Recombine

OUR MISSION

To expand the scope and impact of genomics in medicine.

Through innovative products, dedicated patient care, and collaborative research, Recombine is empowering families and revolutionizing personalized medicine.

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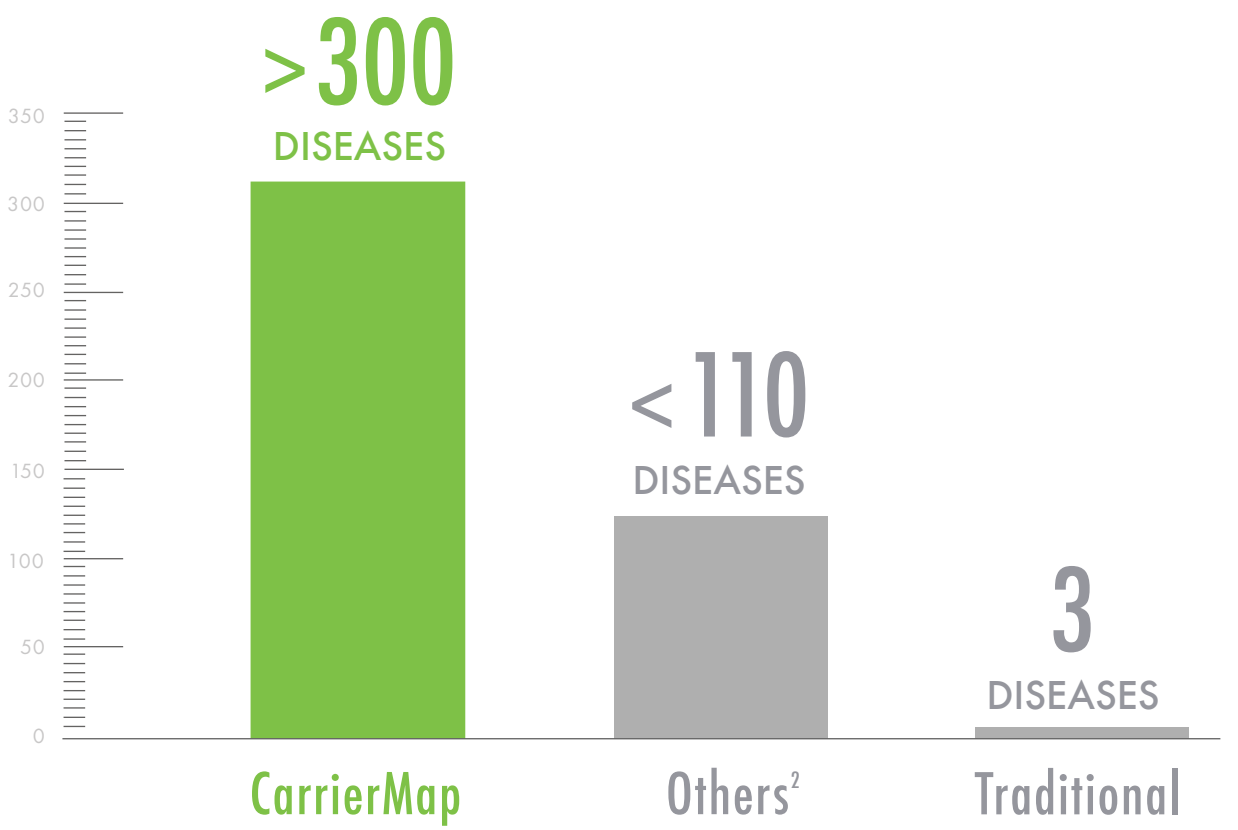
CARRIERMAP

Genetic diseases, though individually rare, are collectively common; thus, assessing carrier status is one of the most important things you can do for your patients. With over 300 single gene disorders,[1] CarrierMap is the most comprehensive carrier screen, covering the widest range of ancestries and providing impactful information for reproductive planning.



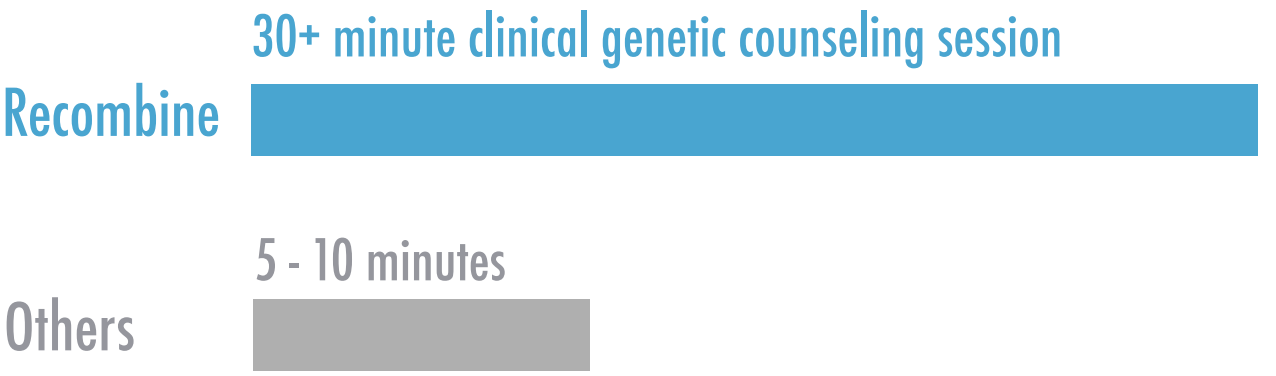
The diseases we screen for have a significant impact and are selected for inclusion based on professional society recommendations.[3]

Physical Impairment	Shortened Lifespan	Cognitive Impairment
290+	185+	145+
Diseases	Diseases	Diseases

[1]For the full list of diseases included on CarrierMap, visit www.recombine.com/diseases.
[2]Data per recent marketing materials.
[3]Edwards JG, Feldman G, Goldberg J, et al. Expanded Carrier Screening in Reproductive Medicine - Points to Consider. Obstet Gynecol. 2015; 125(3):653-662.

GENETIC COUNSELING

Recombine's unique in-depth genetic counseling is available to all patients. Our board-certified genetic counselors[1] carefully prepare for every phone session and explain results in the context of each patient's fertility, family, and medical history. Our sessions are designed based on traditional genetic counseling and facilitate informed decision-making for each patient.



Clinical Counseling Elements	Recombine GC	Others[2]
Pre-test counseling	✓	
Medical & family history	✓	
Education about genetics & inheritance	✓	✓
Overview of testing performed	✓	✓
Results explanation	✓	✓
Risk assessment	✓	
Partner testing & reproductive options	✓	
Consult letter	✓	
Support for physicians	✓	✓

[1]Certified by the American Board of Genetic Counselors (ABGC)
[2]Data per recent marketing materials.

POWERFUL TECHNOLOGY

Cutting-Edge Genotyping

Recombine's genotyping technology is built on Illumina's proven platform, which is used by leading labs worldwide. CarrierMap boasts high disease detection rates due to our careful curation of mutations, including intronic, exonic, and promoter mutations as well as large insertions and deletions often missed by sequencing.

Next-Generation Sequencing

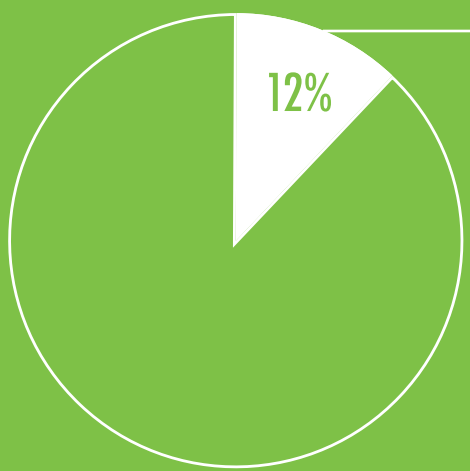
Recombine now offers next-generation sequencing (NGS) for over 300 diseases, enabling detection of novel mutations that may be pathogenic. Our team of experts adheres to the recommendations of the American College of Medical Genetics and Genomics (ACMG) on the interpretation and reporting of genetic variants.

Technological Synergy

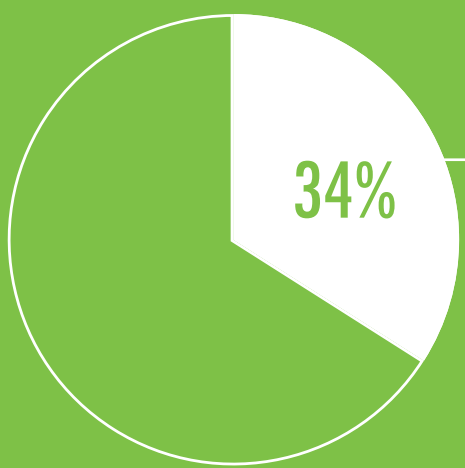
The combination of genotyping and sequencing enables us to better detect the most relevant diseases and mutations, providing the best possible reduction in residual risk.

EXPANDED SCREENING

Limited screening misses two out of every three patients who carry a high-impact disease.[1] Recombine's CarrierMap screens for the most diseases to give your patients the most answers. Expanded screening with CarrierMap empowers individuals with more information and identifies carriers of high-impact diseases that limited screening would miss.



CARRIERS IDENTIFIED BY THE
LIMITED PANEL
OF SOCIETY RECOMMENDED DISEASES



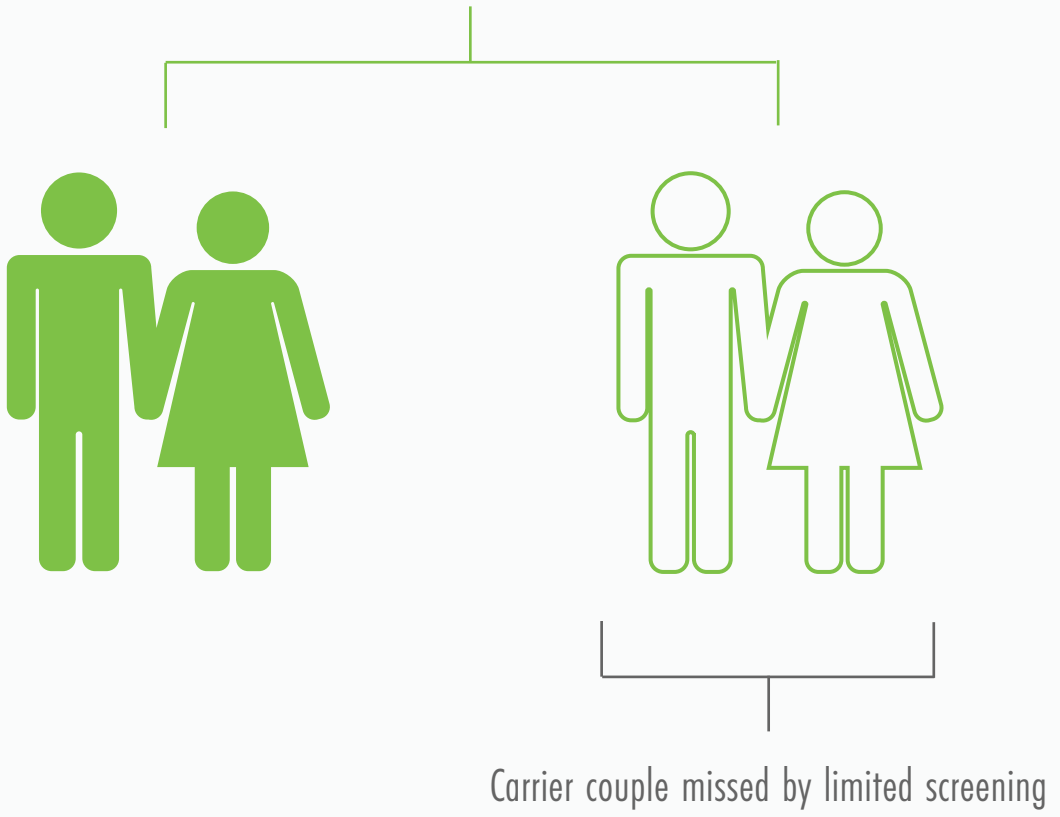
CARRIERS IDENTIFIED BY THE
CARRIERMAP
HIGH-IMPACT PANEL

[1]Internal Recombine data. A limited panel consists only of diseases recommended by professional societies. The CarrierMap high-impact panel consists of diseases that have a significant impact on life expectancy and/or quality of life.

ACTIONABLE ANSWERS

Recombine's CarrierMap identifies 2.1% of couples as carriers for the same autosomal recessive high-impact disease – twice as many when compared to a more limited panel.[1] These couples have a 25% chance of having an affected child. CarrierMap gives couples the power to make more informed reproductive decisions.

ONE OUT OF EVERY TWO COUPLES AT HIGH REPRODUCTIVE RISK ARE MISSED BY THE STANDARD PANEL OF SOCIETY-RECOMMENDED DISEASES



[1] Internal Recombine data. X-Linked diseases are not included in this analysis.

REPRODUCTIVE OPTIONS

Carrier screening enables preimplantation genetic diagnosis (PGD) of embryos. We perform PGD through Reprogenetics for any couple identified as having a high reproductive risk following CarrierMap screening. We are committed to guiding your patients through every step of this process.

CLEAR REPORTS

CarrierMap screens for the most clinically relevant and impactful genetic diseases. Diseases are meticulously selected for inclusion based on carrier rate, clinical severity, and availability of treatment options, as recommended.[1]

HIGH IMPACT

These diseases have a significant impact on life expectancy and/or quality of life.

MODERATE IMPACT

These diseases typically do not affect life expectancy but can affect quality of life.

X-LINKED

These diseases are passed down by female carriers. Carriers may have symptoms.

TREATMENT BENEFITS

Treatment lessens disease symptoms. Newborn screening may be available for timely intervention.

PANEL OPTIONS

Recombine offers various panel options tailored to your clinic's needs.

- CarrierMap Expanded Panel
- ACOG/ACMG Recommended Panel
- Expanded Jewish Panel

[1]Edwards JG, Feldman G, Goldberg J, et al. Expanded Carrier Screening in Reproductive Medicine - Points to Consider. Obstet Gynecol. 2015; 125(3):653-662.

Recombine

CarrierMap

Ordering Practice

Practice Code: 675
Miller MD
374 Broadway
New York, NY 10000
Physician: Dr. Frank Miller
Report Generated: 1/21/2015

Jane Smith

DOB: 02/19/1973
Gender: Female
Ethnicity: European
Procedure ID: 204
Kit Barcode: 20151209229510
Method: Genotyping
Specimen: Saliva, #401
Specimen Collection: 1/12/2015
Specimen Received: 1/13/2015
Specimen Analyzed: 1/21/2015

John Smith

DOB: 01/29/1972
Gender: Male
Ethnicity: European
Procedure ID: 203
Kit Barcode: 20151209229509
Method: Genotyping
Specimen: Saliva, #400
Specimen Collection: 1/12/2015
Specimen Received: 1/13/2015
Specimen Analyzed: 1/21/2015

SUMMARY OF RESULTS

HIGH REPRODUCTIVE RISK

Disease	Jane Smith	John Smith
Cystic Fibrosis ○ High Impact ○ Treatment Benefits	Carrier (1 abnormal copy) Gene: CFTR c.1521_1523delCTT (p.508delF)	Carrier (1 abnormal copy) Gene: CFTR c.G3454C (p.D1152H)
	25% reproductive risk. Genetic counseling is recommended.	
Fragile X Syndrome ○ High Impact ○ X-Linked	Carrier (1 abnormal copy) Gene: FMR1 Intermediate (50 CGG repeats) Wildtype (29 CGG repeats)	Not Applicable
	Females with intermediate-sized expansions are not at increased reproductive risk. Future generations may be at risk. Genetic counseling is recommended.	
Biotinidase Deficiency ○ High Impact ○ Treatment Benefits	No Mutations Detected Gene: BTD	Carrier (1 abnormal copy) Gene: BTD p.Q456H (c.A1368C)
	0.09% (1/1,108) reproductive risk remaining. Genetic counseling is recommended.	

All other mutations analyzed by Recombine were not detected, reducing but not eliminating your chance to be a carrier for the associated genetic diseases. A list of all the diseases and mutations you were screened for is included later in this report. The test does not screen for every possible genetic disease. For disease information, please visit www.recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

♀ Female
Panel CarrierMap Expanded, Diseases Tested 252, Mutations Tested 1970, Genes Tested 243, Null Calls 0

♂ Male
Panel CarrierMap Expanded, Diseases Tested 229, Mutations Tested 1742, Genes Tested 221, Null Calls 0

Assay performed by
Reprogenetics
CLIA ID: 31D1054821
Lab Technician Robert Cohen

Reviewed by Susan Jones, PhD, HCLD, Lab Director

OUR PROMISE

Collaborative Partnership
Empowering your practice

Complete Patient Care
Comprehensive testing and in-depth genetic counseling

Responsible Science
Focused on improving outcomes

Innovative Research
Delving into the science and delivery of genetics

Recombine

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