The Path of Progress in Science and Medicine
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- Incremental change in a shared body of knowledge
- Application of discoveries from one field to another; e.g. organ transplantation from cancer chemotherapy
- Paradigmatic shifts; e.g. antibiotic treatment of ulcers
The Path of Progress in Research on FASD

- Which research paths will lead to breakthroughs and which are dead ends?
- Which discoveries in science and medicine will advance diagnosis and treatment of FASD?
- Which of the current paradigms for diagnosis and treatment of FASD are impeding progress?
A Half Century of Progress in Science, Medicine, and Technology

1972

2019
A Half Century of Progress in Science, Medicine, and Technology

- MRI Brain Imaging
- Functional Brain Imaging
- Gene Editing
- Monoclonal antibodies
- PCR
- Minimally Invasive Surgery
- Electronic Health Record
- Molecular Biology Revolution
- Desktop Computers
- Electronic Calculators
- CT Brain Scan
- Genome Sequencing
- Internet
Promising Sources of New Knowledge
Million Veteran Program (MVP)

- Bioinformatics
- Augmented Intelligence
- Artificial Intelligence
Gene Editing

- CRISPR/Cas9 already allows precise gene editing
- What genes could be edited to impact FASD
Technology

- Devices will get smaller, faster, smarter
- They will be worthless if we don’t apply them to the right problem in the right way
Global Challenge of FASD

- Diagnosis and treatment are unavailable for most affected individuals:
  - Insufficient numbers of clinicians
  - Geographic inaccessibility
  - Stigma

- We can’t even agree on how to diagnose FASD
The Path Forward for FASD

To move forward, we must agree on where we are going and speak a common language.

We need a consensus on diagnosis for FASD.
We need a consensus on diagnosis for FASD.

NIAAA endorses this goal and will support its accomplishment.
Thoughts and challenges on diagnosis: Where are we going and why

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Things do change

Circa 1975

2018
Diagnostic Guidelines

• “A more consistent set of diagnostic procedures and practice guidelines is of utmost importance.”

• “one of the primary problems is inconsistent diagnosing, which I agree represents a real, ongoing concern across the spectrum of mental disorders.”
  – Grohol, 2018, Psych Central
What do we know?

• There is a large disconnect between the number of diagnostic providers and the number of impacted individuals.

• Lack of a consensus diagnostic schema or classification
  – Revised IOM, Old Canadian guidelines, New Canadian guidelines, Australian guidelines, 4-digit code, etc

• Many health service providers are reluctant to make a diagnosis
  – Lack of confidence, stigma
  – Many physicians have stated that they have seen cases of FAS or FASD and did not make a diagnosis
What do we have
Where are we going?
Photographic analysis and telemedicine

- Currently we have a few examples of the use of photographs to assist in diagnosis
  - 2d Imaging
    - Photographic screening tool of the 4 digit code
    - Face2Gene
  - 3d Imaging
    - CIFASD
    - Australian Group
    - South African Group
    - Polish Group
  - Telemedicine

- Photographic screening may be useful in detecting individuals with only subtle facial features, but who do have cognitive or behavioral problems
- Telemedicine may be able to deal with geographical isolation
Where are we going?
Evolution in the technology
FaceScreen Application
- In depth assessment of facial dysmorphology
- Report generation

FaceScreen Upload Portal
- No user interaction
- 3D Image upload -> FASD report
Where are we going?
Brain Imaging

https://journals.plos.org/plosone/article?id=10.1371/journal.pone.0086733

Figure 4. Snapshot of the SBS2 real time brain imaging system running on a Samsung Galaxy Note 2.
Where are we going?
Ehealth technologies

• NEBA is the first of a new kind of medical device cleared by FDA that uses brainwaves (EEG) to help clinicians more accurately diagnosis ADHD in children and adolescents (ages 6 – 17.99 years).
Where are we going?
Biomarkers

• Need evidence of exposure
  – Several proposed biomarkers in the affected individual
    • miRNA
    • Immunological markers
    • Epigenetic markers
    • Metabolomics
    • Proteomics
    • Alcohol metabolites in dried blood spots
But is it possible?

- The number of applications for molecular-based blood tests has risen dramatically.
- A $20 billion market in 2017, projected to be $25 billion in 2022
- Devices that can download results directly to an electronic medical record are more common
- Tests that use analytes other than blood—saliva, urine, or even breath
- Looking for the startrek body scanner
  - Xprize competition: $10 million prize
Ethical issues

- Consent
- Privacy
THANK YOU

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