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RESEARCH, EDUCATION, SERVICE

The Science, Social, and Cultural Aspects of Disability: The Intersection of Disability Identity and Science

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Disability Identity and Attitudes Towards Prenatal Testing in the Osteogenesis Imperfecta (OI) Community

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University of Cincinnati Center for Excellence in Developmental Disabilities

Disability Identity

Disability identity refers to possessing a positive sense of self and feelings of connection to, or solidarity with, the disability community. A coherent disability identity is believed to help individuals adapt to disability, including navigating related social stresses and daily hassles.

Disability identity:

- Is challenging to measure
- Is complex
- Is dynamic

Prenatal Testing and the Disability Community

Some members of the disability community view prenatal diagnostic testing:

- as eugenic because it may result in decreasing the number of people with disabilities.
- as helpful in making decisions about biological parenting
- as neither positive or negative

Attitudes toward Prenatal Testing in the OI Community

	With OI (n=74)	Without OI (n=85)	P-value
Age, mean (SD) in years	43 (11.9)	44 (11.4)	0.7508
Race, White, % (n)	96 (71)	92 (78)	0.3402
Sex, female, % (n)	95 (70)	94 (80)	1.0000
Education, bachelor's degree or higher, % (n)	62 (46)	71 (60)	0.2609
Employment, full time, % (n)	39 (29)	49 (42)	0.1959
Respondent, type 1, % (n)	72 (53)	N/A	N/A
***Partner has OI, % (n)**	15 (11)	31 (26)	0.0193
***Has child(ren) with OI, % (n)**	70 (52)	100 (85)	<.0001
***Has >1 child with OI, % (n)**	20 (15)	5 (4)	0.0030

* Significantly different at $p<0.05$, comparing respondents with OI to those without.

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Attitudes toward Prenatal Testing in the OI Community

- Questionnaire on Disability Identity and Opportunity (QDIO)

	With OI Mean (SD)	Without OI Mean (SD)	p-value
***Pride**	3.280 (0.815)	3.529 (0.815)	0.0437
Exclusion	2.412 (0.950)	2.303 (0.814)	0.4412
Social Model	3.779 (0.607)	3.875 (0.604)	0.3240
Medical Model	3.282 (0.602)	3.260 (0.531)	0.8103

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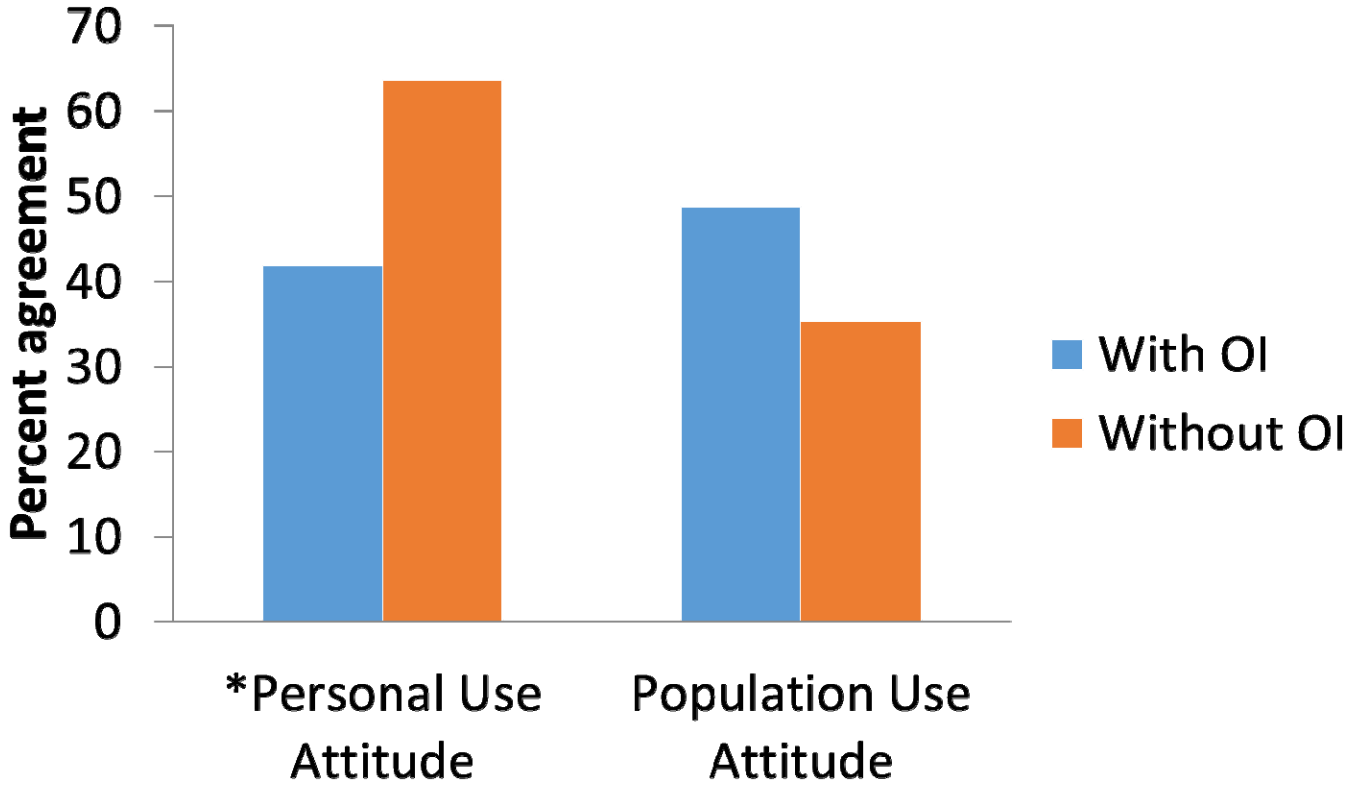
Attitudes toward Prenatal Testing in the OI Community



**Significantly different at p<0.05, comparing respondents with OI to those without OI.*

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Future Research on the Intersection of Disability Identity and Science

- Should prioritize important questions raised by our community
- Should include individuals with disabilities as researchers
- Holds exciting potential for all of us

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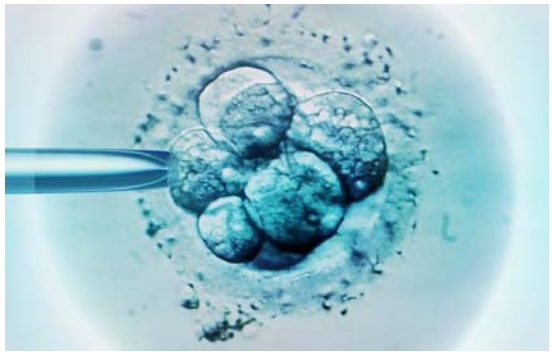
On the Intersection of Disability Identity and Advances in IDD Science



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IDDRC

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A brief update of the state-of science in service of reconciling what exists with what is now within reach

Advances in IDD Science

DD PREDICTION / PREVENTION

The number of Intellectual and Developmental Disabilities for which the **cause** can be traced to a specific variation in the genome is rapidly accelerating, now for over 30 per cent of all people with intellectual disability.

This converts an unexplained condition to one with a known cause, identifies potential opportunity for specific therapy, and specifies risk to family members some who may be silent carriers of an IDD-causing mutation.

"A revolution in clinical genomics"

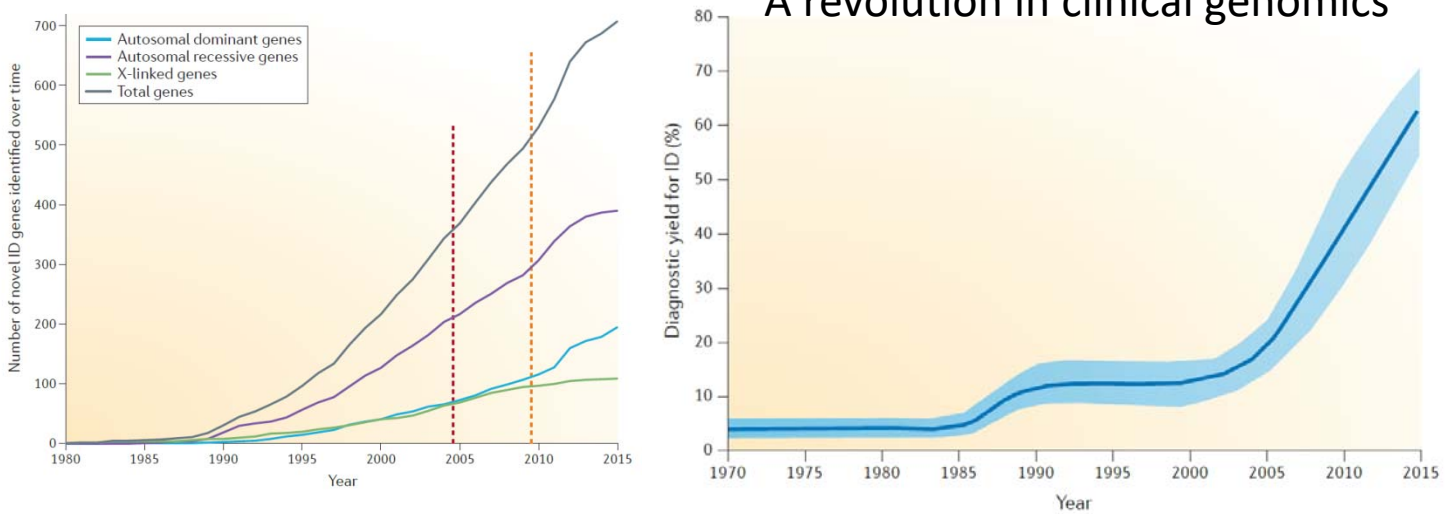


Figure 2 | **Diagnostic yield for ID over time.** Graphical overview of the diagnostic yield for moderate to severe intellectual disability (ID) (excluding Down syndrome, which represents 6–8% of all ID) over time. Solid line indicates the mean of published studies, and the shaded background indicates the lower and upper boundaries of reported diagnostic yields. In the 1970s, conventional karyotyping became a routine diagnostic test and provided a conclusive diagnosis in 3–6.5% of ID cases. The

Intervention at the level of behavior: Personalized Developmental Therapy

ORIGINAL ARTICLE

An evaluation of the effects of intensity and duration on outcomes across treatment domains for children with autism spectrum disorder

E Linstead[1], DR Dixon[2], E Hong[2], CO Burns[2,3], R French[1], MN Novack[2] and D Granpeesheh[2]

Citation: Transl Psychiatry (2017) **7**, e1234; doi:10.1038/tp.2017.207

www.nature.com/tp

ABA-based developmental therapy
N=1,468

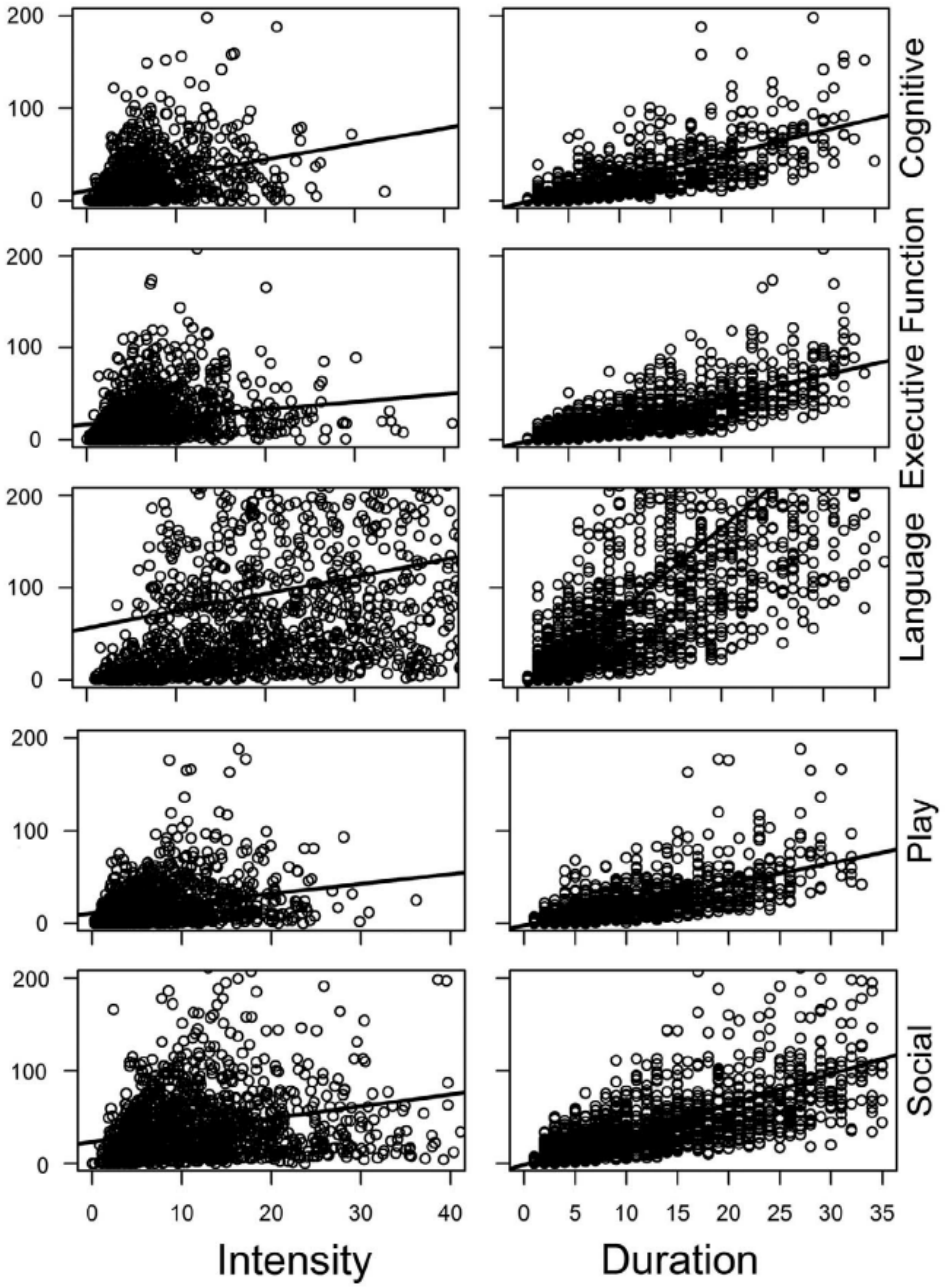


Figure 1. Two-dimensional linear model projections for each treatment domain.

Intervention at the level of circuit:

Management of neuropsychiatric comorbidity

Resolution of Compulsive Behavior with SSRI

Paradigm of Treatment "Indication" from the Y-BOCS:

--O-C symptom burden (inventory of O/C)
--What aspects/proportion of daily life are compromised by symptoms?
--How much power to resist the adverse influence of symptoms?

ORIGINAL RESEARCH

WILEY Brain and Behavior Open Access

February, 2018

The effects of cognitive behavioral therapy on resting-state functional brain network in drug-naive patients with obsessive–compulsive disorder

Ping Li[1*] | Xiangyun Yang[2*] | Andrew J. Greenshaw[3] | Sufang Li[4] | Jia Luo[2] | Haiying Han[2] | Jing Liu[2] | Zhaoxi Zhong[2] | Zhihua Guo[2] | Hongfang Xiong[2] | Shumin Yao[2] | Yunhui Chen[1] | Jing Sun[5] | Zhanjiang Li[2]

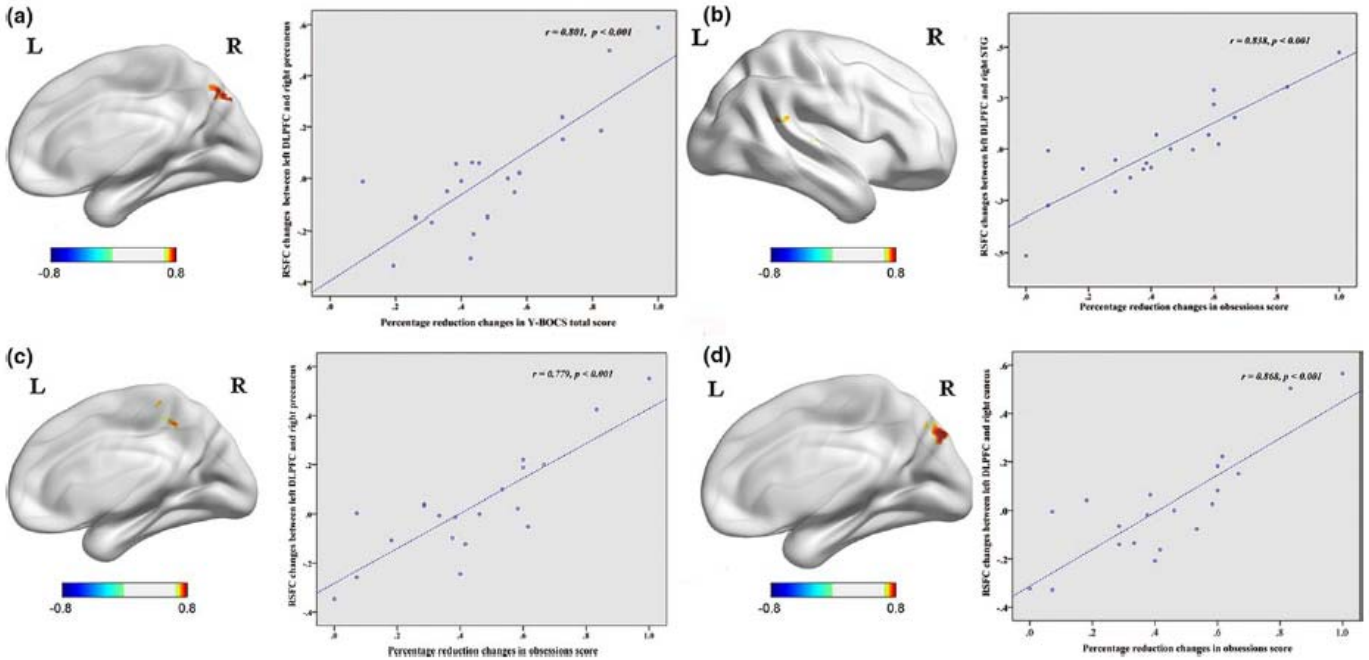


FIGURE 5 (a) Positive correlation between RSFC changes (left DLPFC and right precuneus) and percentage reduction changes in Y-BOCS total score. (b) Positive correlation between RSFC changes (left DLPFC and right superior temporal gyrus) and percentage reduction changes in obsessions score. (c) Positive correlation between RSFC changes (left DLPFC and right precuneus) and percentage reduction changes in obsessions score. (d) Positive correlation between RSFC changes (left DLPFC and right cuneus) and percentage reduction changes in obsessions score. L, left side; R, right side. The threshold was a voxel *p*-value <.001, a cluster *p*-value <.05, two-tailed (GRF correction)

The NEW ENGLAND JOURNAL of MEDICINE

ESTABLISHED IN 1812 NOVEMBER 2, 2017 VOL. 377 NO. 18

Single-Dose Gene-Replacement Therapy for Spinal Muscular Atrophy

J.R. Mendell, S. Al-Zaidy, R. Shell, W.D. Arnold, L.R. Rodino-Klapac, T.W. Prior, L. Lowes, L. Alfano, K. Berry, K. Church, J.T. Kissel, S. Nagendran, J. L'Italien, D.M. Sproule, C. Wells, J.A. Cardenas, M.D. Heitzer, A. Kaspar, S. Corcoran, L. Braun, S. Likhite, C. Miranda, K. Meyer, K.D. Foust, A.H.M. Burghes, and B.K. Kaspar

Advances in IDD Science

INTERVENTION AT THE LEVEL OF GENES AND THEIR FUNCTION

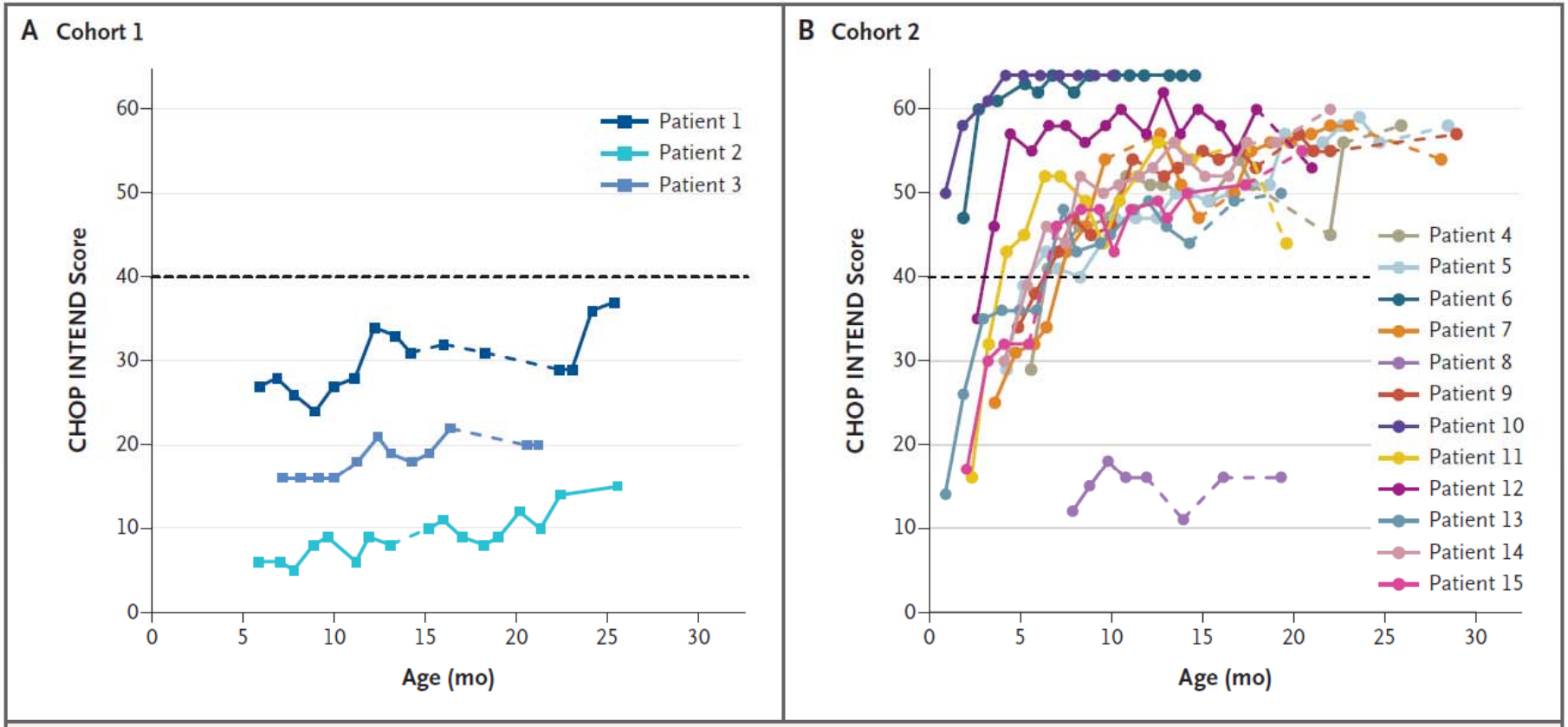
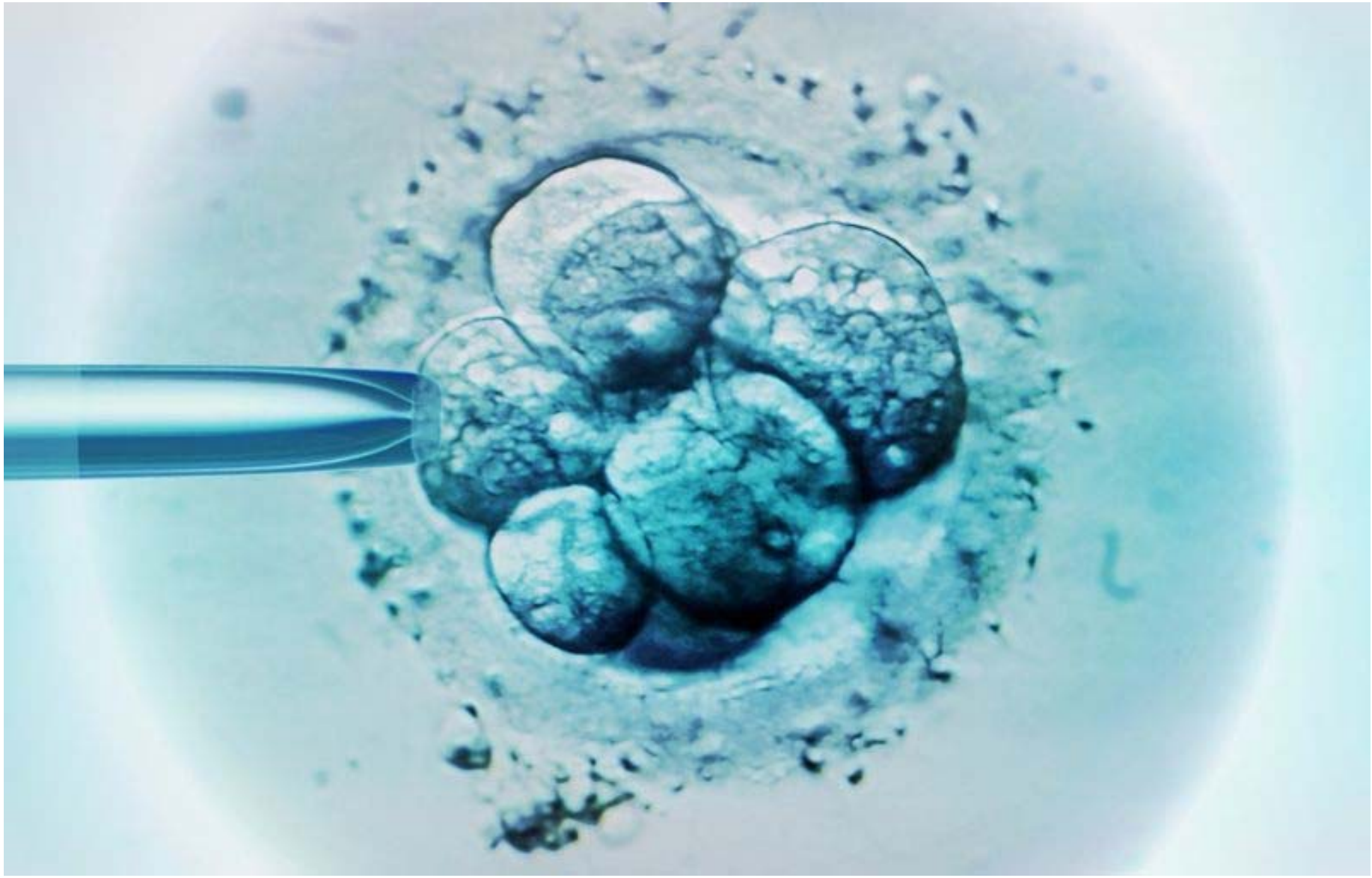


Figure 2. Motor Function after Gene Therapy.

Shown are changes in the score for motor function on the CHOP INTEND (Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders) scale among the 3 patients in cohort 1 (Panel A) and the 12 patients in cohort 2 (Panel B) who received gene therapy with adeno-associated viral vector containing DNA coding for SMN. The scale ranges from 0 to 64, with higher scores indicating better motor function; historical controls with spinal muscular atrophy type 1 never reach 40 points (indicated by the black dashed line). The dashed lines on the individual patient curves indicate either a missed assessment or a partial assessment because of illness, lack of cooperation, or fatigue of the patient; such data were not included in the analyses. The timing of the administration of gene therapy in Figure 1 can be matched with the data shown here for each patient.

Editing the Human Genome in Cells, Tissues, and Whole Organisms



When would an individual opt to pursue a potential “cure”?

How much risk of treatment?
How much risk of non-treatment?
How much benefit of treatment?
How much benefit of non-treatment?
What is the difference between treatments that cure
and treatments that improve adaptation?
Who should be adapting to whom?

Brian Madeux, 44, receives the first human gene editing therapy at the UCSF Benioff Children's Hospital in Oakland, Calif., on Nov. 13, 2017.

ERIC RISBERG / AP

"I'm nervous and excited," Madeux said as he prepared to leave the hospital. "I've been waiting for this my whole life, something that can potentially cure me."

How to think about and prepare
together
for a revolution-in-opportunity
for higher-impact therapy…

Vignettes

DISCUSSION

Common principles in how we think about these intersections between disability identity and science

Language

What language did you hear that you resonated with?

What language was challenging for you to hear?

Identity vs. Disease

How do we understand when a disease process or condition becomes a persons' identity?

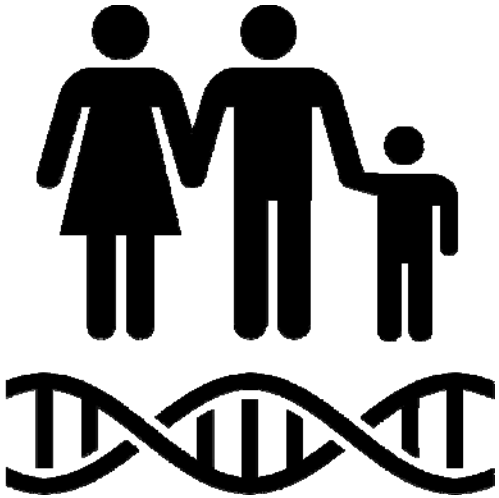
How do we think about this continuum and how have we thought of these two things as being different?

Created by Egon Låstad
from Noun Project

Cure vs. Identity

- How do we decide when cure is reasonable and needed vs. scientific innovations?
- For example, most people think a cure for the common cold or cancer is a good thing, but what about intellectual disability? Autism?

Genetics



What are the implications for prediction, cell selection and gene editing that are used to correct an abnormality of a disability?

How do we as a field prepare for the implications of this reality caused by advances in science?

Interventions

How do we determine if specific interventions should be tried and for what reasons?

For example, if an epilepsy medication can reduce symptoms experienced by people with ASD, should we try them?

Individual choice about quality of life

How do we understand quality of life from the individual perspective? Family perspective?

What are the implications for intervention?

For example, if through scientific intervention we can improve the intellectual functioning of a person with intellectual disability, should we?

Common values and principles

What are the core values and principles we should adhere to when we think about the advancements in science and their intersection disability identity?

Worries and thoughts

What are the things you are thinking about and worrying about related to these issues and the intersection of disability identity and science?

Next Steps

How do we bring voice to these issues by people with disabilities in a proactive way?

What can AUCD do to create bridges between this type of science, people with disabilities, practitioners and allies?