Objectives:

- Challenges Faced by Hermansky-Pudlak Syndrome
- The Role of the Organization Mitigate Our Challenges
- Understand the Contribution That Patients and Non-Profit Organizations Make on Science
HPS Characteristics

Autosomal Recessive – 10 Gene Types

- Albinism
- Low Vision
- Bleeding Problems
- Inflammatory Bowel Disease
- Lung Disease
Challenge: RARE and REMOTE
Cultural and Language
RESEARCH: How we contributed to science

We started at Home!!!

OUR First Science Meeting

The Hermansky-Pudlak Syndrome Network Inc.
Cold Called the NIH!!
Recruitment and Outreach for a drug trial in Puerto Rico
Visual Impairments

Should I participate in Research?!

Cultural Norms

LANGUAGE BARRIER

RELIGIOUS

beliefs

MEDICAL LEAVE

Transportation

Finances

Information privacy

New Patient Engagement Tool!!

Individual Research Plans

My unique challenges with my unique solutions!
INDIVIDUAL RESEARCH PLAN (IRP)

With the help of the IRP TOOL we will be able to educate members on the language and definitions of clinical trials, identify issues preventing our members from taking part in research and develop the resources we need to ensure that they are research ready.

• What are your research choices?
• What are the barriers to your research participation?

• IRP helps to identify our member’s desires regarding research.
• This would include their interest to be involved as well as their interest to NOT be involved.
• If they are interested in research, is it while they are alive or after they pass?
• Are there challenges that might prevent them from being fully engaged in research opportunities?
• Can we help address the individual needs of our members?
Inclusion Initiatives from HPS Network

"WE’RE DRAWN TOGETHER"

Instead of "Patient Engagement" this was "Science Engagement!"

Bringing research to the patients
PATIENT ENGAGEMENT

Multiple Protocols – Single Site
Five different laboratories requested specimens from our members at our 25th Annual HPS Conference

Logistical difficulties included:

- HPS members have low vision so CONSENTS needed to be enlarged or read. Support needed to be provided for consenting.
- Many members do not speak English as the most comfortable language therefore Spanish versions need to be provided or translators assisted.
- Due to the nature of the research, blood had to be immediately sent to the lab for preparation. The collection had to take place within the space of two hours.
EACH PROTOCOL HAD DIFFERENT REQUESTS REGARDING THE DONORS:

1. HPS-1 individuals and their parents
2. All genotypes of HPS
3 & 4. HPS individuals who had not given stool samples before
5. HPS-3 individuals

ALL SPECIMENS HAD TO BE BLINDED AND GIVEN A CODE

The results: IN 1 HOUR AND 55 MINUTES!
All Consents obtained
38 individuals were drawn 96 tubes of blood
4 stool specimens
Research and Social Distancing

Great time for the Symptom Scale Study!!!
ASSESSING FINANCIAL VALUE OF PE:
A QUANTITATIVE APPROACH FROM
CTTI’S PT. GROUPS & CLINICAL TRIALS
PROJECT

• Sponsors are reluctant to go beyond pilot programs because of uncertainty regarding return on investment
• Developed an approach to estimate the value of PE using:  EXPECTED NET PRESENT VALUE (ENPV) integrating key drivers of cost, time, revenue and risk
• Used the impact of PE on ENPV for typical oncology development program entering phase 2 or phase 3
For pre-phase 2 project, the cumulative impact of a PE that avoids ONE protocol amendment and improves enrollment, adherence and retention is an increase in NET PRESENT VALUE of $62MM ($65MM for pre-phase 3) and an increase of ENPV of $35MM pre-phase 2 ($75MM for pre-phase 3).

Compared with an investment of $100,000 in PE the NPV and ENPV increases can exceed 500-fold the investment.

This ENPV increase is the equivalent of accelerating a pre-phase 2 product launch by 2½ years (1½ years for pre-phase 3).
Thank you for listening and please let me know if any of you have great ideas on this topic!

*It’s a matter of life and death!

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