Electronic Supplementary Material

Journal of Genetic Counseling

Genetic information-seeking behaviors and knowledge among family members and patients with Inherited Bone Marrow Failure Syndromes

Jada G. Hamilton; Sadie P. Hutson; Amy E. Frohnmayer; Paul K. J. Han; June A. Peters; Ann G. Carr & Blanche P. Alter

Correspondence: Please address all correspondence to: Jada G. Hamilton, 641 Lexington Avenue, 7th floor, New York, NY 10022. Phone: 646-888-0049, Fax: 212-888-2584, Email: hamiltoj@mskcc.org.

Online Resource 1.

Responses to measures of general and disorder-specific genetic knowledge among members of families with an IBMFS disorder

	True (%)	False (%)	Don't Know (%)
General Genetic Knowledge Scale Questions			
A. "We are all born with matched pairs of genes, one set from our mother and one set from our father"	**76**	8	16
B. "Genes are made up of DNA"	**81**	4	15
C. "Cells in different body organs have different genes in the nucleus"	22	**33**	45
D. "Mutations occur when the information carried by a gene is damaged in some way"	**73**	4	23
E. "Certain conditions in males can only be inherited from a mother and not from a father"	**58**	11	31
F. "Some genetic diseases occur when a child inherits two abnormal genes, one from each parent"	**80**	3	17
G. "A woman with breast cancer cannot have inherited the abnormal breast cancer gene from her father"	7	**42**	51
H. "If only males can get a certain disease (such as testicular cancer), then the alteration in DNA must have been inherited from the father"	9	**44**	47
I. "Some genetic diseases occur when a child inherits one copy of an abnormal gene from one parent"	**67**	10	23
J. "Mutations in different genes cannot cause the same disorder"	11	**33**	56
K. "Once a mutation for a disorder is identified in a person, the disorder can definitely be cured"	1	**80**	19
L. "A child shares more genes with a father or mother than with a sister or brother"	15	**53**	32
M. "A scientist can look at a person's chromosomes and tell if that person is male or female"	**78**	3	19
N. "An X-linked recessive genetic trait affects females more frequently than males"	16	**30**	54
O. "Carriers of mutations for autosomal recessive and X-linked recessive inherited conditions usually do not have the condition that affects other members of their families"	**27**	10	63
Diamond-Blackfan anemia Genetic Knowledge Scale			
A. "The most common inheritance pattern of DBA is autosomal dominant"	**20**	13	67
B. "The sex of the person with DBA may be an important clue in determining the inheritance pattern of DBA in their family"	10	**43**	48
C. "The RPS19 gene is one of the mutant genes in DBA"	**76**	0	24
D. "All of the children of a DBA gene carrier will have DBA"	0	**81**	19
E. "The physical features of a person with DBA indicate which specific DBA gene is abnormal"	7	**54**	38
F. "Knowing which family members are affected with	**68**	2	29

DBA may be helpful in determining the inheritance pattern in that particular family”			
Dyskeratosis congenita Genetic Knowledge Scale			
A. “The sex of a person with DC may be an important clue in determining the inheritance pattern of DC in their family”	**45**	13	42
B. “The gene called DKC1 is located on the X chromosome”	**46**	3	51
C. “All the children of a person who has a mutation in a DC gene will have DC”	3	**55**	42
D. “The physical features of a person with DC indicate which specific DC gene is abnormal”	11	**37**	52
E. “Knowing which specific family members are affected with DC may be helpful in determining the inheritance pattern in that particular family”	**69**	1	30
Fanconi Anemia Genetic Knowledge Scale			
A. “The most common inheritance pattern of FA is autosomal dominant”	10	**37**	53
B. “The parents of a child with FA have a 75% chance (3 out of 4) of their next child not having FA”	**60**	23	17
C. “Knowledge of the ethnic background of a family may determine which specific FA gene is tested”	**62**	8	30
D. “All of the children of a carrier of FA will have FA”	3	**87**	10
E. “The physical features of a person with FA indicate which specific FA gene is abnormal”	6	**62**	33
F. “Knowledge of the FA patient’s complementation group determines the mutation test that will be performed”	**43**	8	49
Shwachman-Diamond syndrome Genetic Knowledge Scale			
A. “The most common inheritance pattern of SD is autosomal dominant”	9	**41**	50
B. “The parents of a child with SD have a 75% chance (3 out of 4) of their next child not having SD”	**59**	22	19
C. “Knowledge of the ethnic background of a family may determine which specific SD gene is tested”	16	**44**	41
D. “All of the children of a carrier of SD will have SD”	3	**88**	9
E. “The physical features of a person with SD indicate which specific SD gene is abnormal”	6	**75**	19
F. “Knowing which family members are affected with SD may be helpful in determining the inheritance pattern in that particular family”	**47**	16	37

Note. Values for the correct answer are bolded for each question. DBA=Diamond-Blackfan anemia; DC=dyskeratosis congenita; FA=Fanconi anemia; SDS=Shwachman-Diamond syndrome.

Online Resource 2.

Websites visited by members of families with an IBMFS disorder to learn about the genetic condition in their family (n = 141)

Website	*n*
Disease-specific websites (*n* = 103)	
Fanconi Anemia Research Fund	44
Diamond Blackfan Anemia Foundation	13
DBA Yahoo Group	9
SDSF / SDSI	7
National Organization for Rare Disorders	6
diamondblackfananemia.org	5
Dyskeratosis Congenita Outreach	5
Diamond Blackfan Anemia Registry of North America	4
FA Yahoo Group	3
aamac.ca	1
Fanconi Canada	1
marrow.org	1
Raredisorders.com	1
Schwachman-Diamond Syndrome Canada	1
Shwachman-Diamond America	1
turnersyndrome.org	1
Government websites (*n* = 57)	
NIH	39
PubMed	9
NCI	5
marrowfailure.cancer.gov	2
CDC	1
OMIM	1
Medical search websites (*n* = 47)	
WebMD	31
eMedicine / Medscape	6
Medline	4
Genetics Home Reference	2
Patient.co.uk	2
caringbridge.org	1
medical-dictionary.com	1
Generic search websites (*n* = 33)	
Google	10
Wikipedia	10
Yahoo.com	5
Ask Jeeves	3
msn.com	3
iVillage.com	1
Search engines	1
Hospital and medical center websites (*n* = 20)	
Mayoclinic.com	9
Rockefeller.edu	3
bmt.umn.edu	2
cincinnatichildrens.org	2
Children's Medical Center	1
jeffersonhospital.org	1

pennmedicine.org	1
Steven & Alexandra Cohen Children's Medical Center of New York	1
Other (industry/investigator/scientific literature) websites (*n* = 4)	
ambrygen.com	1
Arleen Auerbach's website	1
Blood journal	1
JAMA	1

Note. Of the 304 individual responses provided by participants, 13% could not be identified or categorized (e.g., “various websites”, “Fanconi Anemia”) and were excluded from this summary.