Childhood Cancer Data Initiative Webinar Series

CCDI Pediatric, Adolescent, and Young Adult Rare Cancer Study

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NIH NATIONAL CANCER INSTITUTE

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Today's Speaker

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Rare Cancers: Definition

Definition: Fewer than 15 per 100,000 people/year in U.S.

- All pediatric cancers are rare

Very rare pediatric cancer:

- Fewer than 2 cases per million per year (11% of all pediatric cancers)
- Tumors not considered in clinical trials
- Molecular characterization of rare cancers creates even smaller groups:
  - Rhabdomyosarcoma:
    - Fusion positive (PAX3-FOXO1, PAX7-FOXO1)
    - Fusion negative:
      - TP53 mutation
      - MYOD1 mutation
      - Multiple other mutations

Ferrari A. et al., European Journal of Cancer, 2019

Rare Cancers: Limitations and Ongoing Efforts

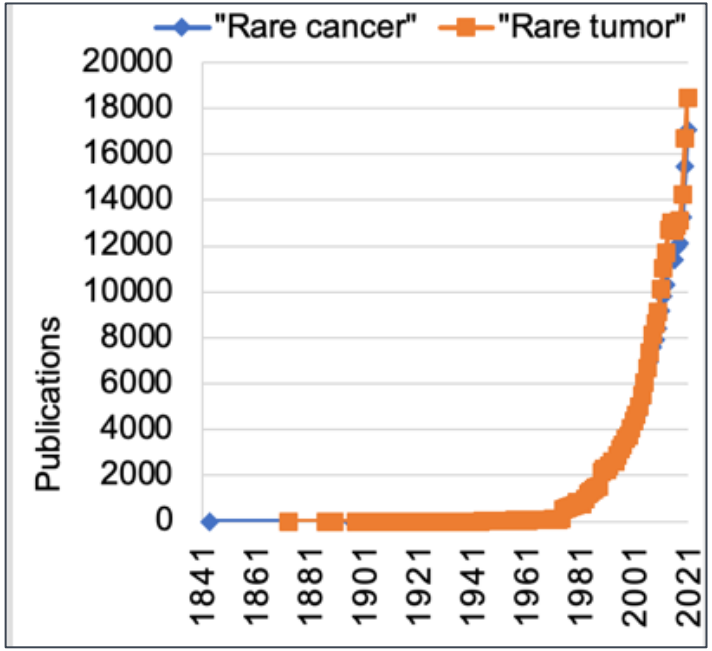
Limitations:

- Focus on few cancers
- Siloed
- Insufficient patient numbers for most cancers
- Data collection limited not standardized/structured

Successful Pediatric Efforts:

- PPB/DICER1
- EXPeRT/Partner

Landscape Analysis: 76 Rare Tumor Programs



Rare tumor/cancer publications

Ferrari A. et al., European Journal of Cancer, 2019 *Vivelo C. et al., The Oncologist 2023*

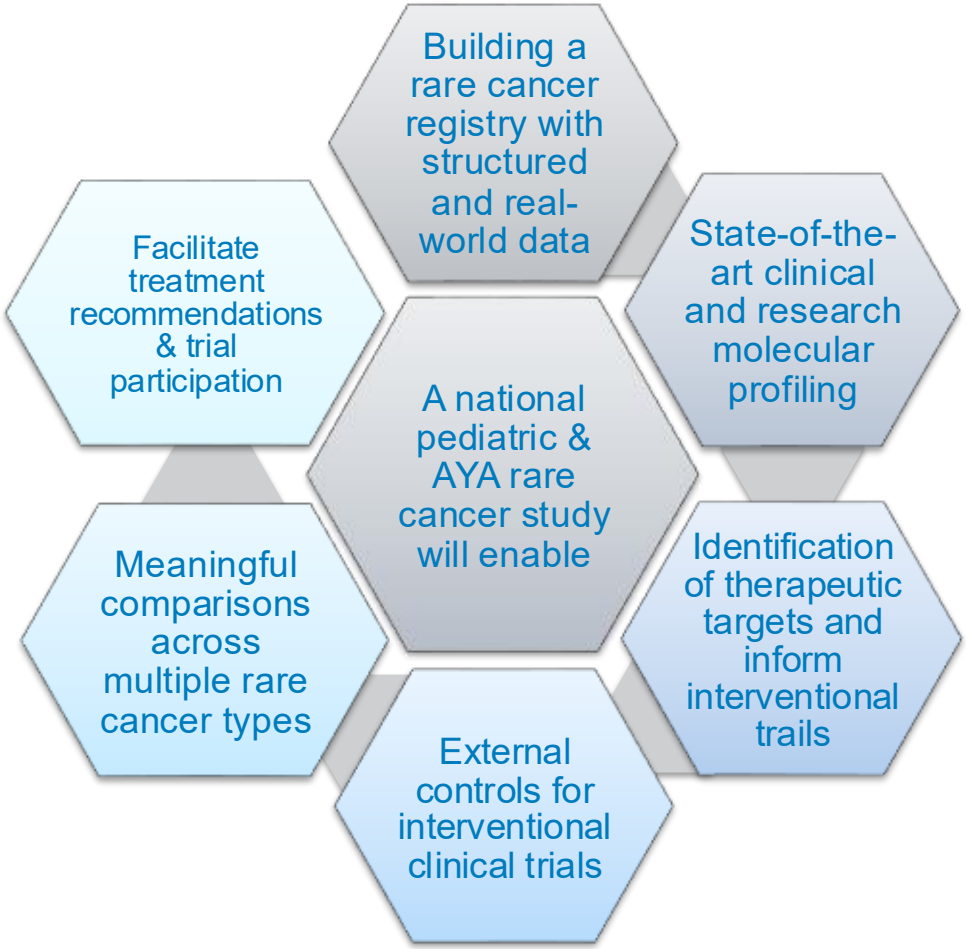
Rare Cancers: Lesson Learned

- Despite ongoing efforts, there remains a large unmet need
- Successful efforts have:
  - Advocacy, patient engagement, and disease champions
- Conducting registry/natural history studies first can facilitate clinical trials
- Achieving meaningful cohorts is time efficient
- Partnership and integration with consortia / COG / PBTC / PNOC / CBTN / disease specific initiatives / community hospitals / advocacy and national experts are critical to accelerate rare tumor efforts
- A national effort will allow enrolling adequate numbers of participants to more rapidly, efficiently, and consistently study multiple rare cancers

CCDI Pediatric and AYA Rare Cancer Study

- A longitudinal observational study for children and young adults with very rare cancers
- A collaboration between:
  - NCI CCDI, Children's Oncology Group (COG), other consortia, advocacy groups, NCI Center for Cancer Research-Pediatric Oncology Branch (CCR-POB)
  - Rare tumor efforts nationally and internationally
  - Regulatory agencies
  - EU Beating Cancer Plan

CCDI Pediatric and AYA Rare Cancer Study



Objectives

Primary Objective:

- To establish a longitudinal observational study and registry for very rare pediatric and AYA solid tumors

Secondary Objectives:

- To evaluate the feasibility of patient reported outcomes (PRO) using validated reporting platforms suitable for pediatric and AYA populations
- To conduct comprehensive clinical molecular characterization, utilizing the CCDI Molecular Characterization Initiative (MCI)

Eligibility and Diagnosis

Eligibility:

- Newly diagnosed very rare solid tumor within 1 year of diagnosis
- Age ≤ 39 years at time of diagnosis

Diagnosis:

- Pathology confirmed rare solid tumor with the exception of:
  - Ewing sarcoma, osteosarcoma, rhabdomyosarcoma, diffuse midline glioma (H3K27 altered), atypical teratoid rhabdoid tumor, pleuropulmonary blastoma, common adult cancers that occur in pediatric/AYA populations (i.e., colorectal cancer, breast cancer)
  - Select molecularly defined rare tumors and rare hematologic malignancies will be added in future

Recruitment and Enrollment

Recruitment:

- COG Project:EveryChild
  - Participants with eligible rare tumors that are also enrolled in MCI
  - Will receive a list of participants from PEC that have consented to “recontact”
- Self-referral
  - Non-COG sites
- Engagement with advocacy groups, other consortia, rare tumor efforts, community hospitals, adult oncology groups

Enrollment:

- Remote via electronic consent
- All participation is remote with no requirement to come to NIH

Study Design

Data Collection:

- Collaborative effort to determine data elements collected
  - CCDI hosted Common Data Element Workshop (July 2024)
  - Harmonization with the European Cooperative Study Group for Pediatric Rare Tumors (EXPeRT)
  - PRO working group
  - Advocacy and patient feedback
- Medical record extraction
  - Medical records requests every 3 months from enrollment
  - Data to include: presenting symptoms, pathology details, treatment details, response to treatment, etc.
- Patient/guardian forms completed electronically
  - Family history, epidemiology questionnaire
  - PROs
    - PROMIS short form completed yearly – examples: depression, anxiety, pain, cognitive function, fatigue
    - PRO-CTCAE/Ped PRO-CTCAE completed every 3 months for those receiving treatment

Study Design

Follow Up:

- All participants will receive yearly request for updated questionnaires/PROs and release of information for medical record collection
- Participants receiving treatment will be asked to complete PRO-CTCAE/Ped PRO-CTCAE every 3 months until off treatment
- Study team will request medical records and extract data every 3 months

Compensation:

- Participants will be compensated for yearly form completion
- Participants who are on treatment and complete the PRO-CTCAE/Ped PRO-CTCAE will also be compensated

Study Design

Data Collection (biospecimens):

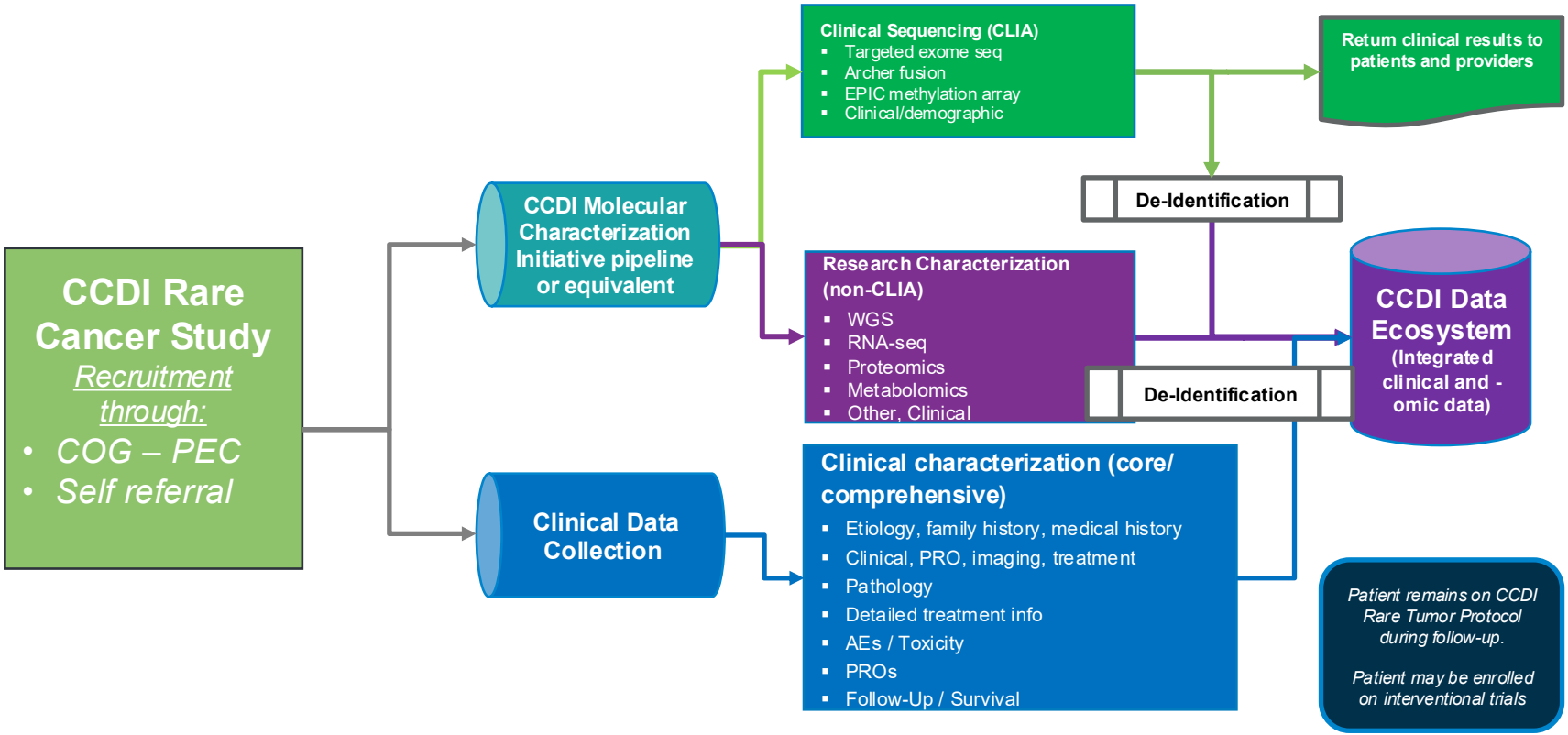
- Clinical molecular characterization
  - Utilizing the CCDI MCI pathway
  - For self-referral patients only
    - Saliva collection tubes sent to participants
  - Results returned to the treating physician
- Research molecular characterization
  - Utilizing CCDI research characterization pathway

Data Sharing:

- Data will be de-identified and deposited in the CCDI Hub every 6 months
- Paired with CCDI MCI data

Study Communications

CCDI Pediatric and AYA Rare Cancer Initiative: Data Flow



Referrals, Questions, and Inquiries

Email NCICCDIRCI@nih.gov

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- **CDE Workshop participants**
- **EXPeRT team members:** Ines Brecht, Ruth Ladenstein
- **PRO working group:** Pamela Hinds, Bryce Reeve
- **Many advocacy organizations and patient participants:** Stupid Cancer (Alison Silberman & Junior Board members), Ann Ramer
- **And so many more!**

Extra thank you to the patients and families!

Q&A / Open Discussion

Join Us at Our Upcoming Webinars

Tuesday, May 12, 2026,
from 1:00–2:00 p.m. ET

Developing Translational and Predictive Imaging Biomarkers for Radiotherapy-Induced Brain Injury in Preclinical Models

Dr. Ethel Ngen
Johns Hopkins University School of Medicine

Tuesday, June 9, 2026,
from 1:00–2:00 p.m. ET

Longitudinal Studies in Genomically Defined Disease Cohorts

Dr. Smita Bhatia
University of Alabama at Birmingham

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Questions? Email us at:
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Thank you for attending!

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