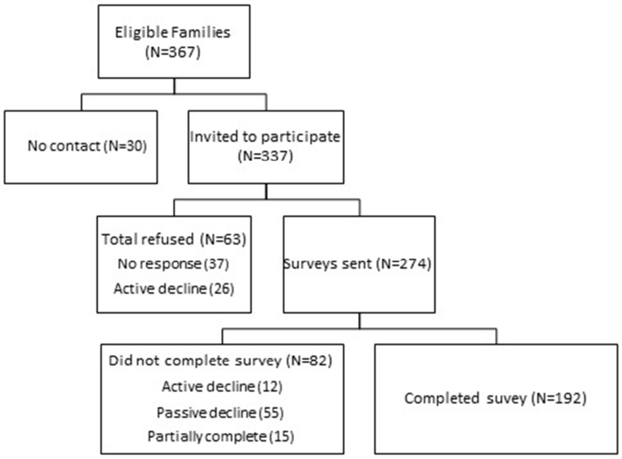

203x162mm (96 x 96 DPI)

Table S1. Survey measures

Measure	**Analysis**	**Interpretation**	**Cronbach α**
Internal Health Locus of Control (IHLC)	Continuous	Higher score= Greater perceived control over health	0.75
Healthy Behaviors (HB)	Continuous	Higher score= Poorer health behavior	0.85
Personal Health Questionnaire (PHQ-9)	Binary	≥ 10 Depressed	0.92
Generalized Anxiety Disorder 7 Item (GAD-7)	Binary	≥ 10 Anxious	0.93
Decision Regret Scale (DRS)	Binary	1= Regret	0.80
Genetic Counseling Satisfaction Scale (GCSS)	Binary	1= Satisfied with genetic counseling	0.91
Adapted* Multidimensional Impact of Cancer Risk Assessment (aMICRA)	Continuous	Higher score = Greater impact of results	0.89
aMICRA Uncertainty subdomain	Continuous	Higher score = More uncertainty	
aMICRA Distress subdomain	Continuous	Higher score = More distress	
aMICRA Positivity subdomain	Continuous	Higher score = Lower positivity	

Table 2. Interpretation of exome sequencing results. Clinician interpretation compared to laboratory test report. Parental interpretation of child's results compared to clinician interpretation. N and column %.

	Laboratory Report of Exome Sequencing Results						
Clinician	**Positive**		**Uncertain**		**Negative**		**Total**
Interpretation	**N**	**%**	**N**	**%**	**N**	**%**	**N**
Positive	63	95%	16	31%	0	0%	79
Uncertain	3	5%	26	51%	6	8%	35
Negative	0	0%	9	18%	69	92%	78
Total	66		51		75		192

	Clinician Interpretation of Exome Sequencing Results						
Parental	**Positive**		**Uncertain**		**Negative**		**Total**
Interpretation	**N**	**%**	**N**	**%**	**N**	**%**	**N**
Positive	70	89%	4	11%	2	3%	76
Uncertain	8	10%	25	71%	13	17%	46
Negative	1	1%	5	14%	56	72%	62
Blank	0	0%	1	3%	7	9%	8
Total	79		35		78		192

Table S3. Parental report of process of decision to have CES and receiving results. N, %

Which medical professionals did you speak with to learn about CES and make the decision to have CES for your child?		
Geneticist	142	74%
Genetic Counselor	113	59%
Pediatrician	6	3%
Primary Care Provider	32	17%
Cardiologist	18	9%
Nurse	25	13%
Oncologist	8	4%
Neurologist	7	4%
Other*	48	25%

* Dermatologist, obstetrician, orthopedist, heptologist, ophthalmologist, rheumatologist, pulmonologist

What other resources outside of your doctors or health care providers did you use to learn more about CES		
Internet search	38	20%
Educational Materials	11	6%
Educational Websites	102	53%
Friends	20	10%
Book	8	4%
Educational Videos	4	2%
Other*	9	5%

*Relative/ friend in medical or science industry, parent support group, Jewish Genetics organization (Bonei Olam)

How much time did you spend discussing CES testing with your child's health care provider(s) before deciding to have CES for your child		
<15 minutes	31	16%
15-30 minutes	50	26%
30-60 minutes	45	23%
>1 hour	39	20%
Cannot remember	23	12%

How much money did you personally have to pay out of pocket to have CES for your child?		
No cost	119	62%
<$100	18	9%
$100-$1000	19	10%
>$1000	9	5%
Cannot remember	18	9%

Before your child had CES, had he/she had any other genetic testing for the medical condition?		
Yes	118	61%
No	62	32%
Cannot remember	12	6%

How did you FIRST learn about the CES test results?		
Phone	103	54%
In person	13	7%
Letter	30	16%
Cannot remember	45	23%

Abbreviations: Clinical Exome Sequencing (CES)

Table S4a. Frequency of parent incorrectly interpreting child's CES reuslts by categorical variables of interpretation of results, demographics, testing experience and parental psychology.

Variable	**Category**	**% Incorrect**	**p-value**
All		**21%**	
Parental Intp. of CES Results	Positive	11%	**0.02**
	Uncertain	29%	
	Negative	28%	
Parental Gender	Male	25%	0.64
	Female	21%	
Child Gender	Male	24%	0.36
	Female	18%	
Marital status	Married	22%	0.90
	Unmarried	23%	
Race/ Ethnicity	Non-White	21%	0.99
	White	21%	
Education	< College	24%	0.59
	≥ College	25%	
Employment	Employed	20%	0.70
	Unemployed	23%	
Medical Geneticist	MD 1	22%	0.59
	MD 2	18%	
	Others	37%	
Genetic Counselor Experience*	≥ 5yr	22%	0.72
	< 5yr	19%	
Cost	No cost	16%	0.05
	Cost	28%	
Location	Clinic	20%	0.56
	In patient	24%	
Results Disclosure	Phone	22%	0.75
	Other	20%	
Indication	Single System	22%	0.89
	Multiple Systems	18%	
	Neurological	22%	
Decision Regret Scale (DRS)	Regret	31%	0.12
	No Regret	24%	
Genetic Counseling Satisfaction Scale (GCSS)*	Regret	24%	0.48
	No Regret	19%	
Personal Health Questionnaire (PHQ-9)	Not Depressed	18%	0.14
	Depressed	32%	
Generalized Anxiety Disorder 7 Item (GAD-7)	No Anxiety	18%	0.07
	Anxiety	33%	
Genetic Knowledge	Not All Correct	22%	0.86
	All Correct	21%	
Exome Sequencing Knowledge	Not All Correct	24%	0.46
	All Correct	19%	

p-values generated by chi squared analysis

Abbreviations: Clinical Exome Sequencing (CES)

*Analysis excludes in patients

Table S4b. Mean values of continuous variables stratified by those parents who had correct and incorrect interpretation of child's exome sequencing results.

		Incorrect		**Correct**		
Variable	**Interpretation**	**Mean**	**SD**	**Mean**	**SD**	**p-value**
Parental Age in years		40.9	9.1	40.0	9.0	0.45
Child Age in years		7.3	6.2	6.7	5.4	0.56
Time from disclosure		16.4	9.4	16.5	8.8	0.95
Internal Health Locus of Control (IHLC)	higher=more control	13.5	3.6	12.2	3.0	**0.03**
Healthy Behaviors (HB)	higher= more healthy behaviors	21.8	4.3	24.4	4.4	**0.001**
Negative Experiences	higher= more negative experiences	1.6	1.7	1.2	1.7	0.21
Positive Experiences	higher=more positive experiences	1.9	2.1	3.2	2.8	**0.007**
Non-Healthcare Utility	higher=more non-healthcare utility	0.5	0.7	0.6	1.0	0.40
Healthcare Utility	higher=more health utility	1.2	0.9	1.3	0.9	0.58

p-values generated by two sample t-test

Abbreviations: Standard Deviation (SD), Exome Sequencing (ES)

Table S5a. Frequency of reported satisfaction with genetic counseling by categorical variables of parental interpretation of results, demographics, testing experience and parental psychology. Analysis includes only parents of children who were evaluated in the clinic.

Variable	**Category**	**% Satisfied**	**p-value**
All		**68%**	
Parental Intp. of CES Results	Positive	68%	0.33
	Uncertain	60%	
	Negative	76%	
Parental Misinterpreation of CES Results	Correct	69%	0.48
	Incorrect	62%	
Parental Gender	Male	69%	0.92
	Female	67%	
Child Gender	Male	58%	**0.005**
	Female	80%	
Marital status	Married	64%	0.37
	Unmarried	80%	
Race/ Ethnicity	Non-White	70%	0.72
	White	67%	
Education	< College	73%	0.42
	≥ College	66%	
Employment	Employed	64%	0.22
	Unemployed	74%	
Medical Geneticist	MD 1	65%	0.61
	MD 2	69%	
	Others	79%	
Genetic Counselor Experience	≥ 5yr	76%	**0.024**
	< 5yr	58%	
Cost	No cost	65%	0.45
	Cost	71%	
Results Disclosure	Phone	63%	0.28
	Other	72%	
Indication	Single System	77%	0.34
	Multiple Systems	63%	
	Neurological	74%	
Decision Regret Scale (DRS)	Regret	50%	**0.012**
	No Regret	74%	
Personal Health Questionnaire (PHQ-9)	Not Depressed	68%	0.81
	Depressed	71%	
Generalized Anxiety Disorder 7 Item (GAD-7)	No Anxiety	68%	1.00
	Anxiety	68%	
Genetic Knowledge	Not All Correct	71%	0.29
	All Correct	62%	
Exome Sequencing Knowledge	Not All Correct	71%	0.40
	All Correct	64%	

p-values generated by chi squared analysis

Abbreviations: Clinical Exome Sequencing (CES)

*Analysis excludes inpatients

Table S5b. Mean values of continuous variables stratified by those satisfied and not satisfied with genetic counseling experience.

		Satisfied		**Not Satisfied**		
		Mean	**SD**	**Mean**	**SD**	**p-value**
Parental Age in years		39.4	7.3	41.1	9.0	0.15
Child Age in years		6.2	5.1	7.8	6.0	0.06
Time from disclosure		17.1	9.1	17.8	8.7	0.67
Internal Health Locus of Control (IHLC)	higher=more control	12.5	3.3	12.6	3.0	0.73
Healthy Behaviors (HB)	higher= more healthy behaviors	24.7	4.3	22.8	4.4	**0.006**
Negative Experiences	higher= more negative experiences	1.2	1.7	1.4	1.3	0.47
Positive Experiences	higher=more positive experiences	3.4	2.8	2.2	2.3	**0.005**
Healthcare Utility	higher=more health utility	1.3	0.9	1.2	0.8	0.28
Non-Healthcare Utility	higher=more non-healthcare utility	0.5	0.9	0.8	1.2	**0.045**

p-values generated by two sample t-test

Abbreviations: Standard Deviation (SD), Clinical Exome Sequencing (CES)

Table S6a. Frequency of reported regret with decision (DSR) to have CES by categorical variables of parental interpretation of results, demographics, testing experience and parental psychology.

Variable	Category	% Regret	p-value
All		**21%**	
Parental Intp. of CES Results	Positive	15%	0.20
	Uncertain	15%	
	Negative	27%	
Parental Misinterpreation of CES Results	Male	19%	0.80
	Female	21%	
Child Gender	Male	26%	0.13
	Female	16%	
Marital status	Married	23%	0.34
	Unmarried	14%	
Race/ Ethnicity	Non-White	28%	0.15
	White	18%	
Education	< College	29%	0.09
	≥ College	18%	
Employment	Employed	20%	0.61
	Unemployed	23%	
Medical Geneticist	MD1	24%	0.80
	MD2	36%	
	Others	19%	
Genetic Counselor Experience*	≥ 5yr	16%	**0.024**
	< 5yr	32%	
Cost	No cost	19%	0.50
	Cost	23%	
Location	Clinic	25%	0.21
	In patient	15%	
Results Disclosure	Phone	25%	0.16
	Other	17%	
Indication	Single System	28%	0.50
	Multiple Systems	17%	
	Neurological	21%	
Genetic Counseling Satisfaction Scale (GCSS)*	Regret	32%	**0.012**
	No Regret	17%	
Personal Health Questionnaire (PHQ-9)	Not Depressed	17%	**0.017**
	Depressed	24%	
Generalized Anxiety Disorder 7 Item (GAD-7)	No Anxiety	20%	0.79
	Anxiety	23%	
Genetic Knowledge	Not All Correct	22%	0.47
	All Correct	18%	
Exome Sequencing Knowledge	Not All Correct	21%	0.59
	All Correct	21%	

p-values generated by chi squared analysis

Abbreviations: Clinical Exome Sequencing (CES)

*Analysis excludes inpatients

Table S6b. Mean values of continuous variables stratified by those with and with out regret about the decision to have WES.

		No Regret		Regret		
Variable	**Interpretation**	**Mean**	**SD**	**Mean**	**SD**	**p-value**
Parental Age in years		40.1	8.0	38.6	7.8	0.31
Child Age in years		6.8	5.6	6.9	5.2	1.00
Time from disclosure		16.7	9.2	16.4	21.7	0.93
Internal Health Locus of Control (IHLC)	higher=more control	12.7	3.1	11.9	3.2	0.16
Healthy Behaviors (HB)	higher= more healthy behaviors	23.8	4.5	24.4	4.7	0.49
Negative Experiences	higher= more negative experiences	1.2	1.4	1.5	2.1	0.32
Positive Experiences	higher=more positive experiences	3.2	2.8	1.7	2.2	**0.002**
Non-Healthcare Utility	higher=more non-healthcare utility	0.6	1.1	0.4	0.8	0.45
Healthcare Utility	higher=more health utility	1.3	0.9	1.2	0.9	0.17

p-values generated by two sample t-test

Abbreviations: Standard Deviation (SD), Clinical Exome Sequencing (CES)

Table S7a. aMICRA uncertainty subdomain scores (outcome) and parental interpretation of child CES results (primary predictor) and other demographic and parental experience variables. ANOVA for categorical predictors, dummy reference variable created for predictor variables with three categories, two sample t-test for continuous predictors.

Variable	Reference	Estimate	95% CI U	95% CI L	p-value	N
Parental Intp. of CES Results	Pos vs Neg	5.27	1.92	8.63	**0.002**	176
	Unc vs Neg	3.72	-0.09	7.53	0.06	
	Pos vs Unc	1.56	-2.12	5.23	0.40	
Parental Misinterpreation of CES Results	Incorrect vs Correct	2.5	-1.3	6.2	0.19	181
Parental Gender	Male vs Female	1.87	-2.74	6.47	0.42	183
Child Gender	Male vs Female	1.90	-1.16	4.96	0.22	183
Parental Age in years		0.19	-0.16	0.53	0.29	181
Child Age in years		-0.27	-0.80	0.26	0.32	183
Marital status	Married vs Unmarried	2.73	-1.88	7.33	0.24	172
Race/ Ethnicity	Non-White vs White	0.49	-2.79	3.76	0.77	183
Education	< College vs ≥ College	-1.12	-4.45	2.21	0.51	183
Employment	Employed vs Unemployed	1.21	-1.97	4.39	0.45	183
Time in months between results returned and survey		-0.02	-0.20	0.149	0.78	181
Medical Geneticist	MD1 vs All	-0.08	-4.73	4.57	0.97	176
	MD2 vs All	-0.54	-5.38	4.29	0.83	
Genetic Counselor experience*	≥ 5yr vs <5yr	-1.05	-4.73	2.63	0.57	143
Reported cost	No cost vs cost	-1.98	-5.25	1.28	0.23	175
Reported method of results disclosure	Phone vs other	-0.99	-4.05	2.08	0.53	183
Location	In Patient vs Clinic	-2.04	-5.72	1.65	0.28	183
Indication	Single system vs Neurological	-2.22	-6.39	1.95	0.29	183
	Multiple systems vs Neurological	-2.45	-6.38	1.49	0.22	
	Single system vs Multiple systems	0.22	-4.83	5.28	0.93	183
Decision Regret Scale (DRS)	No Regret vs Regret	-2.25	-6.05	1.54	0.24	177
Genetic Counseling Satisfaction Scale (GCSS)*	Not Satisfied vs Satisfied	-0.26	-4.21	3.68	0.90	139
Personal Health Questionnaire (PHQ-9)	Not Depressed vs Depressed	-14.07	**-18.21**	-9.92	**<.0001**	174
Generalized Anxiety Disorder 7 Item (GAD-7)	No Anxiety vs Anxiety	-12.68	**-16.95**	-8.41	**<.0001**	174
Genetic Knowledge	Not All Correct vs All Correct	-0.64	-3.99	2.72	0.71	182
Exome Sequencing Knowledge	Not All Correct vs All Correct	-1.05	-4.12	2.02	0.50	182
Internal Health Locus of Control (IHLC)	higher=more control	0.17	-0.31	0.66	0.48	177
Healthy Behaviors (HB)	higher= more healthy behaviors	-0.72	-1.04	-0.39	**<.0001**	180

Abbreviations: Adapted Multidimensional Impact of Cancer Risk Assessment (aMICRA), Clinical Exome Sequencing (CES), 95% Confidence Interval Upper and Lower (95% CI U, L).

p-values from ANOVA for categorical predictors, dummy reference variable created for predictor variables with three categories, two sample t-test for continuous predictors.

**Analysis excludes in patients

Table S7b. aMICRA distress subdomain scores (outcome) and parental interpretation of child CES results (primary predictor) and other demographic and parental experience variables. ANOVA for categorical predictors, dummy reference variable created for predictor variables with three categories, two sample t-test for continuous predictors.

Variable	**Reference**	**Estimate**	**95% CI U**	**95% CI L**	**p-value**	**N**
Parental Intp. of CES Results	Pos vs Neg	-0.05	-1.96	1.86	0.96	177
	Unc vs Neg	1.14	-1.04	3.31	0.30	
	Pos vs Unc	-1.19	-3.28	0.90	0.26	
Parental Misinterpreation of CES Results	Incorrect vs Correct	1.2	-0.8	3.2	0.24	184
Parental Gender	Male vs Female	-1.36	-5.04	2.32	0.47	188
Child Gender	Male vs Female	0.81	-1.66	3.28	0.52	188
Parental Age in years		0.01	-0.14	0.16	0.90	188
Child Age in years		-0.34	-0.57	-0.11	**0.005**	173
Marital status	Married vs Unmarried	2.61	-1.21	6.42	0.18	177
Race/ Ethnicity	Non-White vs White	-1.88	-4.52	0.75	0.16	187
Education	< College vs ≥ College	-0.55	-3.21	2.11	0.68	188
Employment	Employed vs Unemployed	0.29	-2.27	2.84	0.83	188
Time in months between results returned and survey		-0.01	-0.22	0.83	0.05	188
Medical Geneticist	MD1 vs All	0.86	-2.95	4.68	0.66	181
	MD2 vs All	2.71	-1.24	6.66	0.18	
Genetic Counselor experience*	≥ 5yr vs <5yr	-1.83	-4.63	0.98	0.20	145
Reported cost	No cost vs cost	-0.78	-3.45	1.89	0.57	179
Reported method of results disclosure	Phone vs other	-3.29	-5.71	-0.87	0.01	187
Location	In Patient vs Clinic	0.67	-2.26	3.59	0.65	188
Indication	Single system vs Neurological	-0.78	-4.14	2.58	0.65	188
	Multiple systems vs Neurological	-0.33	-3.51	2.85	0.84	
	Single system vs Multiple systems	-0.45	-4.53	3.62	0.83	
Decision Regret Scale (DRS)	No Regret vs Regret	-0.23	-3.30	2.83	0.88	181
Genetic Counseling Satisfaction Scale (GCSS)*	Not Satisfied vs Satisfied	0.57	-2.48	3.63	0.71	140
Personal Health Questionnaire (PHQ-9)	Not Depressed vs Depressed	-12.16	-15.54	-8.77	**<.0001**	179
Generalized Anxiety Disorder 7 Item (GAD-7)	No Anxiety vs Anxiety	-9.89	-13.33	-6.46	**<.0001**	181
Genetic Knowledge	Not All Correct vs All Correct	-0.81	-3.50	1.88	0.55	187
Exome Sequencing Knowledge	Not All Correct vs All Correct	-1.82	-4.28	0.63	0.15	187
Internal Health Locus of Control (IHLC)	higher=more control	0.40	0.01	0.80	0.05	181
Healthy Behaviors (HB)	higher= more healthy behaviors	-0.64	-0.90	-0.38	**<.0001**	185

Abbreviations: Adapted Multidimensional Impact of Cancer Risk Assessment (aMICRA), Clinical Exome Sequencing (CES), 95% Confidence Interval Upper and Lower (95% CI U, L).

p-values from ANOVA for categorical predictors, dummy reference variable created for predictor variables with three categories, two sample t-test for continuous predictors.

**Analysis excludes in patients

Table S7c. aMICRA positivity subdomain scores (outcome) and parental interpretation of child CES results (primary predictor) and other demographic and parental experience variables. ANOVA for categorical predictors, dummy reference variable created for predictor variables with three categories, two sample t-test for continuous predictors.

Variable	**Reference**	**Estimate**	**95% CI U**	**95% CI L**	**p-value**	**N**
Parental Intp. of CES Results	Pos vs Neg	-0.05	-1.96	1.86	0.96	177
	Unc vs Neg	1.14	-1.04	3.31	0.30	
	Pos vs Unc	-1.19	-3.28	0.90	0.26	
Parental Misinterpreation of CES Results	Incorrect vs Correct	1.2	-0.8	3.2	0.24	184
Parental Gender	Male vs Female	1.04	-1.42	3.51	0.40	184
Child Gender	Male vs Female	1.01	-0.66	2.67	0.23	184
Parental Age in years		0.07	-0.04	0.17	0.20	184
Child Age in years		0.19	0.03	0.35	**0.017**	169
Marital status	Married vs Unmarried	-0.97	-3.57	1.62	0.46	174
Race/ Ethnicity	Non-White vs White	0.56	-1.22	2.35	0.53	184
Education	< College vs ≥ College	1.12	-0.69	2.92	0.22	184
Employment	Employed vs Unemployed	-0.72	-2.45	1.01	0.41	184
Time in months between results returned and survey		0.03	-0.07	0.12	0.59	184
Medical Geneticist	MD1 vs All	1.25	-1.31	3.81	0.34	177
	MD2 vs All	1.63	-1.03	4.29	0.23	
Genetic Counselor experience*	≥ 5yr vs <5yr	0.71	-1.16	2.58	0.45	143
Reported cost	No cost vs cost	0.12	-1.67	1.91	0.90	176
Reported method of results disclosure	Phone vs other	-0.62	-2.62	1.37	0.54	184
Location	In Patient vs Clinic	0.72	-0.95	2.39	0.40	183
Indication	Single system vs Neurological	2.60	0.38	4.83	0.02	184
	Multiple systems vs Neurological	0.43	-1.68	2.54	0.69	
	Single system vs Multiple systems	2.17	-0.52	4.86	0.11	
Decision Regret Scale (DRS)	No Regret vs Regret	-3.90	-5.92	-1.89	**0.0002**	178
Genetic Counseling Satisfaction Scale (GCSS)*	Not Satisfied vs Satisfied	2.02	-0.02	4.06	0.05	138
Personal Health Questionnaire (PHQ-9)	Not Depressed vs Depressed	-2.67	-5.26	-0.08	**0.044**	174
Generalized Anxiety Disorder 7 Item (GAD-7)	No Anxiety vs Anxiety	-2.04	-4.53	0.45	0.11	177
Genetic Knowledge	Not All Correct vs All Correct	0.40	-1.41	2.21	0.66	183
Exome Sequencing Knowledge	Not All Correct vs All Correct	1.21	-0.44	2.86	0.15	183
Internal Health Locus of Control (IHLC)	higher=more control	-0.14	-0.41	0.12	0.28	178
Healthy Behaviors (HB)	higher= more healthy behaviors	-0.15	-0.34	0.03	0.10	181

Abbreviations: Adapted Multidimensional Impact of Cancer Risk Assessment (aMICRA), Clinical Exome Sequencing (CES), 95% Confidence Interval Upper and Lower (95% CI U, L).

p-values from ANOVA for categorical predictors, dummy reference variable created for predictor variables with three categories, two sample t-test for continuous predictors.

**Analysis excludes in patients

SUPPLEMENTARY METHODS

Study Instrument

The survey went through four revisions, incorporating feedback from three additional GCs, a genetic research coordinator and two psychologists before the start of the study. The survey included demographic questions as well as questions about the process of testing, including counselling and resources utilized, cost of the test, and time needed to make a decision. Questions regarding emotions experienced at the time of results disclosure, parental perceived medical and non-medical utility of results, with whom results were shared, positive and negative experiences, and genetic knowledge were developed for this study. The parent's interpretation of the child's clinical exome sequencing (CES) results was assessed by a single multiple-choice question. Measures previously validated in multiple adult patient populations were used to assess respondent's psychological state including depression (Personal Health Questionnaire-9 (PHQ-9))(1) and anxiety (Generalized Anxiety Disorder 7 Item (GAD-7))(2) . Health perception and behaviors were measures with the validated instrument internal health locus of control (IHLC)) (3) and a scale that was developed for healthy behaviors (HB). Regret about the decision to have CES and satisfaction with genetic counseling were assessed with validated measures: decision regret scale (DRS) (4) and genetic counseling satisfaction scale (GCSS)(5). The psychological impact of genetic test results was evaluated via an adapted version of the multidimensional impact of cancer risk assessment (aMICRA) to assess CES test results with subdomains of positivity (reverse scored), uncertainty and distress experienced related to results (6) (Table S1). This MICRA scale was developed and validated in a population of women undergoing breast cancer genetic testing, it has not been validated in other populations. The survey took 20 minutes on average to complete.

Statistical analysis

Descriptive statistics are presented in frequencies for categorical variables and means and standard deviations for quantitative variables. Associations between parental interpretations of child's CES results and binary variables were assessed through Chi-square tests. Results for the DRS and GCSS scales were skewed and therefore were analyzed as binary outcomes: DRS: no regret (for any of the questions) versus some regret (for any of the questions), GCSS: completely satisfied (for all questions) versus some dissatisfaction (for any of the questions). The GCSS outcome was assessed only for patients who were seen in clinic and therefore interacted with a GC in the pre-test session. Associations between parental interpretation of their child's test results and continuous variables were assessed by ANOVA.

A multiple linear regression model of the association between parental interpretation of child's CES results and aMICRA score was fit, adjusting for confounders. A variable was considered a potential confounder if it was not theorized to be in the causal pathway between parental interpretation of the child's results (exposure) and aMICRA score (outcome), and was associated with both the exposure and the outcome at a p-value threshold of 0.2.

Analysis was completed in duplicate excluding the 77 parents who completed the survey within 12 months of receiving their child's CES results. The results were similar and for simplicity only the analysis of the full cohort is shown.

The analysis was completed including the 54 fathers from the paired parents using a generalized estimating equation to adjust for paired parent response correlations. The results were similar and for simplicity only the analysis without the 54 fathers is presented.

A p-value of 0.05 was considered relevant for our exploratory analysis. Statistical analysis was completed in SAS (7).

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Figure S1: Flow diagram of participant enrollment. Final enrollment rate 57%

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Enter your study ID (provided in the email):_______________

What is your date of birth? __ __/__ __/__ __ __ ___ (mm/dd/yyyy)

What is your gender?

- ○ Male
- ○ Female
- ○ Other, please specify ____________________

What is your marital status?

- ○ Never married
- ○ Married or living as married with partner
- ○ Separated
- ○ Divorced
- ○ Widowed

Are you of Hispanic, Latino or Spanish origin?

- ○ Yes
- ○ No

Which of the following best describes your race? (check all that apply)

- ☐ White
- ☐ Black or African American
- ☐ Asian
- ☐ American Indian or Alaska Native
- ☐ Native Hawaiian or other Pacific Islander
- ☐ More than one race
- ☐ Other race, please specify ____________________

Counting yourself, how many members currently make up your household?: ___________

What is the highest level of education you completed?

- ○ Less than 9th grade
- ○ Some high school but did not graduate
- ○ Graduated high school or completed GED
- ○ Some post high school training including vocational, associate degree of some college
- ○ College degree
- ○ Graduate degree (Master's degree or Doctoral degree)

Which of the following best describes your current employment status, main daily activities and/or responsibilities?

- ○ Employed full-time (including full-time self-employment)
- ○ Employed part-time (including part-time self-employment)
- ○ Keeping house or raising children full-time
- ○ Unemployed
- ○ Retired
- ○ Disabled
- ○ Other, please specify ____________________

What is the medical condition that first brought your child (your family) to genetics?:

____

For the remainder of this survey, we will refer to this as the <u>medical condition</u> when asking you questions about it and your experience in genetics.

What do you think is the cause of the medical condition? (check all that apply)

- ☐ A mutation in a gene/ a genetic cause
- ☐ Medication exposure (either a medication your child took or one your child was exposed to during the pregnancy)
- ☐ Environmental exposure (either an exposure your child had or one your child was exposed to during the pregnancy)
- ☐ Infection (either an infection your child had or one your child was exposed to during the pregnancy)
- ☐ My fault – poor care of my child's health
- ☐ Stress
- ☐ Difficult delivery, not enough oxygen to the brain at the time of my child's birth
- ☐ Bad luck
- ☐ I don't know

How many children do you have with this medical condition (currently living or deceased)? ______________________

How many children do you have without this medical condition (currently living or deceased)? ___________________

If you have any children without this medical condition, how likely is it that they will develop this medical condition?

- ○ Very unlikely
- ○ Unlikely
- ○ Somewhat unlikely
- ○ Undecided
- ○ Somewhat likely
- ○ Likely
- ○ Very likely
- ○ N/A, I don't have any children without this medical condition

If you decide to have more children, how likely it is that the child will have or develop this medical condition?

- ○ Very unlikely
- ○ Unlikely
- ○ Somewhat unlikely
- ○ Undecided
- ○ Somewhat likely
- ○ Likely
- ○ Very likely

Do you have this medical condition?

- ○ Yes
- ○ No
- ○ Uncertain

If you do not have the medical condition, how likely are you to develop this medical condition in the future?

- ○ Very unlikely
- ○ Unlikely
- ○ Somewhat unlikely
- ○ Undecided
- ○ Somewhat likely
- ○ Likely
- ○ Very likely

Your child had a genetic test called whole exome sequencing (WES) to try to identify the genetic cause of his/her medical condition. Please think back to when you first learned about WES. Think about your experiences during this time to answer the questions for this section.

Before your child had WES, had he/she had any other genetic testing for the medical condition?
- ○ Yes
- ○ No
- ○ I cannot remember

Which medical professionals did you speak with to learn about WES and make the decision to have the test for your child? (check all that apply)
- ☐ Clinical geneticist (a genetics doctor)
- ☐ Genetic counselor
- ☐ Nurse
- ☐ Pediatrician
- ☐ Primary care doctor/ general practitioner
- ☐ Cardiologist
- ☐ Oncologist
- ☐ Other, please specify ____________________

How did you communicate with these medical professionals? (check all that apply)
- ☐ In person, at an appointment
- ☐ By email
- ☐ By phone

Did you have more than one appointment with your geneticist or genetic counselor to discuss WES before you decided to have testing for your child?
- ○ No, I had one appointment
- ○ Yes, I had two appointments
- ○ Yes, I had more than two appointments
- ○ N/A, I did not have an appointment with a geneticist or genetic counselor
- ○ I cannot remember

What other resources outside of your child's doctors or healthcare providers did you use to learn more about WES? (check all that apply)
- ☐ Educational materials provided by the genetic counselor/ medical professionals
- ☐ Educational videos provided by the genetic counselor/ medical professionals
- ☐ Internet searches
- ☐ Educational websites
- ☐ Magazine articles
- ☐ TV
- ☐ Books
- ☐ Friends/ Family
- ☐ Other, please specify ____________________

When did you decide to pursue WES for your child?
- ○ The same day the test was offered to me.
- ○ I took several (2-6) days before deciding to pursue the test.
- ○ I took longer than a week but less than a month before deciding to pursue the test.
- ○ I took more than a month before deciding to pursue the test.
- ○ I took several months or longer before deciding to pursue the test.
- ○ I cannot remember.

How much time did you spend discussing WES testing with your health care provider(s) (doctor, nurse, and genetic counselor) before you decided to have the test for your child?
Please think of all the time that was spent even if it was over several appointments or in an appointment and by phone or email.

- ○ Less than 15 minutes
- ○ 15-30 minutes
- ○ 30-60 minutes
- ○ 60-90 minutes
- ○ 90-120 minutes
- ○ More than 2 hours
- ○ I cannot remember

How much money did you personally have to pay out of pocket to have WES?

- ○ $0, the cost was covered entirely by my insurance
- ○ More than $0, but less than or equal to $100
- ○ More than $100, but less than or equal to $500
- ○ More than $500, but less than or equal to $1000
- ○ More than $1000
- ○ I had to pay some money but I cannot recall how much

Please think about when you received the results of your child's WES and the information you learned about your child's medical condition when answering this next set of questions.

What were the results of your child's WES testing?

- ○ Positive test results. A genetic mutation that is the cause of my child's medical condition was identified.
- ○ Negative genetic test results. No genetic mutations were identified but there still could be a genetic mutation that causes my child's medical condition.
- ○ Negative genetic test results. No genetic mutation was identified and this means that my child's medical condition is not genetic.
- ○ Uncertain genetic test results. A genetic variant was identified but it is not certain whether or not the genetic variant is the cause of my child's medical condition.
- ○ The results of the genetic testing are not clear to me.

What was your reaction when you received the results of the WES? (check all that apply)

- ☐ Happy
- ☐ Content
- ☐ Surprised
- ☐ Relieved
- ☐ Guilty
- ☐ Depressed
- ☐ Worried
- ☐ Excited
- ☐ Curious
- ☐ Fearful
- ☐ Nervous/ Anxious
- ☐ Hopeful
- ☐ Frustrated
- ☐ Confused
- ☐ Other, please specify ____________________

How did you FIRST learn about the WES test results?

- ○ By phone
- ○ In a follow up visit
- ○ In a letter
- ○ I do not remember
- ○ Other, please specify ____________________

Which health care provider FIRST told you about the WES test results?

- ○ A genetic counselor
- ○ A genetics doctor
- ○ Both a genetic counselor and a genetics doctor
- ○ I do not remember
- ○ Other, please specify ____________________

What other resources other than your doctors or healthcare providers did you use to learn more about the WES results? Please indicate the helpfulness of each of these resources.

	N/A, did not use this resource	Very Helpful	Somewhat Helpful	Helpful	Not Helpful
Educational materials provided by the genetic counselor/ medical professionals	○	○	○	○	○
Educational videos provided by the genetic counselor/ medical professionals	○	○	○	○	○
Internet searches	○	○	○	○	○
Educational websites	○	○	○	○	○
Patient support groups	○	○	○	○	○
Facebook	○	○	○	○	○
Friends/ Family	○	○	○	○	○

Please answer the questions below about you experience with your genetic counselor. Please read each statement below carefully and tell us how much you agree or disagree with each statement by indicating strongly agree to strongly disagree.

	Strongly agree	Agree somewhat	Uncertain	Disagree somewhat	Strongly disagree
My genetic counselor seemed to understand the stress I was facing.	○	○	○	○	○
My genetic counselor helped me to identify what I needed to know to make decisions about what would happen to me.	○	○	○	○	○
The counseling session was about the right length of time.	○	○	○	○	○
My genetic counselor was truly concerned about my well-being.	○	○	○	○	○
The genetic counseling session was valuable to me.	○	○	○	○	○

Please think about the decision you made to have WES for your child after talking to your healthcare provider (doctor, nurse, health professional, etc.). Please read each statement below and tell us how much you agree or disagree with each statement by indicating strongly agree to strongly disagree.

	Strongly Agree	Agree	Neither Agree nor Disagree	Disagree	Strongly Disagree
It was the right decision.	○	○	○	○	○
I regret the choice that was made.	○	○	○	○	○
I would make the same choice if I had to do it all over again.	○	○	○	○	○
The choice did me a lot of harm.	○	○	○	○	○
The decision was a wise one.	○	○	○	○	○

What was positive about the experience of WES testing? (check all that apply)

- ☐ The results provided an answer for my child’s medical condition.
- ☐ The results provided reassurance that my child’s medical condition is likely not genetic.
- ☐ The results helped my family.
- ☐ The results helped us to determine who else might be at risk for the medical condition.
- ☐ The results provided closure for our family.
- ☐ The results have made me hopeful about possible treatments in the future.
- ☐ The results helped me know what to expect in the future.
- ☐ The results helped me to identify other individuals with the same condition.
- ☐ The results allow me to identify the scientists and doctors who are specialists in this condition.
- ☐ The results allow my child to participate in research on my child’s condition.
- ☐ The results allow me to follow the new scientific developments about my child’s condition.
- ☐ The results allow me to feel more in control.
- ☐ Nothing was positive about the experience.
- ☐ Other, please specify ____________________

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What was negative about the experience of WES testing? (check all that apply)

- ☐ The results have not helped my child.
- ☐ The results have created more questions about my child’s medical condition.
- ☐ I worry about the things I now know could happen in the future.
- ☐ Knowing what the future is likely to hold is depressing.
- ☐ I have had to have difficult conversations with relatives.
- ☐ The results have caused conflict or arguments with my partner.
- ☐ The results have caused conflict or arguments with my other family members.
- ☐ I feel like I have lost privacy.
- ☐ It has negatively affected my child’s medical insurance.
- ☐ It has negatively affected my child’s life insurance.
- ☐ It has negatively affected my child’s ability to get a job.
- ☐ It has negatively affected my child’s to have a spouse or long term partner.
- ☐ Nothing was negative about the experience.
- ☐ Other, please specify ____________________

Have the results of the WES affected your child’s medical care in the any of the follow ways? (check all that apply)

- ☐ My child saw a doctor to discuss the implications of the test results.
- ☐ My child saw a doctor who specializes in this specific condition.
- ☐ My child had an additional medical evaluation or test to determine if my child has features of the medical condition.
- ☐ My child plans to have regular screening for the medical condition.
- ☐ The results have not changed my child’s medical care.
- ☐ Other, please specify ____________________

Will /did the results of WES help to guide your reproductive decisions? (check all that apply)

- ☐ Yes, I now know that my future children are not at risk to have the medical condition.
- ☐ Yes, I was or will be able to pursue testing to prevent having (another) child affected with the medical condition.
- ☐ Yes, I elected not to have (additional) biological children.
- ☐ No, the results did not guide my reproductive decisions.
- ☐ I don't know.
- ☐ Other, please specify ____________________

Please indicate if you shared the WES results with any of the following individuals. (check all that apply)

- ☐ My primary care physician
- ☐ My child's pediatrician
- ☐ My spouse/ partner
- ☐ My children
- ☐ My siblings
- ☐ My extended family
- ☐ My parents
- ☐ My friends
- ☐ My employer
- ☐ My health insurance
- ☐ I did not share my results
- ☐ Other, please specify ____________________

If you shared the results with your family members what was their reaction?

- ☐ N/A, I did not share my child’s results with any family members.
- ☐ All of my family members were grateful to learn about the results.
- ☐ Some of my family members were grateful to learn about the results but some were upset.
- ☐ All of my family members were upset to learn about the results.

Please tell us a little more about your family members’ specific reactions: ________________________

____

Please indicate how the results of WES affected you in ways unrelated to medical management. (check all that apply)

- ☐ I or my child was able to connect with other families/ individuals with the same medical condition online or in a support group.
- ☐ I or my child met another person with the medical condition.
- ☐ I or my child attended a meeting for individuals with the medical condition.
- ☐ I or my child started a foundation/ support group for the medical condition.
- ☐ I learned that my child is the only person known to have the medical condition.
- ☐ I or my child joined a research study.
- ☐ None of the above.

If you could have chosen when in your life to learn about the WES results, when would that have been?

- ○ I would not change when I learned about the results.
- ○ I wish I knew about the results before I had my child(ren).
- ○ I wish I knew about the results immediately after my child was diagnosed with the medical condition.
- ○ I wish I knew about the results as soon as my child became an adult (18 years).
- ○ I wish I never learned about the results.
- ○ Other, please specify when you would have liked to learn about your results. ____________________

If you had known your child's WES results before you had children, would the results have changed your decision to have children?

- ○ No, the results would not have changed my decision to have biological children.
- ○ Yes, I would have chosen not to have biological children.
- ○ Yes, I would have chosen to pursue reproductive options to prevent my children from inheriting the medical condition.

WES Post-Disclosure, Parental (retro)

The questions below are about some specific responses you may have had after receiving your child's WES test results. Please answer every question regardless of whether you were given a positive or negative test result. Please indicate whether you have experienced each statement never, rarely, sometimes, or often, by indicating the corresponding number.

	Never	Rarely	Sometimes	Often
Feeling upset about the test results.	○	○	○	○
Feeling sad about the test results.	○	○	○	○
Feeling anxious or nervous about the test results.	○	○	○	○
Feeling guilty about the test results.	○	○	○	○
Feeling relieved about the test results.	○	○	○	○
Feeling happy about the test results.	○	○	○	○
Feeling a loss of control.	○	○	○	○
Having problems enjoying life because of the test results.	○	○	○	○
Worrying about my risk of getting the medical condition.	○	○	○	○
Being uncertain about what the test results mean about my child's health risk.	○	○	○	○
Being uncertain about what the test results mean for my child(ren) and/or family's risk.	○	○	○	○
Having difficulty making decisions about screening or prevention (e.g. having preventive surgery or getting medical tests done).	○	○	○	○
Understanding clearly my choices for disease prevention or early detection.	○	○	○	○
Feeling frustrated that there are no definite prevention guidelines for me or my children.	○	○	○	○
Thinking about the test results has affected my work or my family life.	○	○	○	○
Feeling concerned about how the test results will affect my child's insurance status.	○	○	○	○
Having difficulty talking about the test results with my family members.	○	○	○	○
Feeling that my family has been supportive during the genetic counseling and testing process.	○	○	○	○
Feeling satisfied with family communication about the genetic test results.	○	○	○	○
Worrying that the genetic counseling and testing process has brought about conflict.	○	○	○	○
Feeling regret about getting the test results.	○	○	○	○
Feeling that the genetic test results have made it harder to cope with my medical condition.	○	○	○	○
Feeling that the genetic test results have made it easier to cope with my child's medical condition.	○	○	○	○
Worry about the possibility of my other children getting the medical condition.	○	○	○	○
Feeling guilty about possibly passing on the disease risk to my child(ren).	○	○	○	○

Please answer the following questions thinking about how you feel.
These symptoms may or may not be related to the WES testing.

Over the last 2 weeks, how often were you bothered by any of the following problems? Please indicate not at all to nearly every day.

	Not at all	Several days	More than half the days	Nearly every day
Little interest or pleasure in doing things.	○	○	○	○
Feeling down, depressed or hopeless.	○	○	○	○
Trouble falling or staying asleep, or sleeping too much.	○	○	○	○
Feeling tired or having little energy.	○	○	○	○
Poor appetite or overeating.	○	○	○	○
Feeling bad about yourself – or that you are failure and have let yourself or your family down.	○	○	○	○
Trouble concentrating on things, such as reading the newspaper or watching television.	○	○	○	○
Moving or speaking so slowly that other people could have noticed, or the opposite being so fidgety or restless that you have been moving around a lot more than usual.	○	○	○	○
Thoughts that you would be better off dead, or of hurting yourself in some way.	○	○	○	○

Over the last 2 weeks, how often were you bothered by any of the following problems? Please indicate not at all to nearly every day.

	Not at all	Several days	More than half the days	Nearly every day
Feeling nervous, anxious or on edge.	○	○	○	○
Not being able to stop or control worrying.	○	○	○	○
Worrying too much about different things.	○	○	○	○
Trouble relaxing.	○	○	○	○
Being so restless that it is hard to sit still.	○	○	○	○
Becoming easily annoyed or irritable.	○	○	○	○
Feeling afraid as if something awful might happen.	○	○	○	○

Do you have a diagnosis of a psychiatric or emotional condition?

○ No
○ Yes
○ No, not currently but have been diagnosed in the past with a psychiatric condition.

What psychiatric or emotional condition do you have or did you have in the past? (check all that apply)

☐ Depression
☐ Anxiety
☐ Bipolar disorder
☐ Schizophrenia
☐ Panic disorder
☐ Attention deficit/ hyperactive disorder (ADHD)
☐ Obsessive compulsive disorder
☐ Eating disorder
☐ Other, please specify ____________________

For each answer, choose what best describes your feelings. Please indicate strongly agree to strongly disagree.

	Strongly agree	Moderately agree	Moderately disagree	Strongly disagree
If I get sick, it is my own behavior which determines how soon I get well again.	○	○	○	○
I am in control of my health.	○	○	○	○
When I get sick, I am to blame.	○	○	○	○
The main thing which affects my health is what I do myself.	○	○	○	○
If I take care of myself, I can avoid illness.	○	○	○	○
If I take the right actions, I can stay healthy.	○	○	○	○

Please tell us about how you care for your health. Please indicate strongly agree to strongly disagree.

	Strongly agree	Moderately agree	Moderately disagree	Strongly disagree
I make sure I get plenty of rest.	○	○	○	○
I make sure I get regular exercise.	○	○	○	○
I try very hard to avoid having too much stress in my life.	○	○	○	○
I make sure I eat a healthy diet.	○	○	○	○
I make sure to get regular medical check-ups.	○	○	○	○
I make sure to follow up on recommendations that my doctors make.	○	○	○	○
I make sure that I take time to relax.	○	○	○	○
I try to keep up with the latest recommendations on how to stay healthy.	○	○	○	○

Please answer each true and false question to the best of your ability.

Fifty percent of inherited genetic information is passed down from each parent.

- ○ True
- ○ False

All individuals with a genetic mutation will eventually develop the genetic disease.

- ○ True
- ○ False

If a woman learned from genetic testing that she does not have a breast cancer gene mutation, then that means that she will never develop breast cancer.

- ○ True
- ○ False

A healthy couple can have a child with a genetic condition.

- ○ True
- ○ False

Whole exome sequencing (WES) is able to detect all genetic mutations.

- ○ True
- ○ False

A negative result for whole exome sequencing (WES) indicates that an individual definitely does not have a genetic condition.

- ○ True
- ○ False

A positive result for whole exome sequencing (WES) indicates that an individual’s medical condition is caused by specific genetic mutation.

- ○ True
- ○ False

When an individual is found to have a genetic condition, his/her relatives may also be at risk for the condition.

- ○ True
- ○ False

Would you be willing to participate in a 60 minute phone interview about your experience with WES?

- ○ Yes
- ○ No

Thank you for taking the time to complete this survey. We will be sending you a $20 amazon gift card to show our appreciation.