

# The Importance of Citizen Science in Autism Research



Jill Escher, MA, JD  
@JillEscher

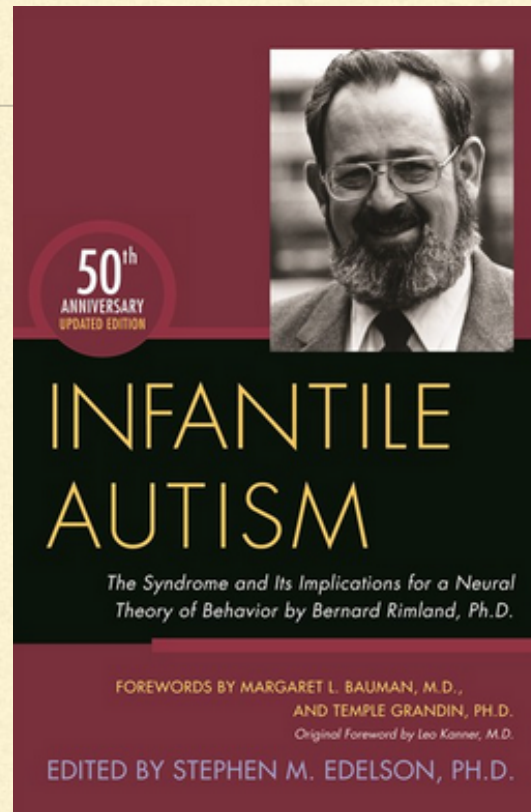
Escher Fund  Humans start as molecules  
for Autism  
[GermlineExposures.org](http://GermlineExposures.org)



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# Citizen Science: Part of ASA's Heritage

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ASA co-founder **Bernie Rimland**, Ph.D: Infantile Autism  
The Syndrome and Its Implications for a Neural Theory of Behavior

Citizen science can change history.

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# I love formal science, but...

“Science” can err

Dogma, funding, conservatism  
can stifle innovation

Ivory tower syndrome

Technology empowers citizen  
science

We have data, too





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# Four paths of autism citizen science

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- Hypothesis Hunting (causation, treatment)

- Philanthropy (you don't need to be rich)

- Crowdsourcing

- Educating and Advocating

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# An accidental hypothesis hunter



I was born in 1965 in Los Angeles.

Three beautiful children from normal pregnancies.

No history of ASD, no other risk factors.

**Yet two kids nonverbal autistic.**



Son, 17

Daughter, 10



Why?  
What happened?



# My kids' autism is a puzzle

## Genetics?

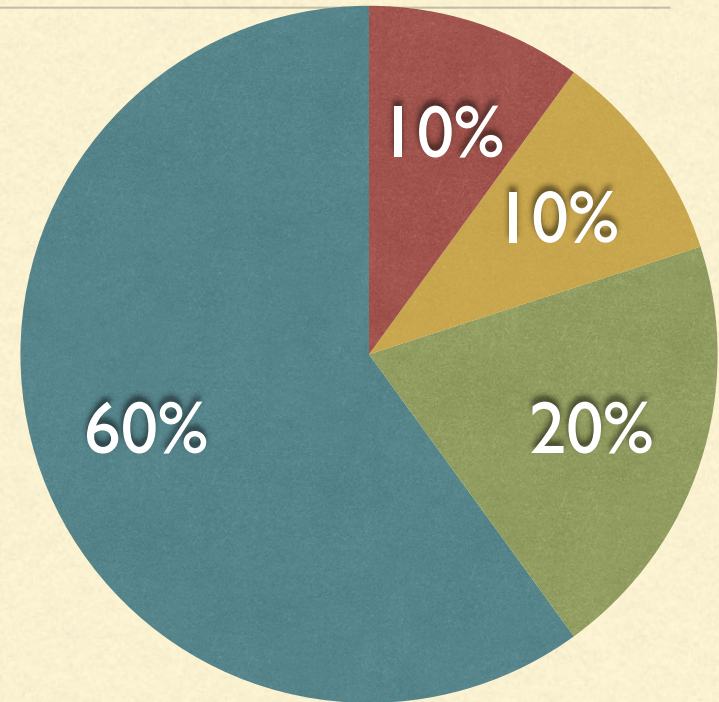
About 10% of cases can be attributed to known genomic errors, 10% more forecast. But not found in my kids.

## Environment?

Exogenous factors—ie, prematurity, certain drugs, maybe 20%. No environmental risk factors with my kids.

## What else could it be?

At least 60% of ASD cases unexplained, but strong evidence of heritability. My kids fall in here: a puzzle.



What we know about causes of ASD



# Then, a trio of bizarre discoveries

1.

Handwritten medical record from 1965, likely a prenatal or obstetric record. The document is filled with handwritten notes, including dates (e.g., 3-2-65, 3-8-65, 4-12-65, 5-12-65, 5-26-65), measurements (e.g., 145, 150, 154, 157, 162, 165, 168, 170, 172, 175, 178, 180, 182, 185, 188, 190, 192, 195, 198, 200, 202, 205, 208, 210, 212, 215, 218, 220, 222, 225, 228, 230, 232, 235, 238, 240, 242, 245, 248, 250, 252, 255, 258, 260, 262, 265, 268, 270, 272, 275, 278, 280, 282, 285, 288, 290, 292, 295, 298, 300, 302, 305, 308, 310, 312, 315, 318, 320, 322, 325, 328, 330, 332, 335, 338, 340, 342, 345, 348, 350, 352, 355, 358, 360, 362, 365, 368, 370, 372, 375, 378, 380, 382, 385, 388, 390, 392, 395, 398, 400, 402, 405, 408, 410, 412, 415, 418, 420, 422, 425, 428, 430, 432, 435, 438, 440, 442, 445, 448, 450, 452, 455, 458, 460, 462, 465, 468, 470, 472, 475, 478, 480, 482, 485, 488, 490, 492, 495, 498, 500, 502, 505, 508, 510, 512, 515, 518, 520, 522, 525, 528, 530, 532, 535, 538, 540, 542, 545, 548, 550, 552, 555, 558, 560, 562, 565, 568, 570, 572, 575, 578, 580, 582, 585, 588, 590, 592, 595, 598, 600, 602, 605, 608, 610, 612, 615, 618, 620, 622, 625, 628, 630, 632, 635, 638, 640, 642, 645, 648, 650, 652, 655, 658, 660, 662, 665, 668, 670, 672, 675, 678, 680, 682, 685, 688, 690, 692, 695, 698, 700, 702, 705, 708, 710, 712, 715, 718, 720, 722, 725, 728, 730, 732, 735, 738, 740, 742, 745, 748, 750, 752, 755, 758, 760, 762, 765, 768, 770, 772, 775, 778, 780, 782, 785, 788, 790, 792, 795, 798, 800, 802, 805, 808, 810, 812, 815, 818, 820, 822, 825, 828, 830, 832, 835, 838, 840, 842, 845, 848, 850, 852, 855, 858, 860, 862, 865, 868, 870, 872, 875, 878, 880, 882, 885, 888, 890, 892, 895, 898, 900, 902, 905, 908, 910, 912, 915, 918, 920, 922, 925, 928, 930, 932, 935, 938, 940, 942, 945, 948, 950, 952, 955, 958, 960, 962, 965, 968, 970, 972, 975, 978, 980, 982, 985, 988, 990, 992, 995, 998, 1000). The notes include clinical observations, measurements, and possibly treatment details.

In 2010, I obtained my mother's 1965 obstetric records. What did they mean?

2.

## Prenatal Exposure to Synthetic Progestins and Estrogens: Effects on Human Development

June Machover Reinisch, Ph.D.,<sup>1</sup> and William G. Karow, M.D.<sup>2</sup>

Seventy-one offspring of mothers administered combinations of synthetic progestins and estrogen for the maintenance of at-risk pregnancy were evaluated for their performance on IQ and personality tests. Siblings born of untreated pregnancies acted as controls. Hormone-exposed subjects were partitioned into three treatment subgroups dependent on the ratio of progestin to estrogen administered to their mothers during pregnancy. No difference in IQ was obtained among the three treatment subgroups even when scores were adjusted for sibling score and prenatal and perinatal complications. Responses to the personality questionnaire provided significant differences among the three groups. The group exposed to the progestin regime (progestin alone or in combination with very low doses of estrogen) and the estrogen regime (higher doses of estrogen than progestin) were most dissimilar. Progestin regime exposed subjects were characterized as more independent, sensitive, self-assured, individualistic, and self-sufficient. In contrast, the subjects exposed to the estrogen regime were more group oriented and group dependent. Analysis of difference scores generated by subtracting the score of an unexposed sibling from that of the exposed cosibling provided similar results. A general discussion is presented on the efficacy of hormone treatment for pregnancy maintenance, augmented fetal wastage of males, birth order and treatment, maternal knowledge of treatment and its possible postnatal effects on the offspring, and drug effects on the fetus.

**KEY WORDS:** synthetic progestin; estrogen; diethylstilbestrol; humans; personality; IQ; pregnancy maintenance; prenatal.

In 2011, I discovered I had been a subject in a study (Reinisch 1977) examining fetal effects of synthetic steroid hormone drugs.

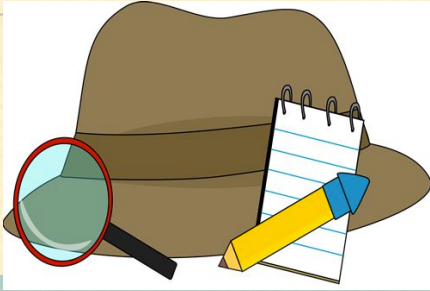
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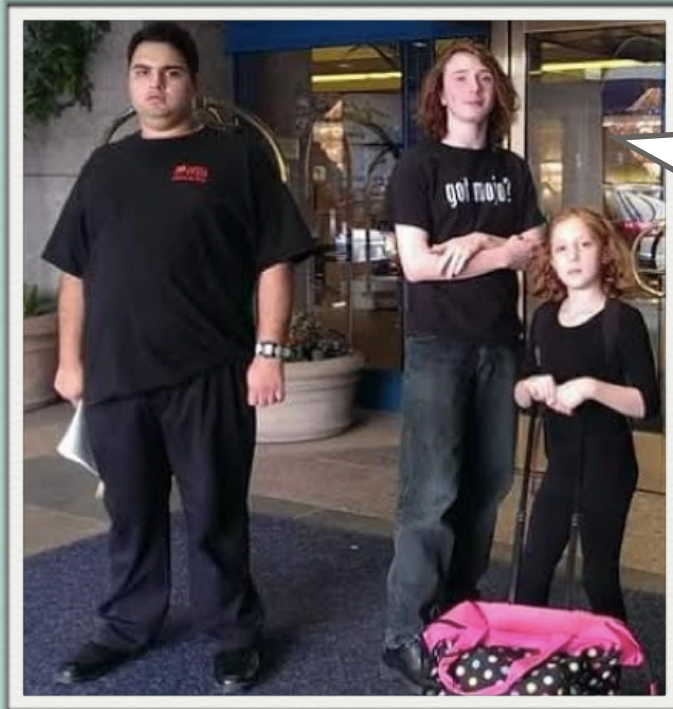
In 2013, I obtained records from the Kinsey Institute detailing my prenatal drug exposures.

Bottom line: Unbeknownst to me, I had been prenatally exposed to a truckload of gene-tinkering drugs





## Others shared this story: An example



We started as  
eggs when our  
mom was a fetus.

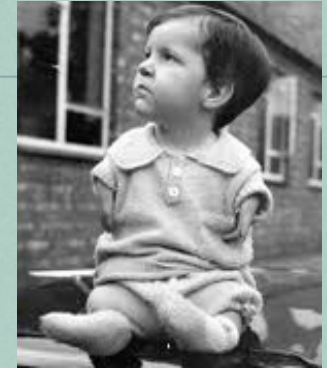
Joan Hutchens was also exposed prenatally  
to an “anti-miscarriage” hormone regimen in 1965.

**Three of her five children, above, have idiopathic autism.**

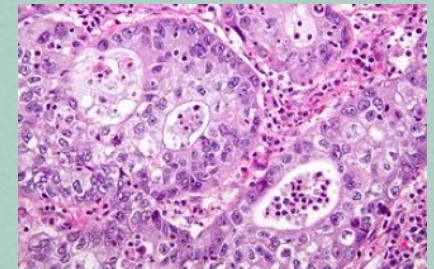
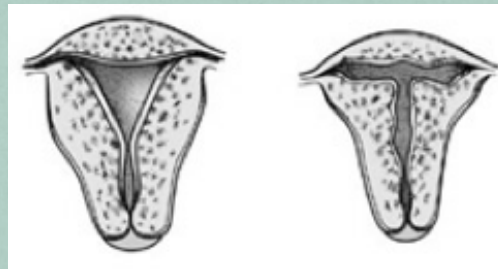
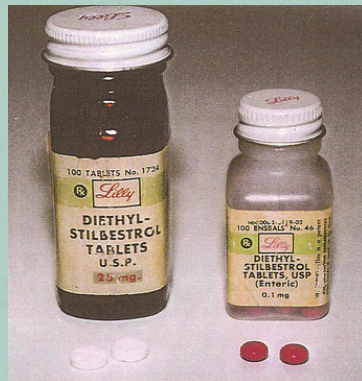


# No question that toxic pregnancy exposures can have devastating effects

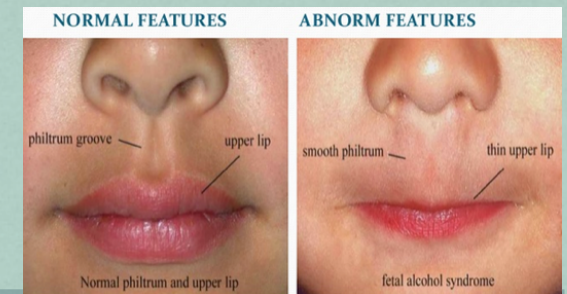
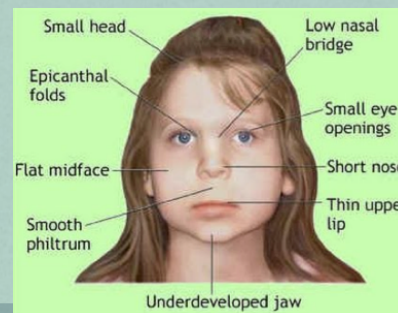
## Thalidomide



## DES (Diethylstilbestrol)



## Fetal Alcohol Syndrome



**BUT there's more ... pregnancy exposures may also affect grandchildren**



**F0 = Mother**

**F1 = Fetus**

**F2 = Germ cells / grandchildren**

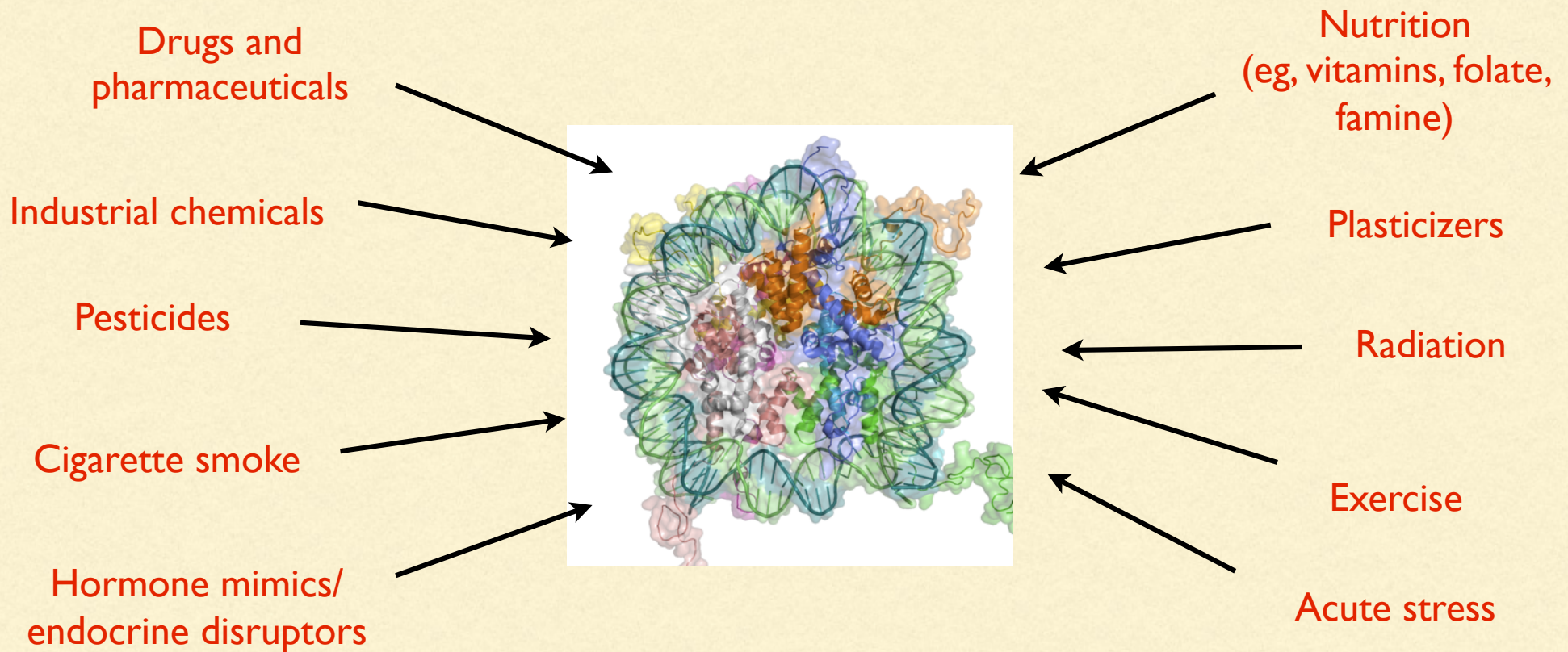
Via the vulnerable fetal germ cells



# Germline mutation or epi-mutation

Mutation: Change in DNA sequence.

Epigenetics: “Heritable changes in gene expression caused by mechanisms other than alterations to underlying DNA sequence.”



# The medicated pregnancy was pretty much a vast human experiment

## Synthetic hormones



**"Really?"**

Yes... **desPLEX**<sup>®</sup>  
to prevent ABORTION, MISCARRIAGE and  
PREMATURE LABOR

recommended for routine prophylaxis  
in ALL pregnancies...

96 per cent live delivery with desPLEX  
in one series of 1200 patients\*—  
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(Each desPLEX tablet starts with 25 mg. of diethylstilbestrol, U.S.P.,  
which is then ultramicrozoned to smooth and accelerate absorption and  
activity. A portion of this ultramicrozoned diethylstilbestrol is even in-  
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desPLEX tablets also contain vitamin C and certain members of the  
vitamin B complex to aid detoxification in pregnancy and the effec-  
tion of estrogen.)

For further data and a generous  
trial supply of desPLEX, write to:  
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REFERENCES:  
1. Cantor, E. H., et al.: *Am. J. Obst. & Gynec.* 68:1296, 1953.  
2. Gorman, L., and Kagan, A. M.: *J. Am. Med. Ass.* 140:1212, 1950.  
3. Kagan, A. M., and Gorman, L.: *J. Am. Med. Ass.* 141:1188, 1953.  
4. Kagan, A. M., and Gorman, L.: *Obst. & Gynec.* 47:41, 1954.  
5. Kagan, A. M., and Gorman, L.: *Obst. & Gynec.* 47:41, 1954.

GRANT CHEMICAL COMPANY, INC., Brooklyn 26, N.Y.

## Sedatives, barbiturates

when  
you  
prescribe  
**CARBRITAL**<sup>®</sup>  
pentobarbital sodium and carbitol  
in Kapseals<sup>®</sup> and Elixir form


you  
prescribe  
sleep



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## Anti-nausea drugs

NOW SHE  
CAN COOK  
BREAKFAST  
AGAIN




... WHEN YOU PRESCRIBE NEW  
**MORNIDINE**<sup>™</sup>  
(BRAND OF MEFAMIDINE)

## Amphetamines

to meet the threat of excess weight gain  
in your obstetrical patients

**PRELUDIN**<sup>®</sup>  
specifically for weight reduction



31 43 44 45 46 47

**GEIGY**  
HEALTH CARE

The fetus and its proto- sperm/egg were of little concern.



# (Other postwar exposures, too)



Pesticides (eg, DDT)



Agent Orange (dioxin)



Plasticizers  
(eg, BPA, phthalates)



Flame retardants  
(eg, PBDEs)



PCBs



Air pollution



Radiation



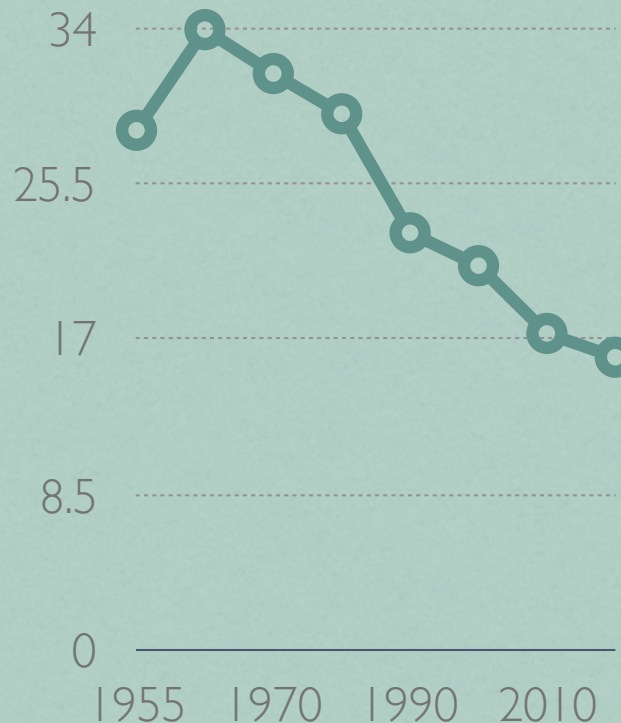
# But the mother of all pregnancy exposures was smoking



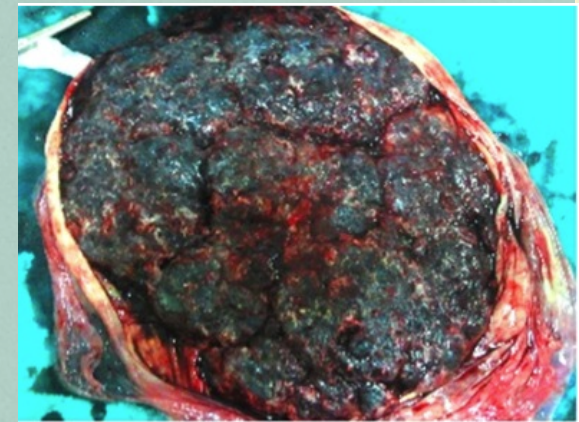
Doctors sometimes recommended it as an appetite suppressant



Smoking prevalence among US females



Cigarette smoke = toxic, mutagenic and epimutagenic components



**Do we feel the consequences today?**



# For example, heavy grandmaternal smoking is common in grandchild ASD

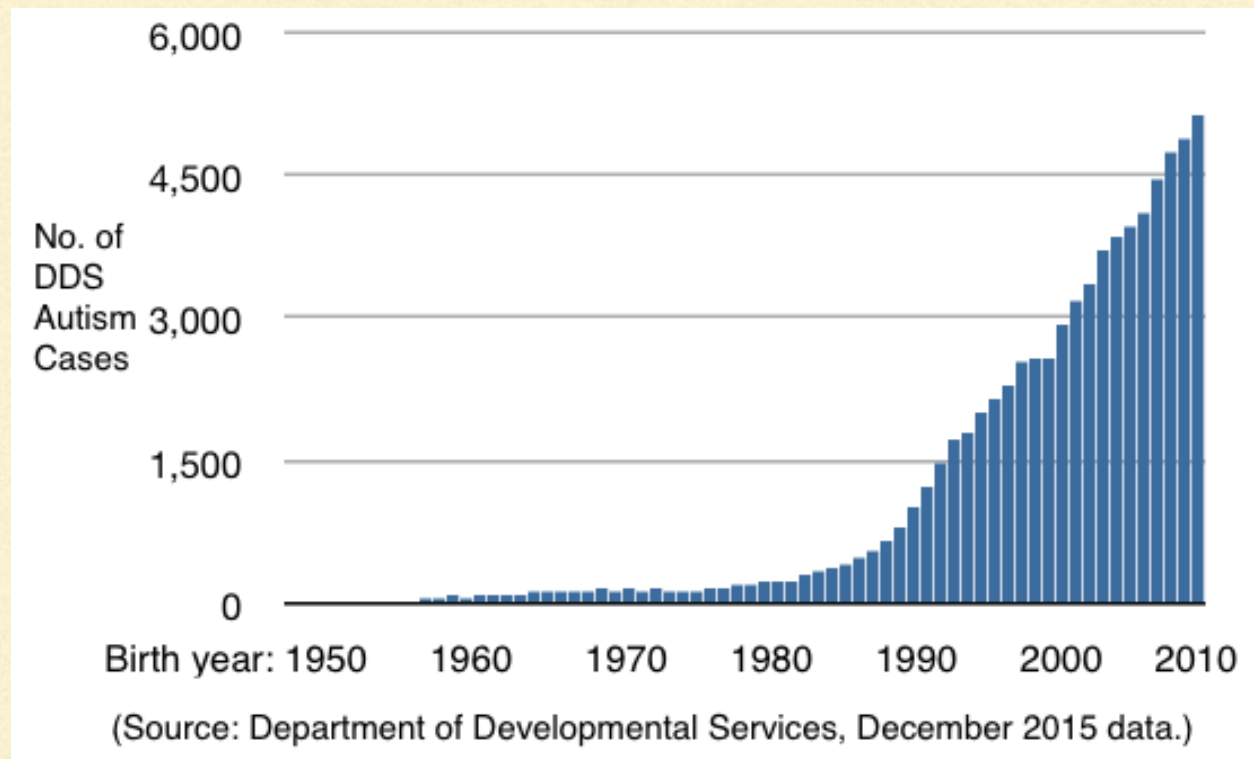
Families had no history of autism. Sampling of the grandchildren.



Yes, these are anecdotes but they may raise important questions about potential germline mutagenesis or epimutagenesis.

# We still have few answers about the main drivers of ASD surge

Calif. Department of Developmental Services  
Autism Cases by Birth Year 1943-2010



## Citizen science can help



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# Jill's Citizen Science Hypothesis

Part of the rise in autism of the 1980s-2000s can be attributed to sperm and egg errors caused by certain pregnancy exposures of the 1950s-1970s, including:

- Smoking
- Artificial hormones
- General anesthesia

**I think autism families are a largely untapped goldmine of ideas for research.  
What are your ideas?**

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## 2: Be a Philanthropist

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\$\$ you raise can seed innovative research and you can help inform the study purpose and design.

- Step 1: Create a donor-advised fund
- Step 2: Create a “Request for Proposals”





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## 3: Crowdsource

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Simons SPARK project



Crowdsourcing data via  
mobile app (eg, Stanford  
project)



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## 4: Educate and Advocate

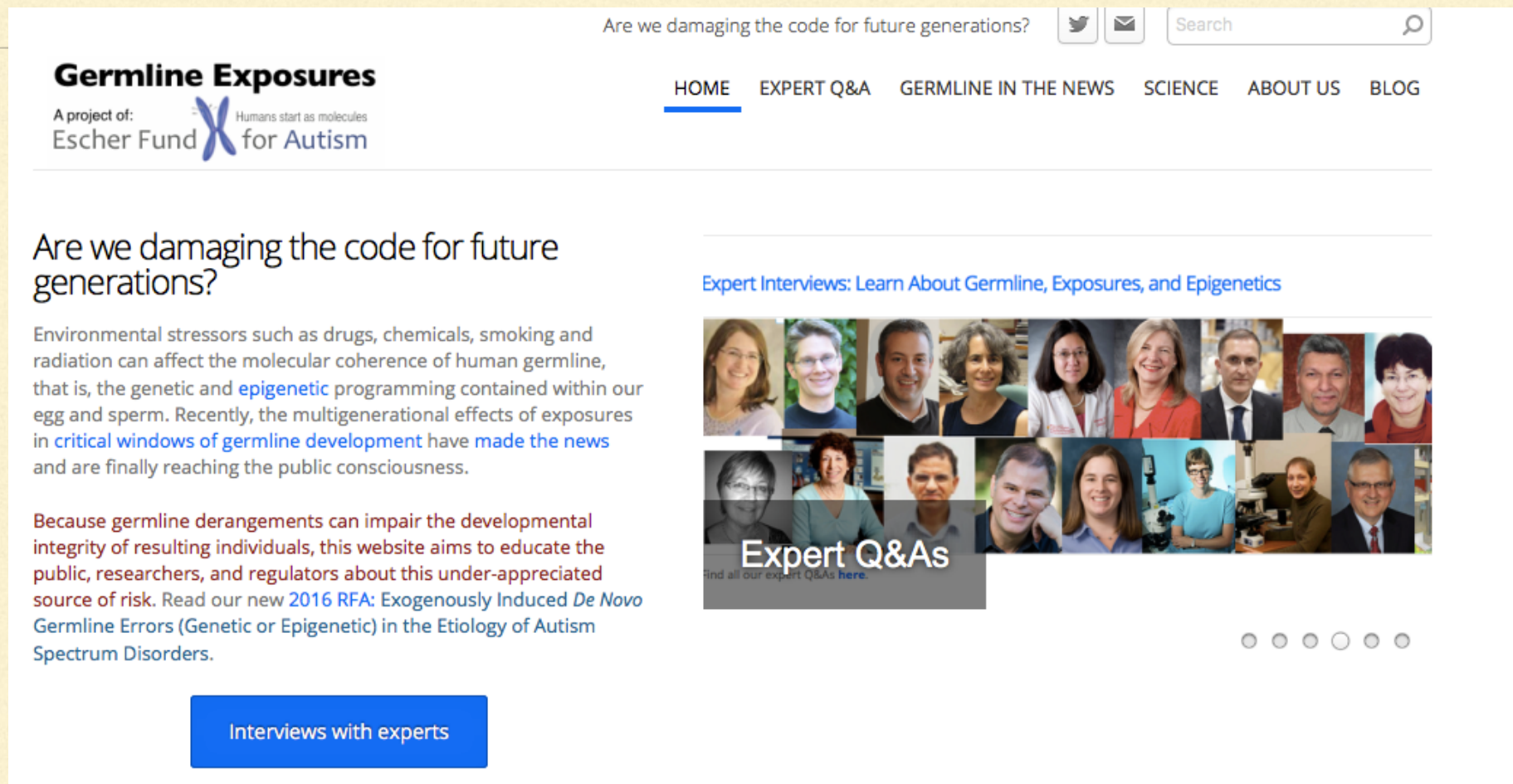
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- Organize a conference
- Build a website
- Write a report





# Build a science education website



[GermlineExposures.org](http://GermlineExposures.org)