolescents and caregivers reported low tolerability for some side effects

Overall adolescent and caregiver views seem similar; however, agreement among adolescent and caregiver pairs was relatively low

Limitations: relatively small and homogenous study cohort



Acknowledgements

REiNS cNF Working Group

- Kavita Y Sarin
- Andrea K Petersen
- Dominique C Pichard
- Pamela L Wolters
- Gregg Erickson
- Andrés J Lessing
- Claas Röhl
- Tena Rosser
- Brigitte C Widemann
- Jaishri O Blakeley
- Scott R Plotkin

Children's Tumor Foundation

UAB Biostatistics

- Peng Li, PhD

Funding

- Francis S. Collins Award (NTAP)

