n-lorem FOUNDATION

2023 Nano-rare Patient Colloquium

# Healthcare Reforms Urgently Required to Meet the Needs of Nano-rare Patients

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Hosted by Biogen

# Reform is Needed to Create Widescale Change

Introduce whole genome sequencing to newborn evaluation protocols

# Incorporate Whole Genome Sequencing Into Newborn Screening Protocols

- Benefits
  - Rapid diagnosis of patients with single gene pathogenic mutations
  - Early intervention and treatment of patients with pathogenic mutations
  - Define the prevalence of all pathogenic mutations, including nano-rare
  - Rapid identification of emergent mutations and mutagenic trends
  - Create a compendium of single gene diseases
  - Immediate cost reduction of diagnoses
  - Enormous long-term reduction of health care costs
  - Establish the genomic basis of molecular epidemeology

# Incorporate (2)

- Issues
  - Risk of misuse of genomic information
  - Immediate cost
  - Mis-alignment of insurer short term profit motivation and the long term reduction of health care costs
- Potential solutions
  - Establish rigid bioethical guidelines and make WGS optional till system proven to be effective
  - Federal and state budget allocations in Medicaid and Medicare
  - Federal subsidies, grants or other inducements for insurers to implement WGS

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Couple genomic data to individual phenotypes

# Couple Genomic Data to Standardized Phenotypic Characterization of Individuals with Pathogenic Mutations

- Benefits
  - Creation of a compendium phenotypes for each mutation
  - Define baseline phenotypes for patents to be followed epidemiologically
  - Define a learning protocol for phenotypic characterization
    - Currently a lack of uniformity in the evaluation of phenotypes makes comparison of phenotypes inaccurate
- Issues
  - Patient confidentiality
  - Defining “omic” technologies mature and cost effective enough to be used population-wide
  - Cost
- Potential solutions
  - Rigid bioethical and privacy standards
  - Technical and cost benefit analyses of various “omics platforms”
  - Divert grant funding for more traditional diagnostics and incentives for diagnostic companies to invest

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# Initiate Molecular Epidemiological Studies

- Benefits
  - Coupling “omics” analytics to epidemiological studies of patients with single gene mutations will create new knowledge of the molecular steps that result in loss of healthy phenotypes to expression of full disease phenotypes
  - Performing the same studies on healthy control patients will establish a compendium of healthy genotype/phenotypes
  - Knowledge with change the way we think about health and disease altogether
- Issues
  - Patient confidentiality
  - Cost
- Potential solutions
  - Rigid bioethical standards
  - Reduce funding for traditional epidemiological and GWAS studies and invest in the future of medicine

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Provide symmetric funding

# Provide Balanced Funding for Treatment and Research to Identify Potential Treatments

- Benefits
  - Immediately improved health for nano-rare patients while continuing a search for more effective treatments
  - Enormous advances in knowledge
  - Cost reductions
- Issues
  - Changing the mindsets of grantors in Government and Charitable organizations to recognize that immediate treatment is available for many
  - Cost
- Potential solutions
  - Continue to show benefit with ASOs for many patients
  - Reduce research funding in favor of treatment and studies in the optimal species, humans with single gene mutation caused diseases

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Encourage research on these unique experiments of nature

# Broaden Research on Nano-rare Patients

- Benefits
  - These “unique experiments of nature” in humans will be fully used to learn
  - Immediate insights into HUMAN disease
  - Understanding how effective treatment reverses the ontogeny of diseases
- Issues
  - Funding
- Potential solutions
  - Make research on nano-rare mutations a funding priority
  - Reduce funding on Animal models of Human disease in favor of Human model of Human disease

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Consider a team approach focused on holistic care

# Establish a Team Approach to Holistic Care of the Entire Nano-rare Patient

- Benefits
  - Improved palliative care
  - Reduced burdens place on families and patients
  - Reduced likelihood of “system weariness” or apathy
- Issues
  - None
- Potential solutions
  - DEMAND BETTER CARE
  - Policy changes that promote a team approach for the patient
  - Create a cohesive nano-rare patient and family collective to demand change

# Reforms
## Conclusions

- Only a cohesive integrated effort can policy changes be effected
- Effecting policy changes will be a marathon, not a sprint
- There will be critics who express legitimate concerns
  - Patient privacy
  - Risk of diagnostic labeling being stigmatic
  - Others
- But the first priority must be to save lives and provide a better future for nano-rare patients and families
  - A patient debilitated for life, or who succumbs to a nano-rare mutation is an insoluble problem and lost futures
  - All other concerns can e addressed