

Results of PGC Survey

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Why a Survey?

- To gather information from a wide cross section of the PGC community and make it available to parents for comparative purposes
- To potentially identify areas for further study

Survey Who?

- Parents of Classic Galactosemic Children
- Adult Classic Galactosemics
- Parents of Variant Galactosemic Children
- Adult Variant Galactosemics
- Both U.S. and International

Method

- Develop Prototype, Then Run Pilot Test
- Finalize Survey and Distribute Surveys
 - Over 600 sent out in US and International
 - PRIVACY !
- Collect surveys
 - Only ~125 returned
- Analyze results
 - Enter into a database, then ask questions

This is ONLY a survey !!
***This is NOT a scientifically
validated study !!***
***These results were NOT
reviewed by Medical
Professionals !!***

Age Demographics of Respondents with Classic Galactosemia

Age Group	Male	Female	Total
0-2 yrs	6	6	12
3-5 yrs	8	13	21
6-12 yrs	22	13	35
13-18 yrs	9	9	18
19-25 yrs	4	4	8
26+ yrs	5	7	12
	54	52	106

Newborn Period

Newborn period

	Average Length (in/cm)	Average Weight – (lbs/Kg)
USA (Full-Term)	20.4 / 51.8	8.0 / 3.6
USA (Premature)	18.8 / 47.6	6.6 / 3.0
International (Full-Term)	20.2 / 51.2	7.8 / 3.5

Newborn period

Age Group (US classics)	Avg. Time to diagnosis (days)	Avg. Time to diet change (days)
0-2 yrs	9.4	7.3
3-5 yrs	10.6	6.7 *
6-12 yrs	10.1	8.1 *
13-18 yrs	12.4	11.2 *
19-25 yrs	20.7	16.7
26+ yrs	69.4	68.3 *

Newborn period

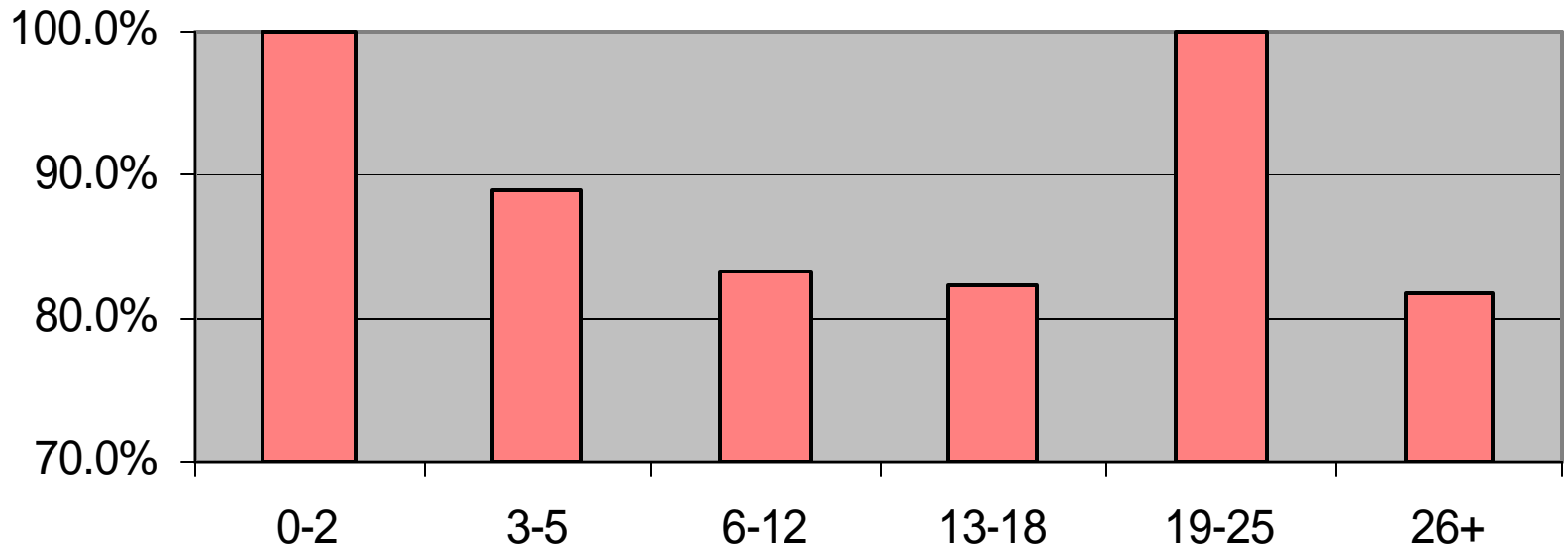
Age Group (International classics)	Avg. Time to diagnosis (days)	Avg. Time to diet change (days)
0-2 yrs	4.7	5.0*
3-5 yrs	14.3	14.3
6-12 yrs	8.8	7.8
13-18 yrs	51	51
19-25 yrs	~	~
26+ yrs	30	27

Newborn period

Age Group (All Classics)	% of Respondents Still on Formula
0-2 yrs	75
3-5 yrs	67
6-12 yrs	51
13-18 yrs	29
19-25 yrs	14
26+ yrs	8

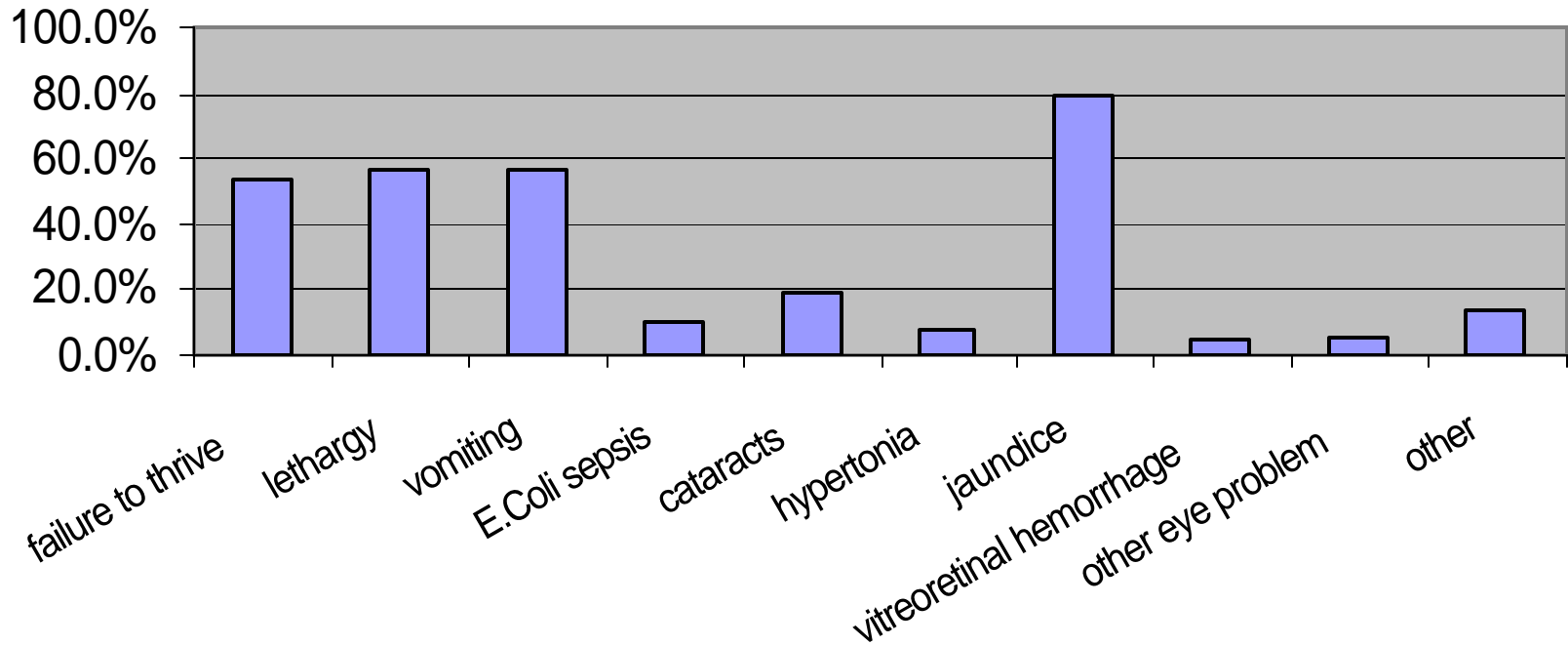
Newborn period

percentage of U.S. classic galactosemics reporting complications in the newborn period, sorted by age at reporting



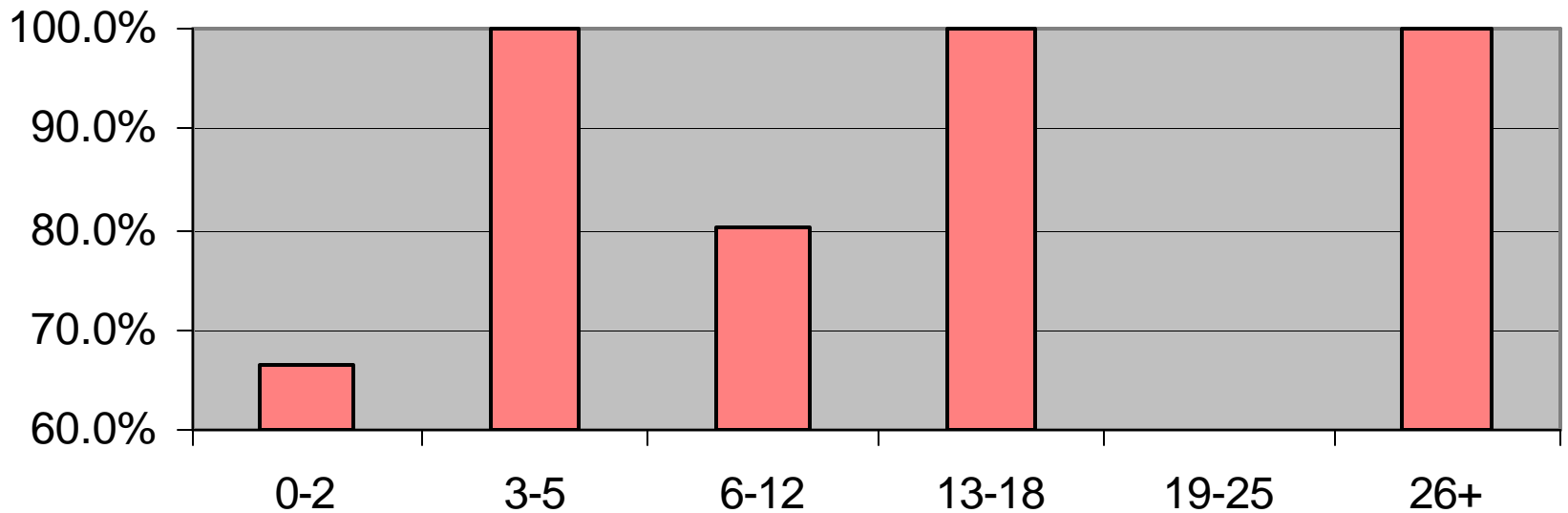
Newborn period

Complications from the newborn period as reported for U.S. Classic Galactosemics



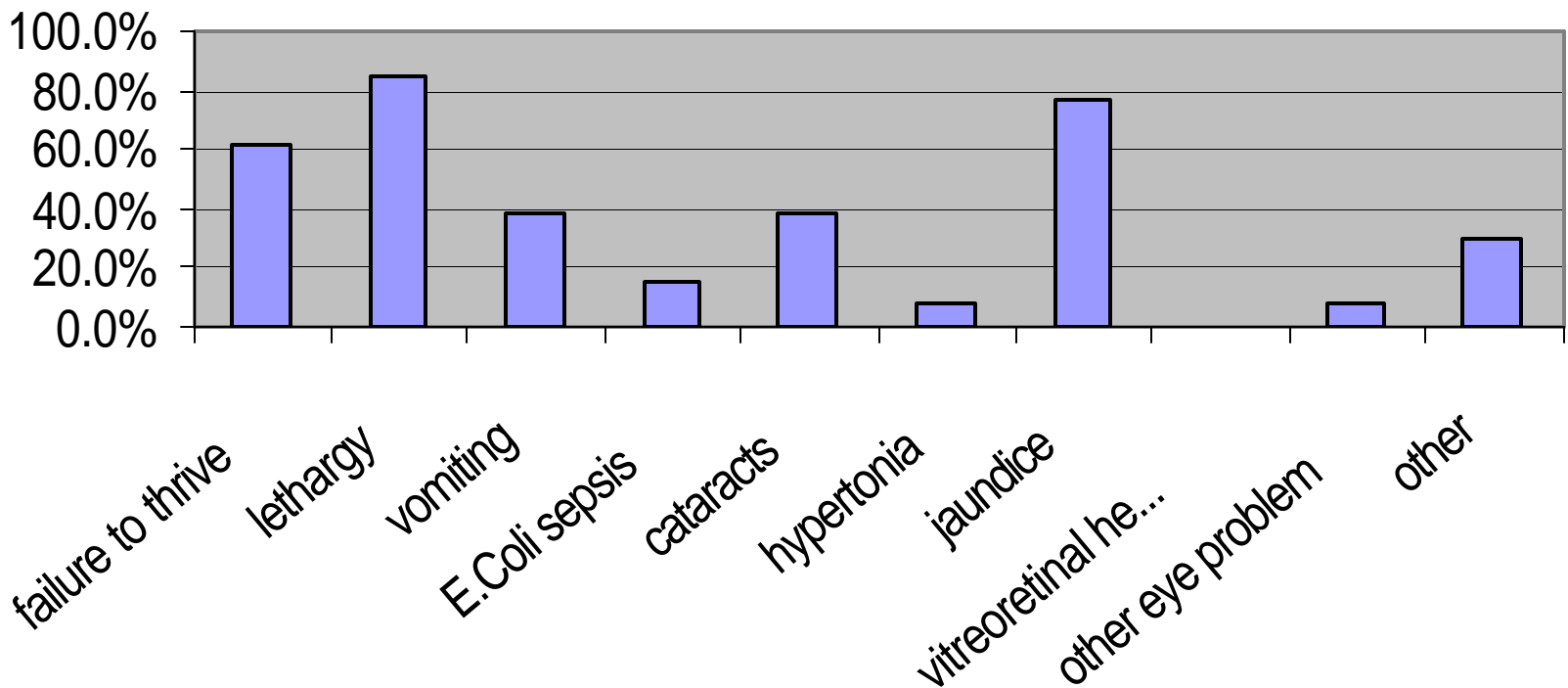
Newborn period

percentage of International classic galactosemics reporting complications in the newborn period, sorted by age at reporting



Newborn period

Complications from the newborn period as reported for International Classic Galactosemics



Clinic Visits

Doctors Visited By Classic Galactosemics

Age 0-2:

Geneticist & Nutritionist = 2 ~3 x per year

Ophthalmologist = 1~2 x per year

Age 3-5:

Geneticist & Nutritionist = ~2 x per year

Ophthalmologist = 1~2 x per year

Age 6-12:

Geneticist & Nutritionist = 1~2 x per year

Ophthalmologist = ~1 x per year

Endocrinologist = 1~2 x per year

Doctors Visited By Classic Galactosemics

Age 13-18:

Geneticist = 1~2 x per year

Nutritionist = 1x per year

Ophthalmologist = 1x per year

Endocrinologist = 1x per year

Age 19-25 (*not many responses*):

Geneticist = 1 x per year

Ophthalmologist = 1x per year

Endocrinologist = 1~2 x per year

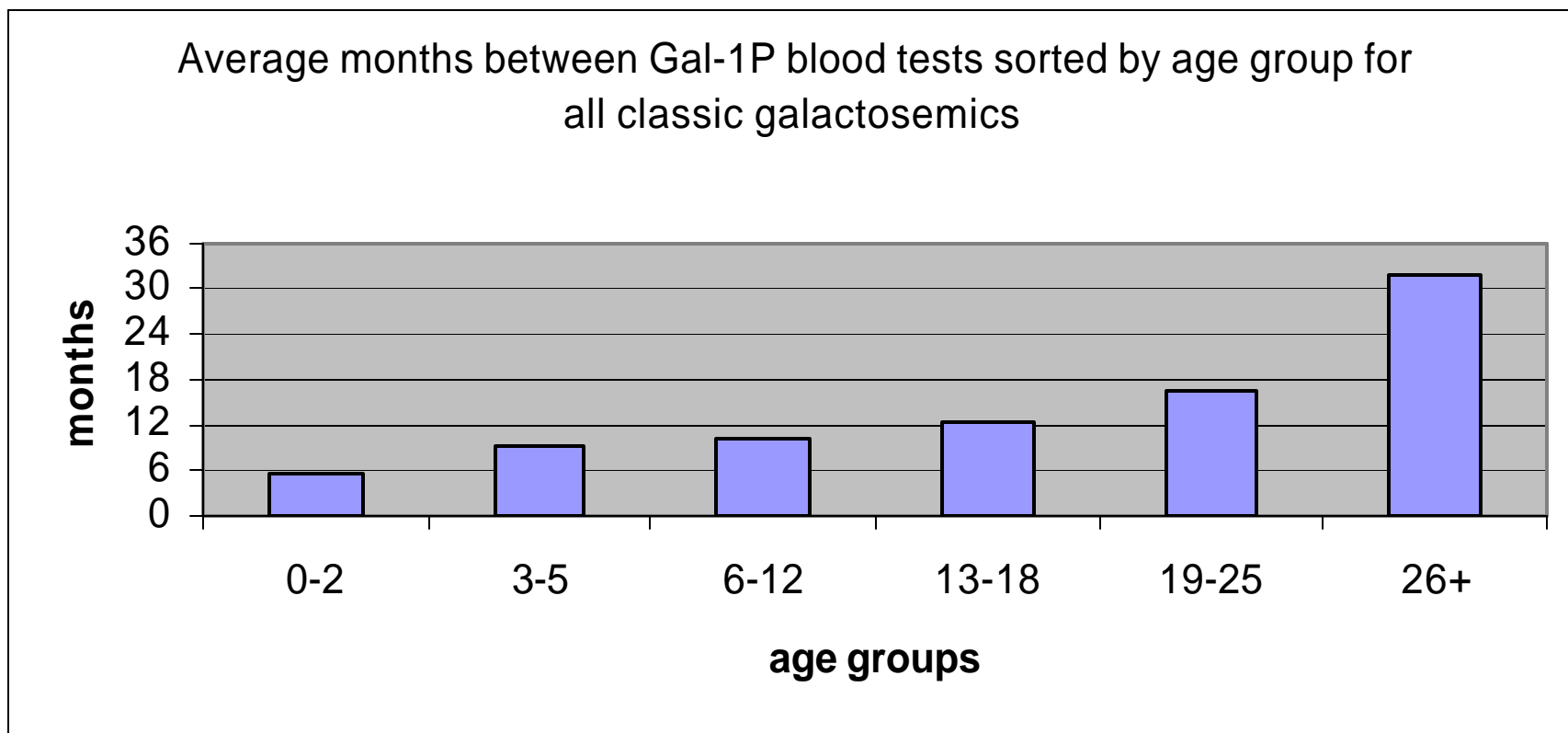
Age 26+ (*not many responses*): :

Geneticist = 2 x per year

Ophthalmologist = 1 x per year

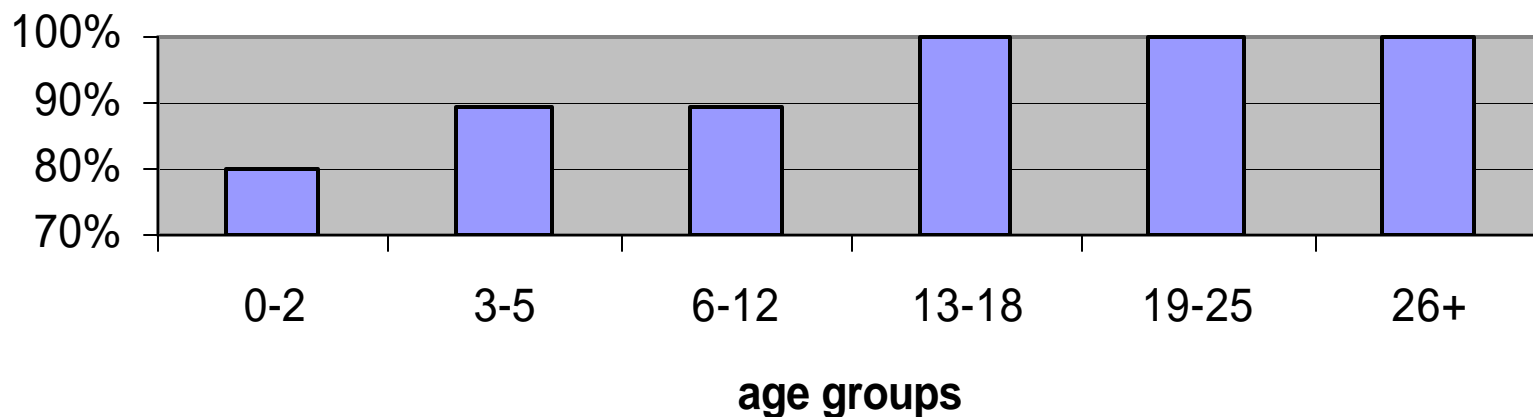
Endocrinologist = 4 x per year

Gal-1-P Blood Testing

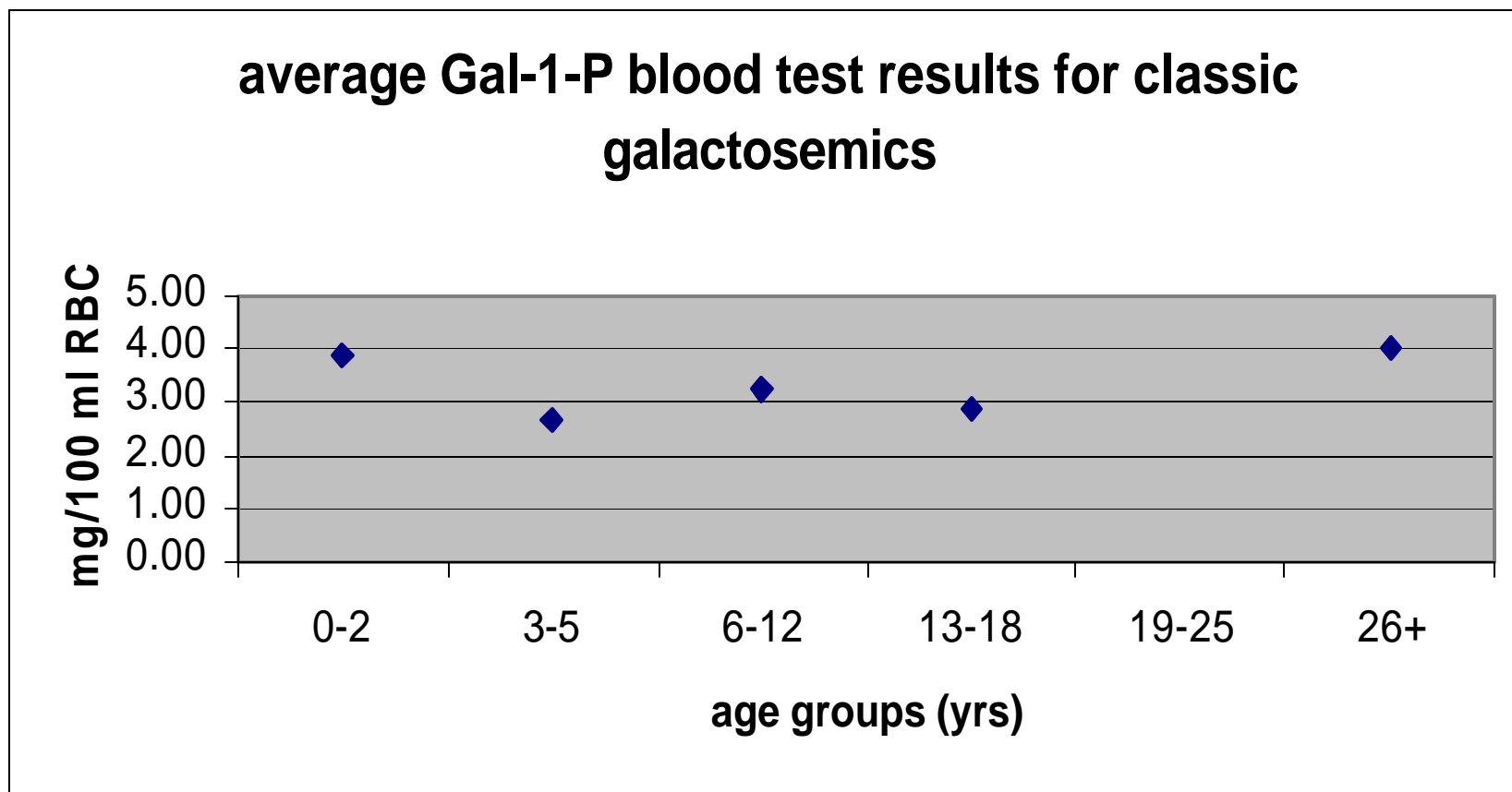


Gal-1-P Blood Testing

percentage of classic galactosemics reporting that most recent Gal-1-P blood test results fall within range desired by physician

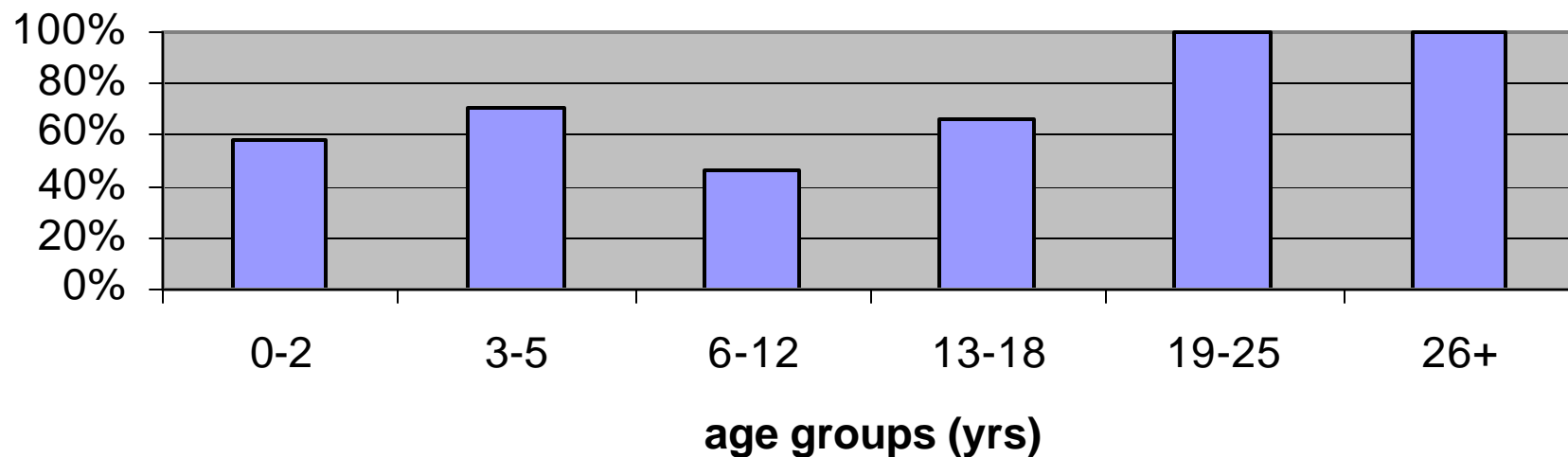


Gal-1-P Blood Testing

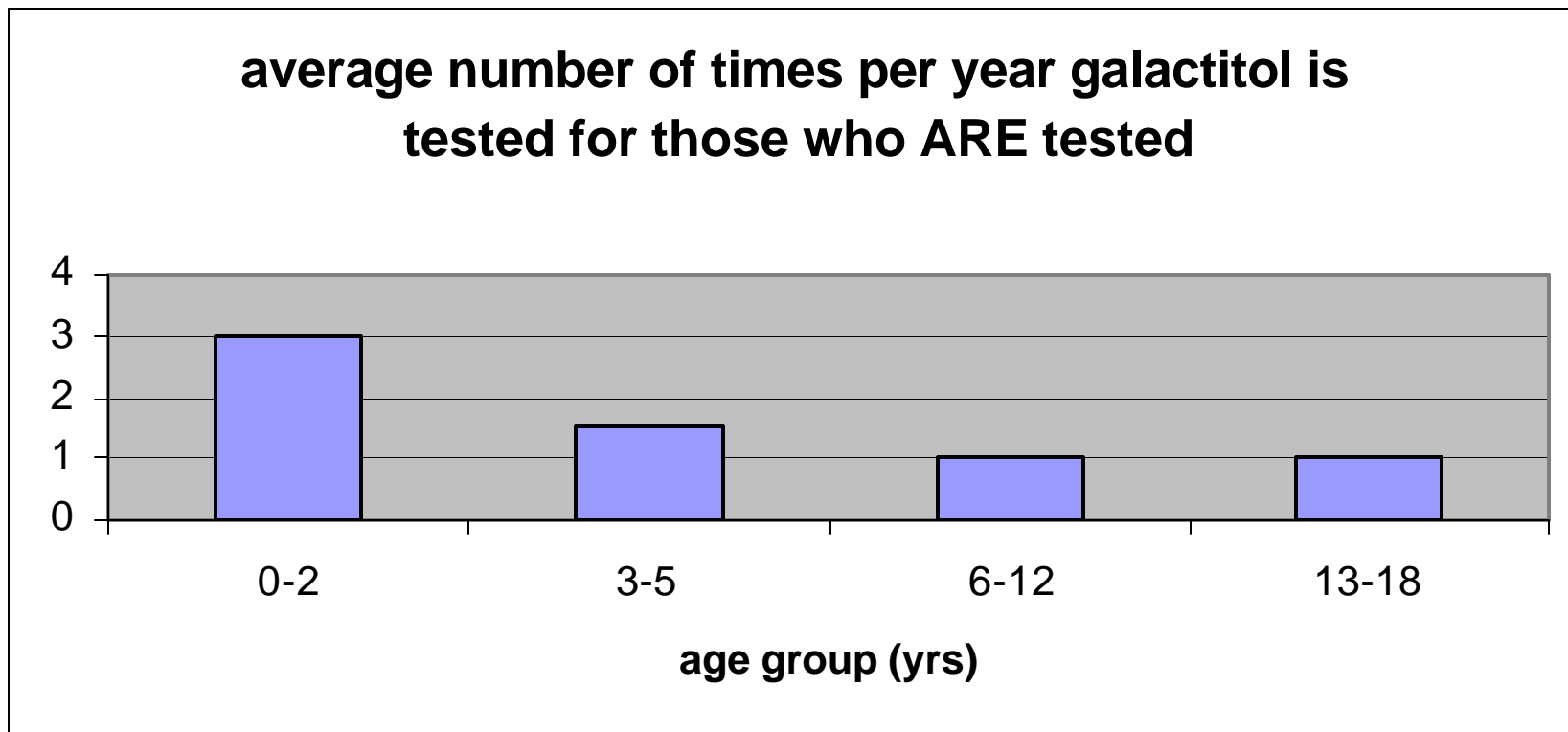


Galactitol Urine Testing

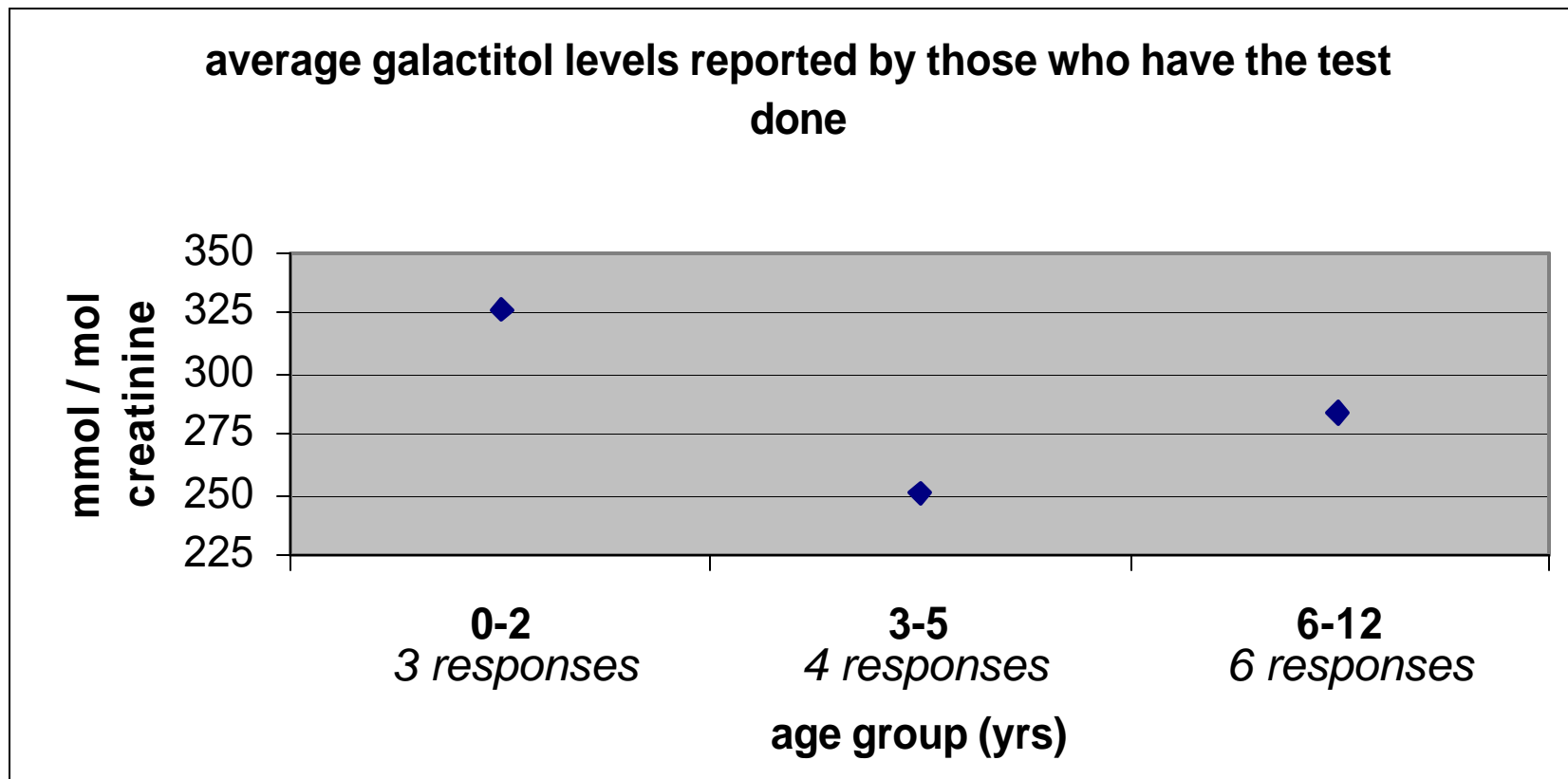
percentage of respondents who never have galactitol tested



Galactitol Urine Testing



Galactitol Urine Testing

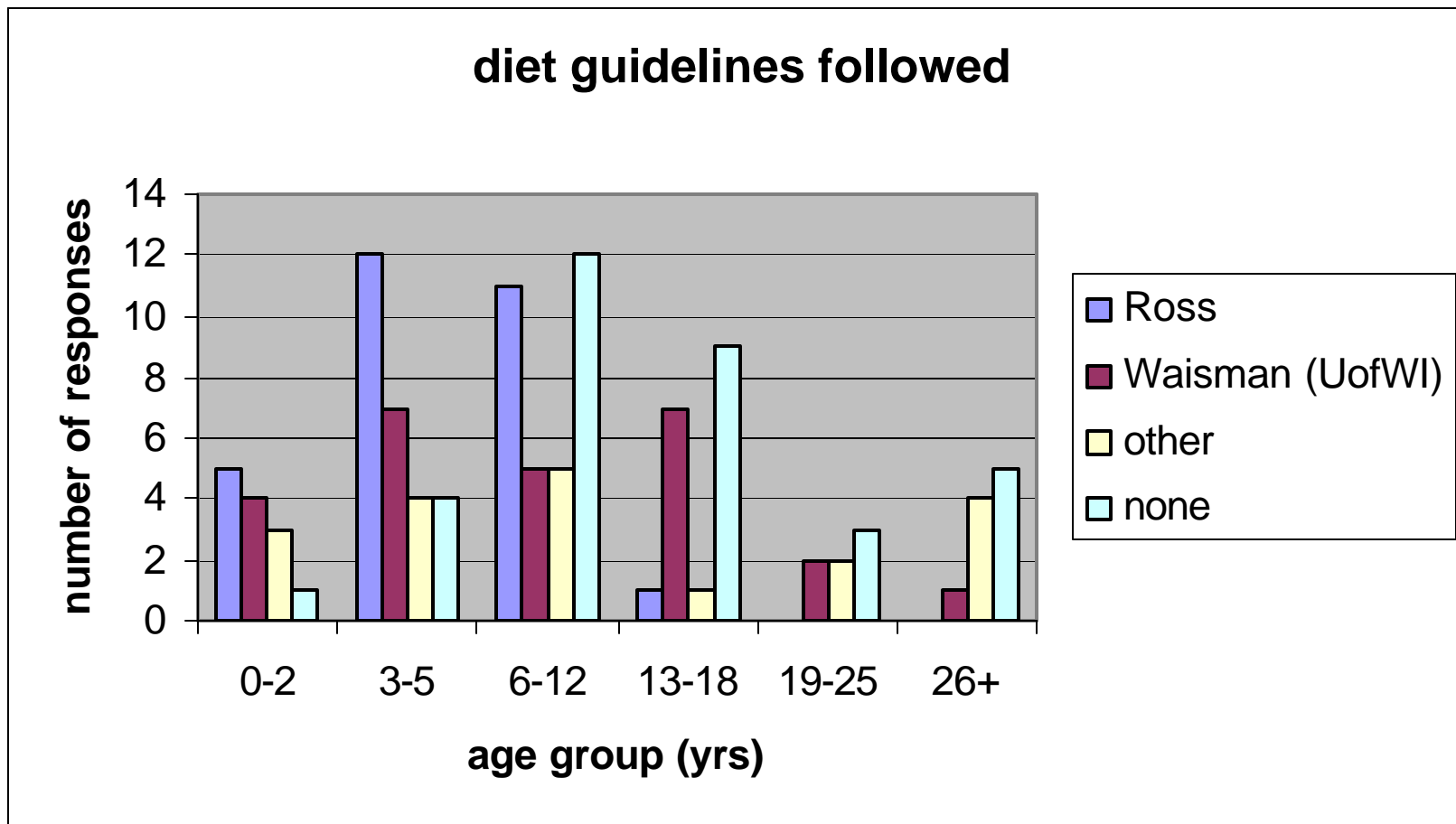


Genotyping

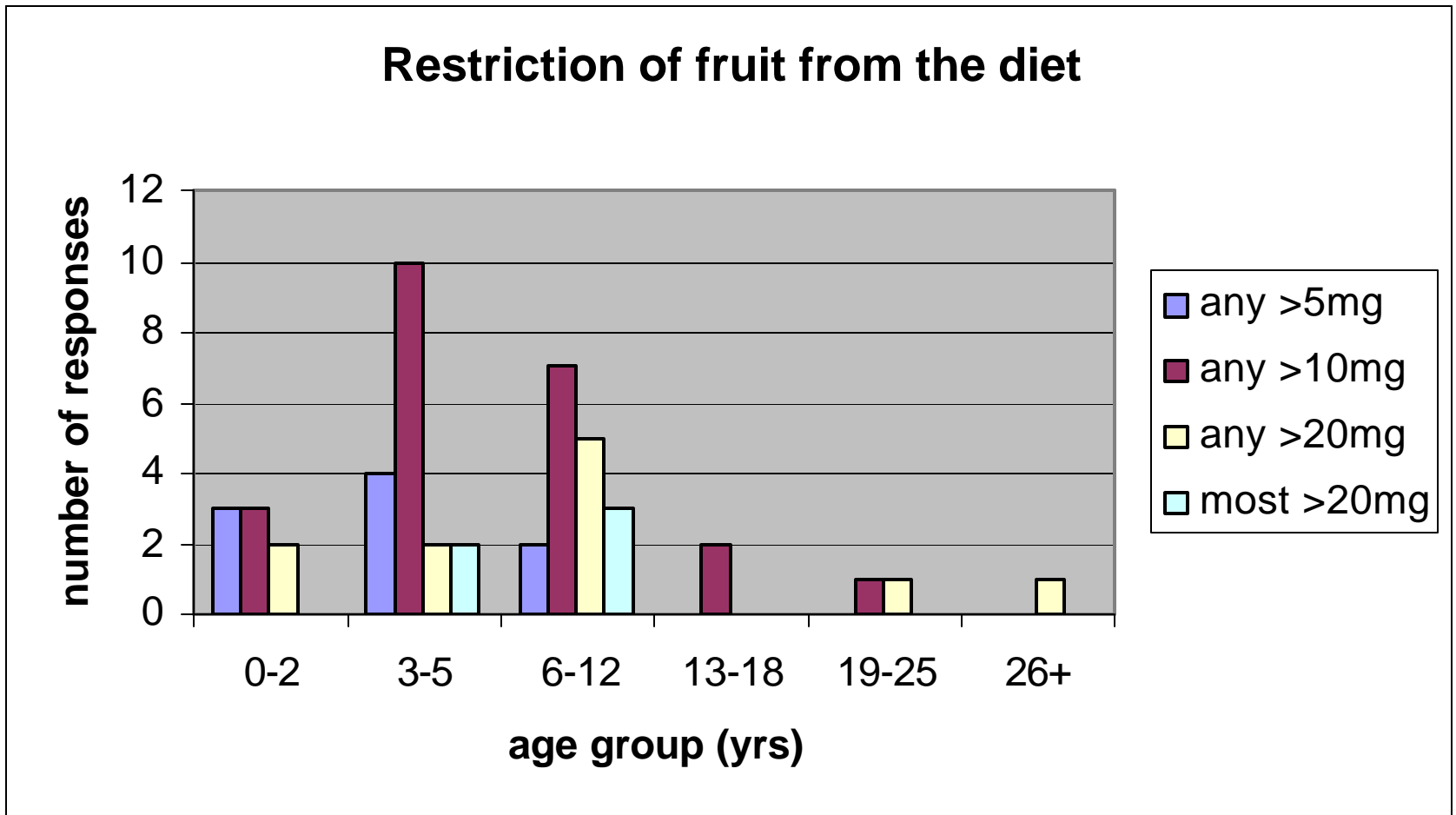
Genotype	Number Reported
Q188R / Q188R	27
Q188R / Unknown	11
Q188R / K285N	3
Q188R / L195P	2
Q188R / other	(1) Y209S, (1) S13L, (1)R333G, (1) L289R, (1) E308K
other / other	(1) L195P / A320P (1) K285N / unknown

Diet Items

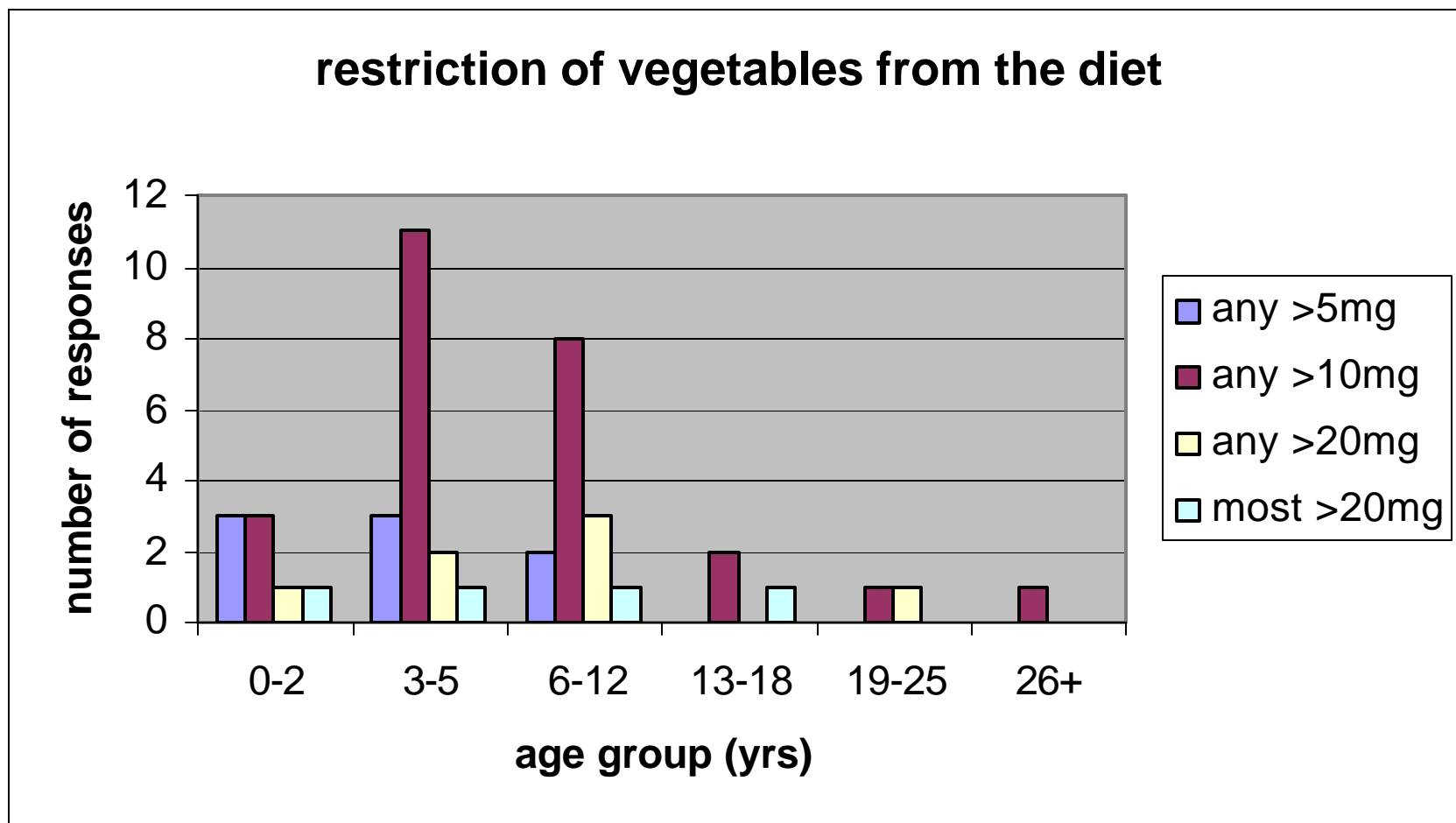
Diet Issues



Diet Issues

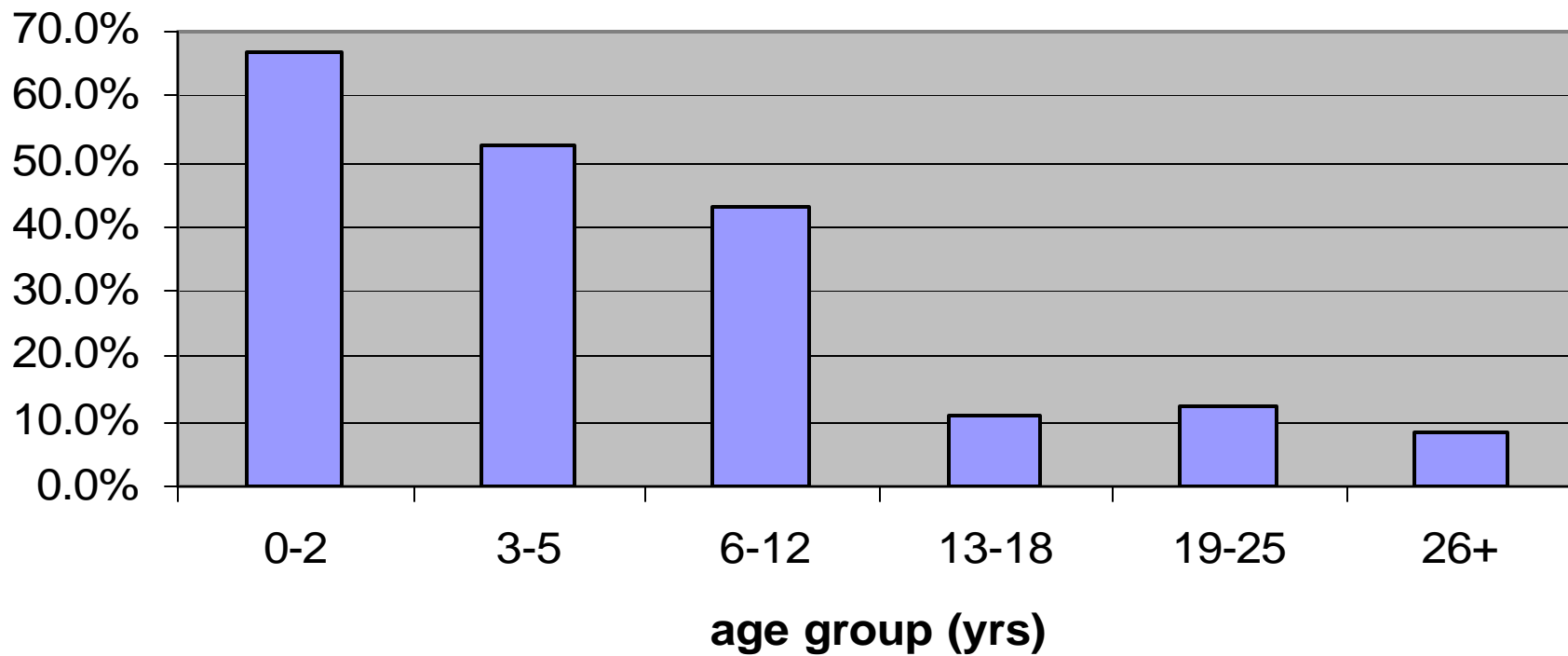


Diet Issues



Diet Issues

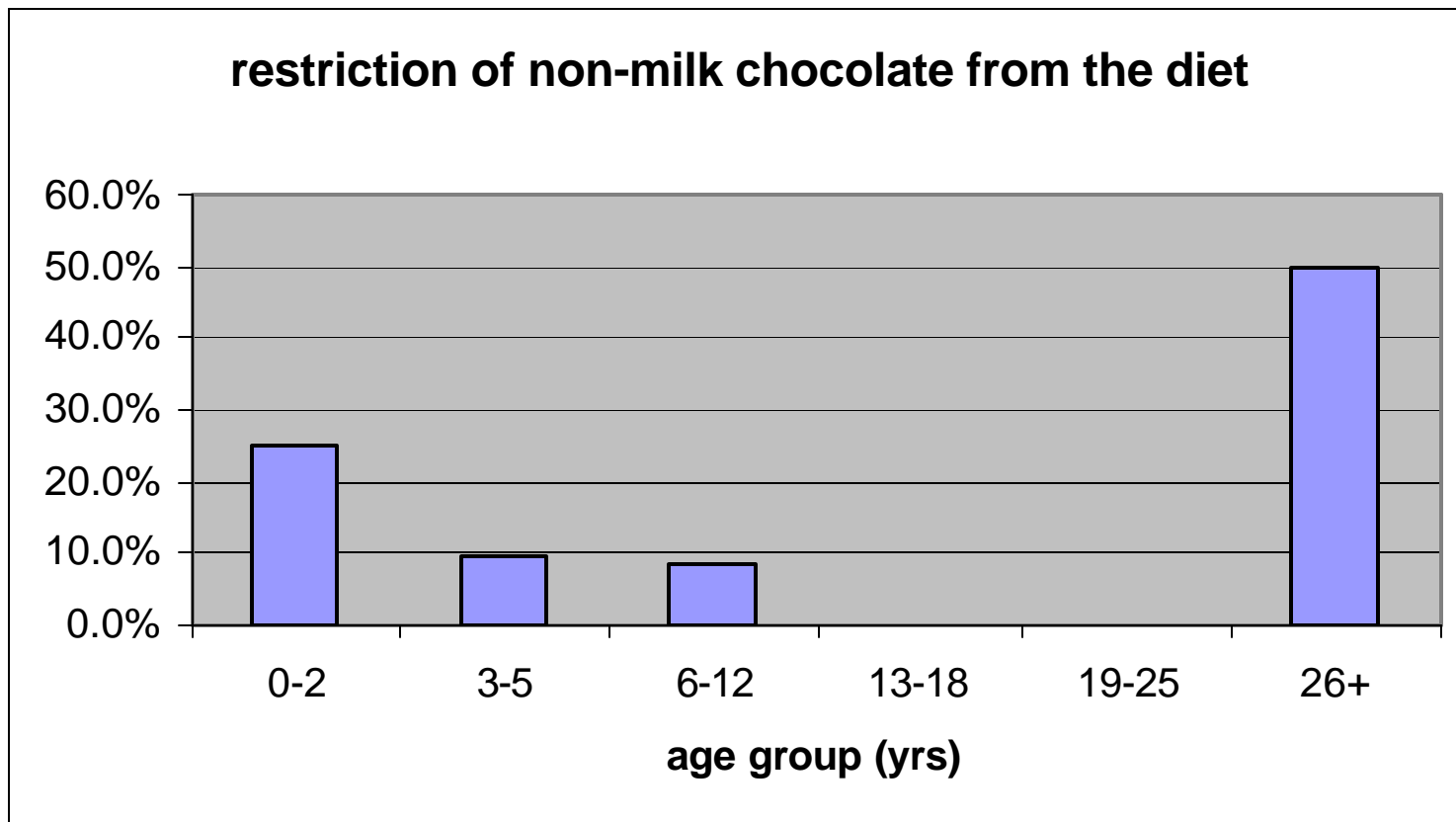
restriction of juice in the diet



Diet Issues

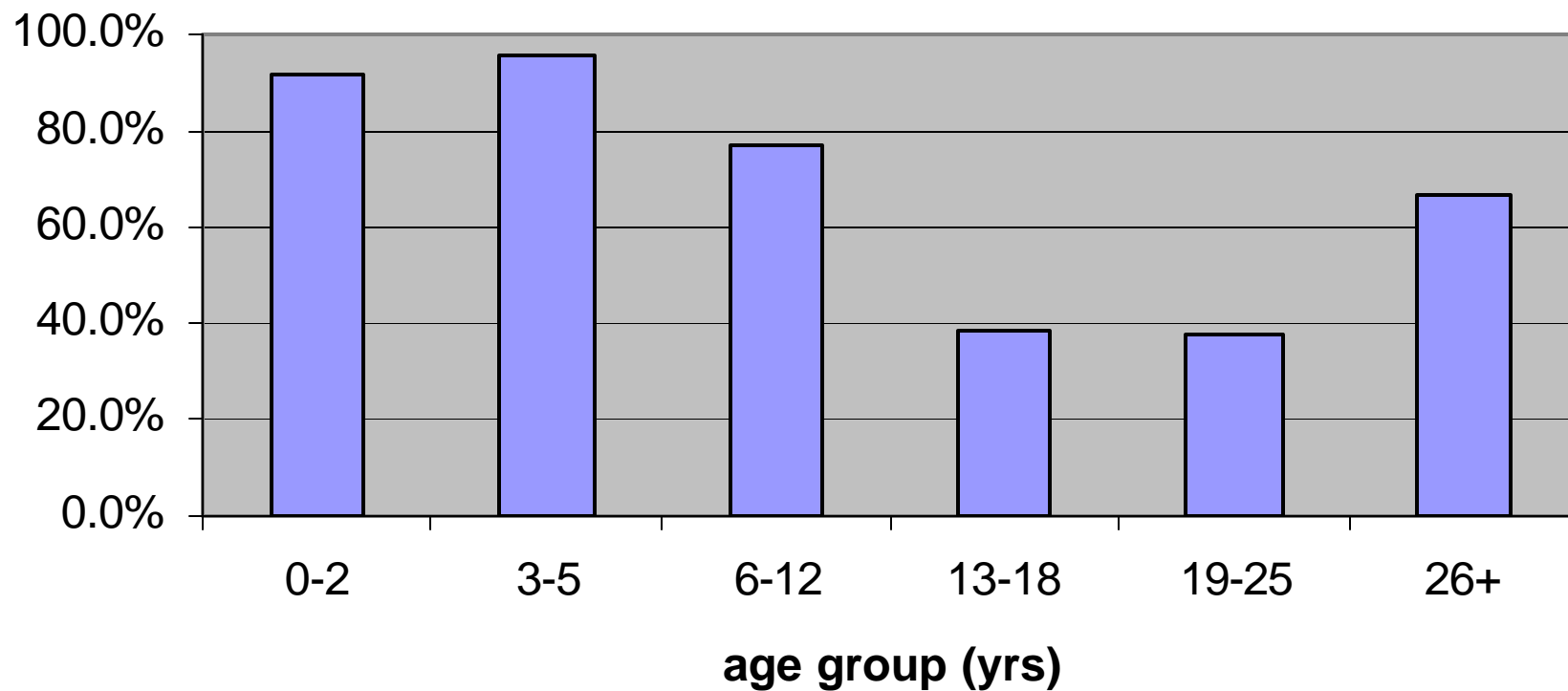


Diet Issues

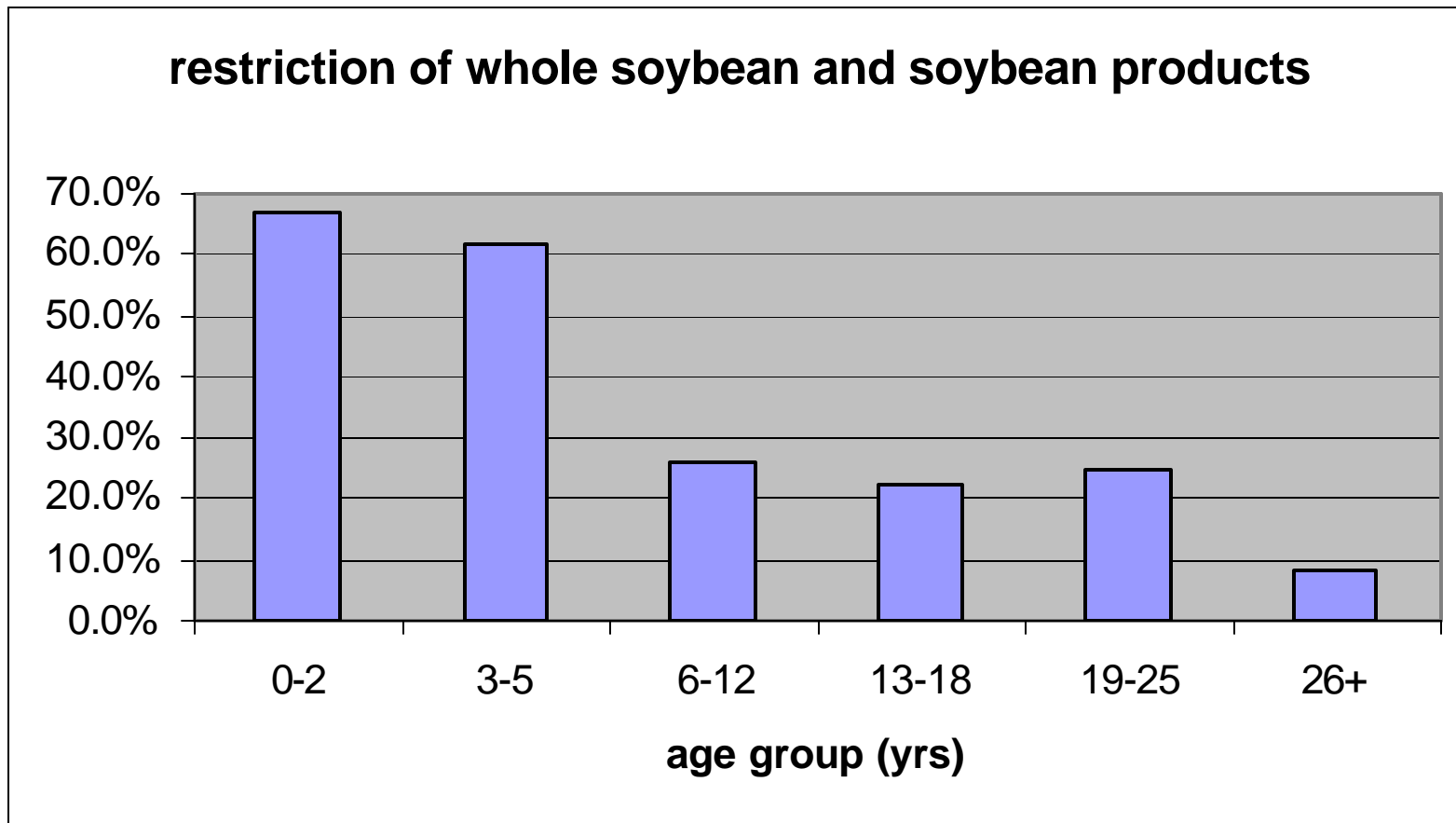


Diet Issues

restriction of legumes from the diet



Diet Issues



Effects of Eating a Restricted Food

- 27 reported an adverse reaction
 - Vomiting: 12
 - Stomach ache: 13
 - Diarrhea: 13
 - Abdominal Gas: 3
 - Mental Effects: 1
 - Appetite Loss: 1

Medical Items

Other Diagnosis - Children

Age group	Learning Disability	Behavior Disorder	ADD / ADHD	Gross Motor	Fine Motor	Sensory Integration Disorder	Auditory Processing Disorder	Speech
0-2 (12)								
3-5 (21)				14%	14%	10%	5%	48%
6-12 (35)	26%	9%	17%	20%	37%	14%	11%	37%
13-18 (18)	33%		22%	11%	7%	22%	17%	56%

Speech Diagnoses - Children

Age Group	Articulation	Dyspraxia	Expressive Language Disorder	Receptive Language Disorder	Aphasia
3-5 (21)	3 / 14 %	2 / 10 %	4 / 19 %	1 / 5 %	
6-12 (35)	7 / 20%	6 / 17 %	6 / 17 %	3 / 9 %	
13-18 (18)	5 / 28%	8 / 44 %	6 / 33 %	1 / 6 %	1 / 6 %

Age Group	Developmental Delay	Fluency	Oral Motor Dysfunction	Stuttering	Recall Disorder
3-5 (21)	3 / 14 %		1 / 5 %	1 / 5 %	1 / 5 %
6-12 (35)	5 / 14 %	3 / 9 %	3 / 9 %	1 / 3 %	4 / 11 %
13-18 (18)	3 / 17 %	1 / 6 %		1 / 6 %	4 / 22 %

Other Diagnoses - Adult Galactosemics

	Males (9)	Females (11)
Special Ed. services	5	6
Learning Disorders	4	5
Behavior Disorders	0	0
ADD/ADHD	0	0
Gross motor	2	5
Fine motor	4	7
Sensory Integration Disorder	2	5
Speech Problems	3	5
Memory Trouble	1	7
Negative Social Effects	4	9

Behavioral Issues

light sensitivity	18%
sound sensitivity	29%
fear of heights	36%
acute shyness	24%
hyperactive	24%
temper tantrums	35%
argumentative	26%
blames others	24%
defiant	28%
high pain threshold	16%
low pain threshold	15%
doesn't transition well	49%
need to spin / swing	9%
hates spin/swing	14%
fearful	34%
often seems to not hear	16%
can only follow 1 direction	31%
little or no imagination	26%
excessive sadness	25%
OCD	16%
immaturity	43%
doesn't get along well	13%
other	7%
fixated	31%
dislikes textures	32%

Male

light sensitivity	22%
sound sensitivity	29%
fear of heights	42%
acute shyness	30%
hyperactive	14%
temper tantrums	32%
argumentative	25%
blames others	10%
defiant	13%
high pain threshold	13%
low pain threshold	26%
doesn't transition well	27%
need to spin / swing	6%
hates spin/swing	8%
fearful	31%
often seems to not hear	28%
can only follow 1 direction	44%
little or no imagination	12%
excessive sadness	14%
OCD	10%
immaturity	46%
doesn't get along well	8%
other	0%
fixated	21%
dislikes textures	37%

Female

Speech / Language / Vision Issues

receptive	31%
expressive	47%
aphasia	4%
unspecified	0%
other	5%
fluency	7%
stuttering	6%
delayed	41%
articulation	33%
oral motor	26%
recall	31%
dispraxia	36%
other	6%
cataracts	36%
vitreoretinal hemorrhage	7%
spatial perception	13%
other	24%

Male

receptive	28%
expressive	45%
aphasia	6%
unspecified	0%
other	0%
fluency	15%
stuttering	3%
delayed	36%
articulation	38%
oral motor	27%
recall	28%
dispraxia	30%
other	7%
cataracts	24%
vitreoretinal hemorrhage	0%
spatial perception	9%
other	16%

Female

Other Medical / Health Issues

gross	39%
fine	60%
low muscle tone	40%
tremors	23%
neurological	15%
seizure	3%
thyroid	0%
ovarian	0%
osteoporosis	0%
joint pain	15%
constipation	25%
ataxia	3%
delayed growth	27%
encopresis	3%
bed wetting	24%
depression	18%
sensory integration disorder	25%
muscle spasms	3%
bad breath	18%
teeth problems	20%
other	15%

Male

gross	39%
fine	49%
low muscle tone	32%
tremors	32%
neurological	22%
seizure	5%
thyroid	6%
ovarian	56%
osteoporosis	18%
joint pain	20%
constipation	27%
ataxia	2%
delayed growth	37%
encopresis	3%
bed wetting	19%
depression	13%
sensory integration disorder	27%
muscle spasms	6%
bad breath	22%
teeth problems	25%

Female

Comparing Genotype

- 27 reported Homozygous Q188R
- 24 reported non-Homozygous Q188R
- Almost identical results for 'other diagnoses'
- Except ADD/ADHD
 - 6 / 27 (22%) Homozygous Q188R
 - 2 / 24 (8%) non Homozygous Q188R

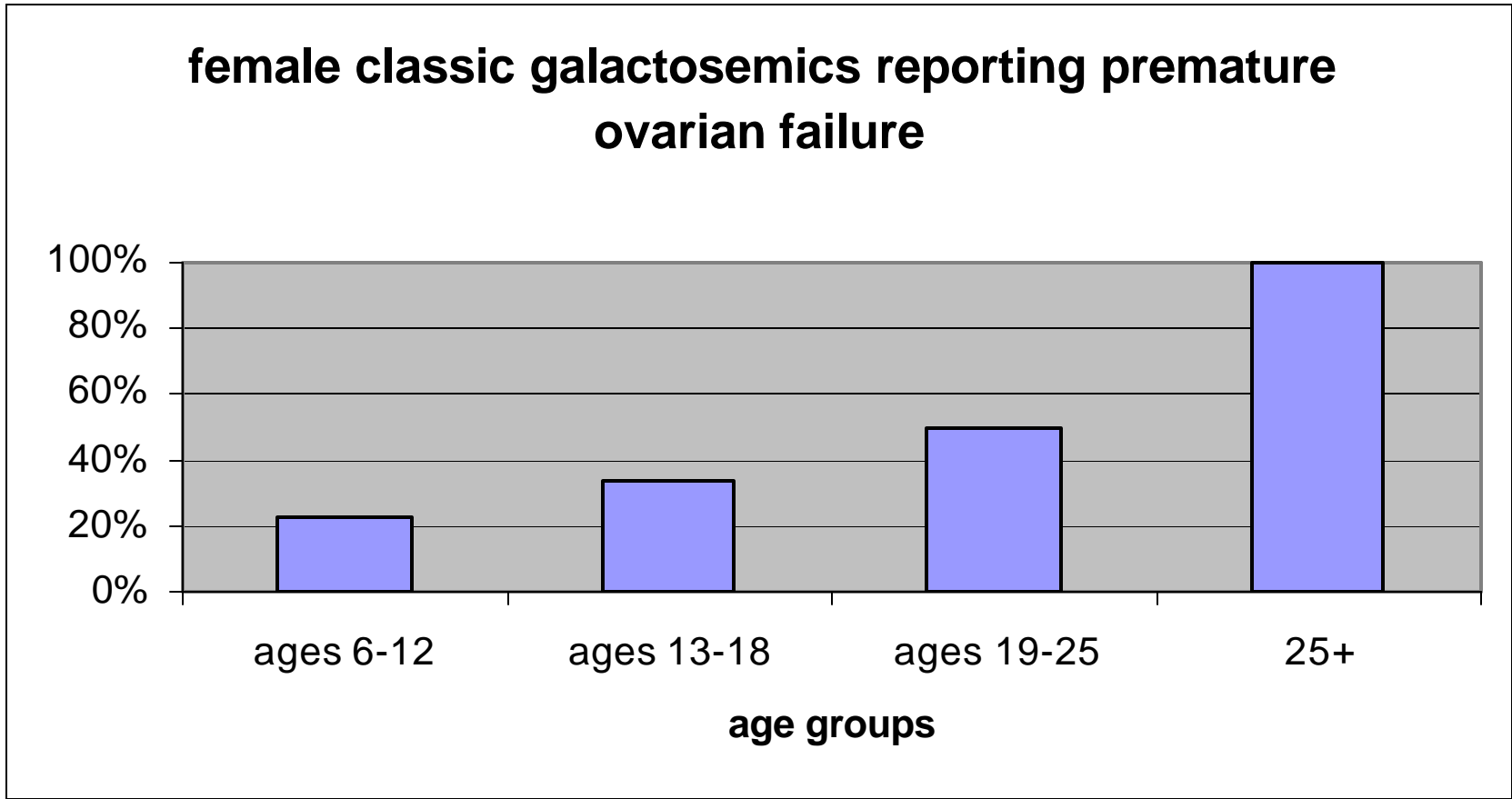
Comparing Time to Diet Change

- Compared
 - 0 days (11 reported)
 - first week (43 reported)
 - after first week (49 reported)
- ‘First week’ and ‘after first week’ very similar
 - Fine motor was worse in ‘after first week’ (13/49) than ‘first week’ (9/43)
 - Sensory integration was worse in ‘after first week’ (8/49) compared to ‘first week’ (3/43)

Comparing Time to Diet Change

- ‘zero days’ group has *substantially* better results – even for speech!
- Only one or two responses for each diagnoses at most

Reproductive Health



Reproductive Health

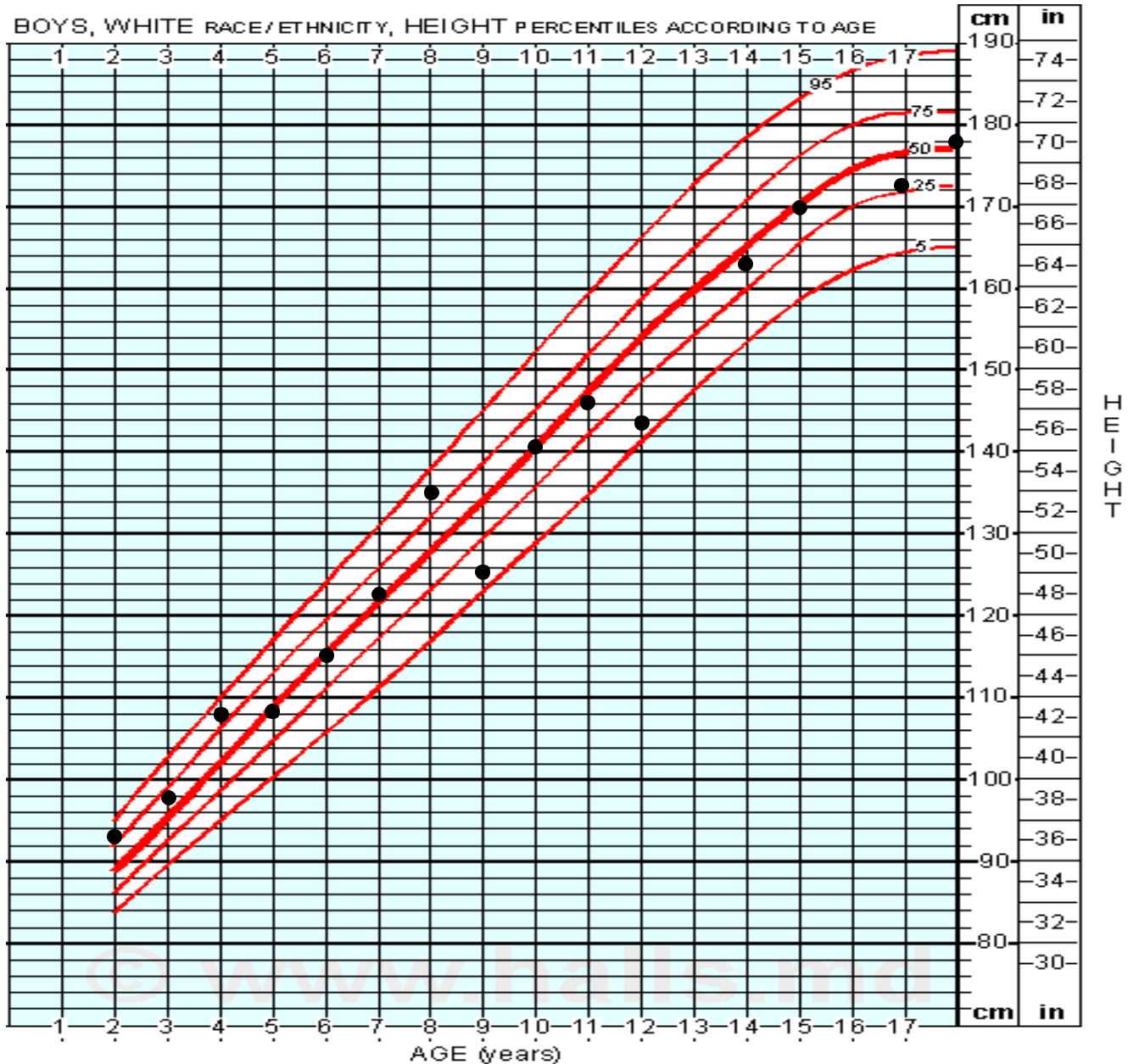
- Almost all females reporting POF also report being on HRT
 - Estroqel Prometrium
 - Demulen
 - Prempro
 - Premarin
- No female respondents report pregnancy
- One non-respondent is known to have had two successful pregnancies

Reproductive Health

- One male respondent reports fathering a child

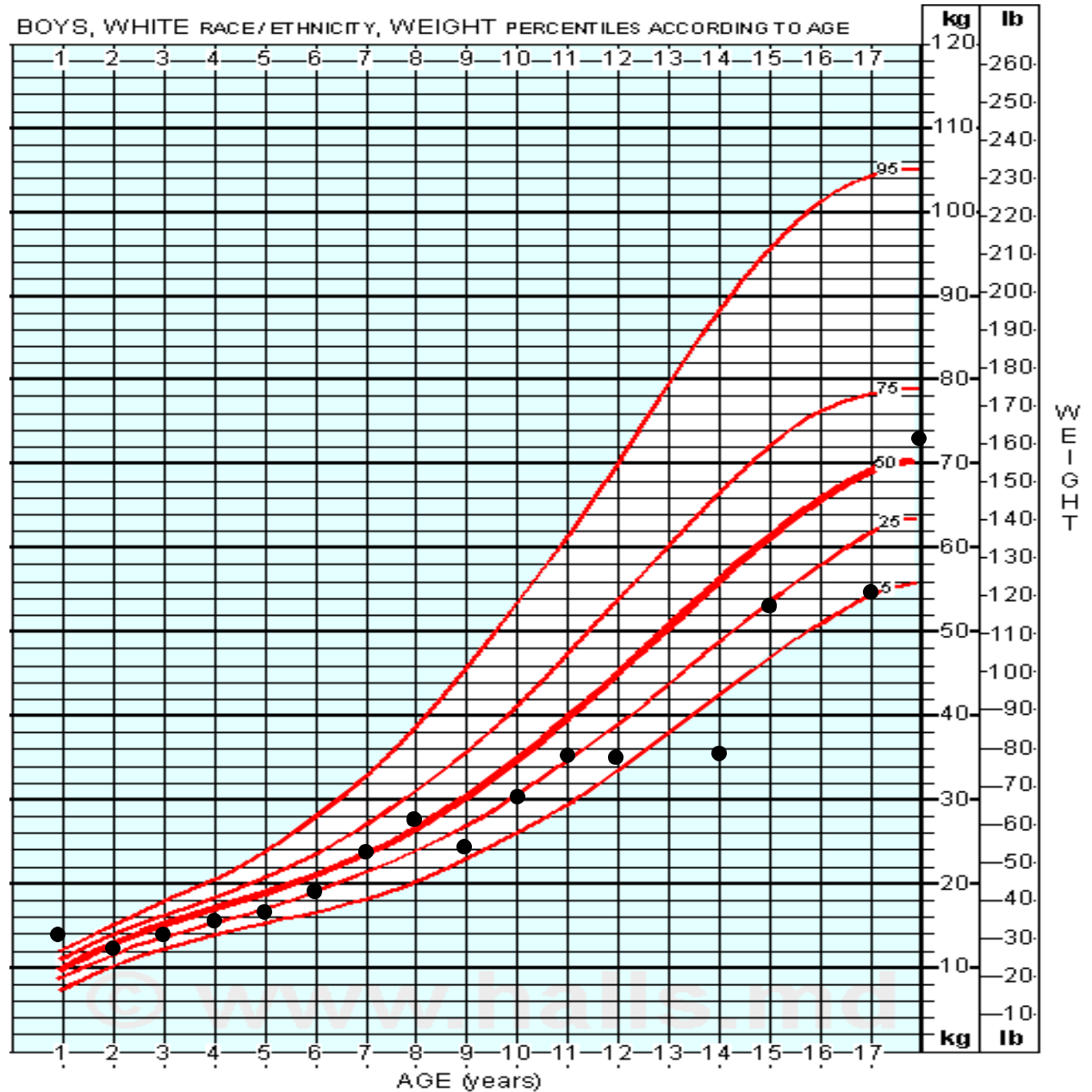
Height and Weight

Classic Galactosemic Boys Average Height



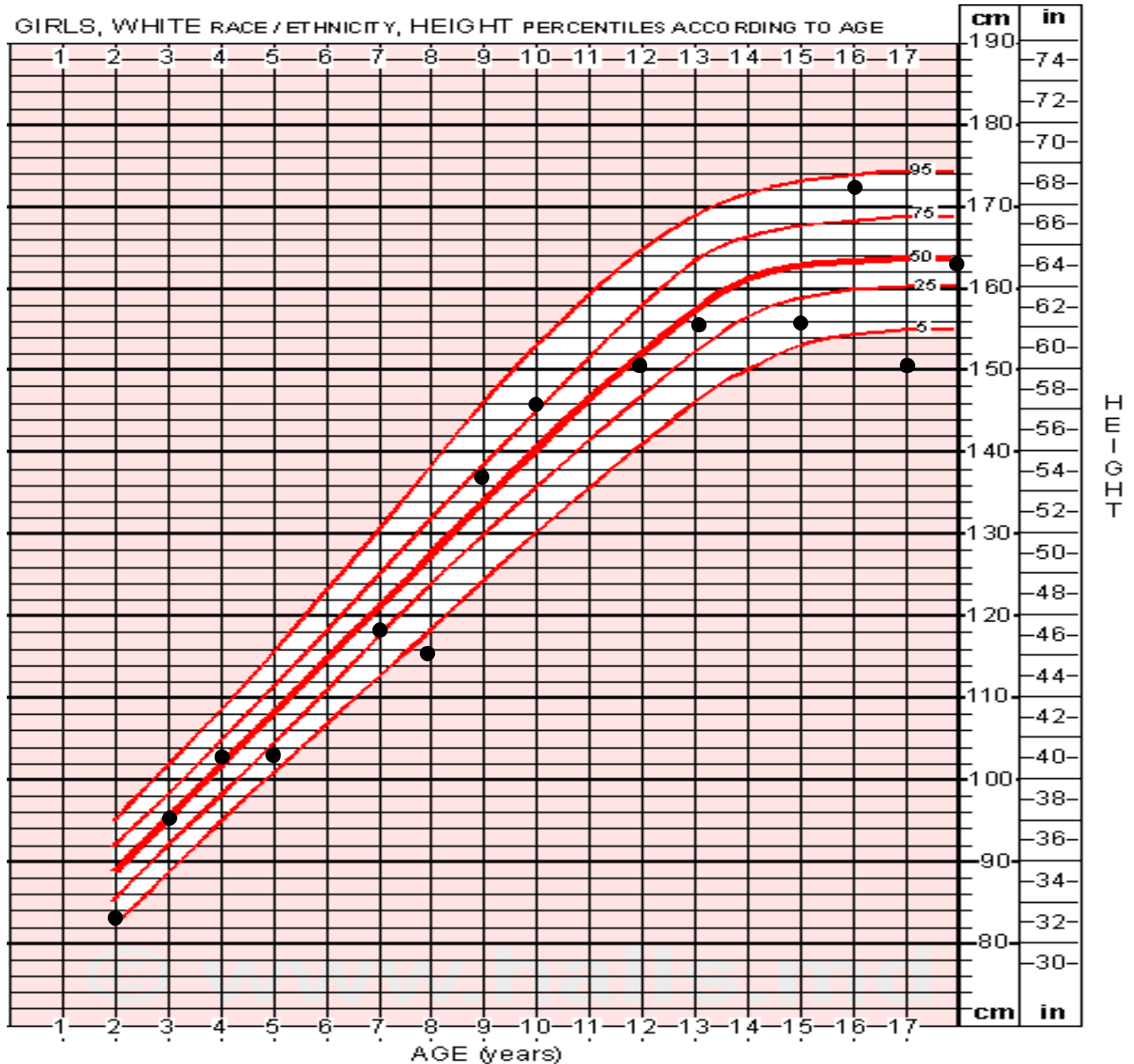
● = average height for galactosemic boys

Classic Galactosemic Boys Average Weight



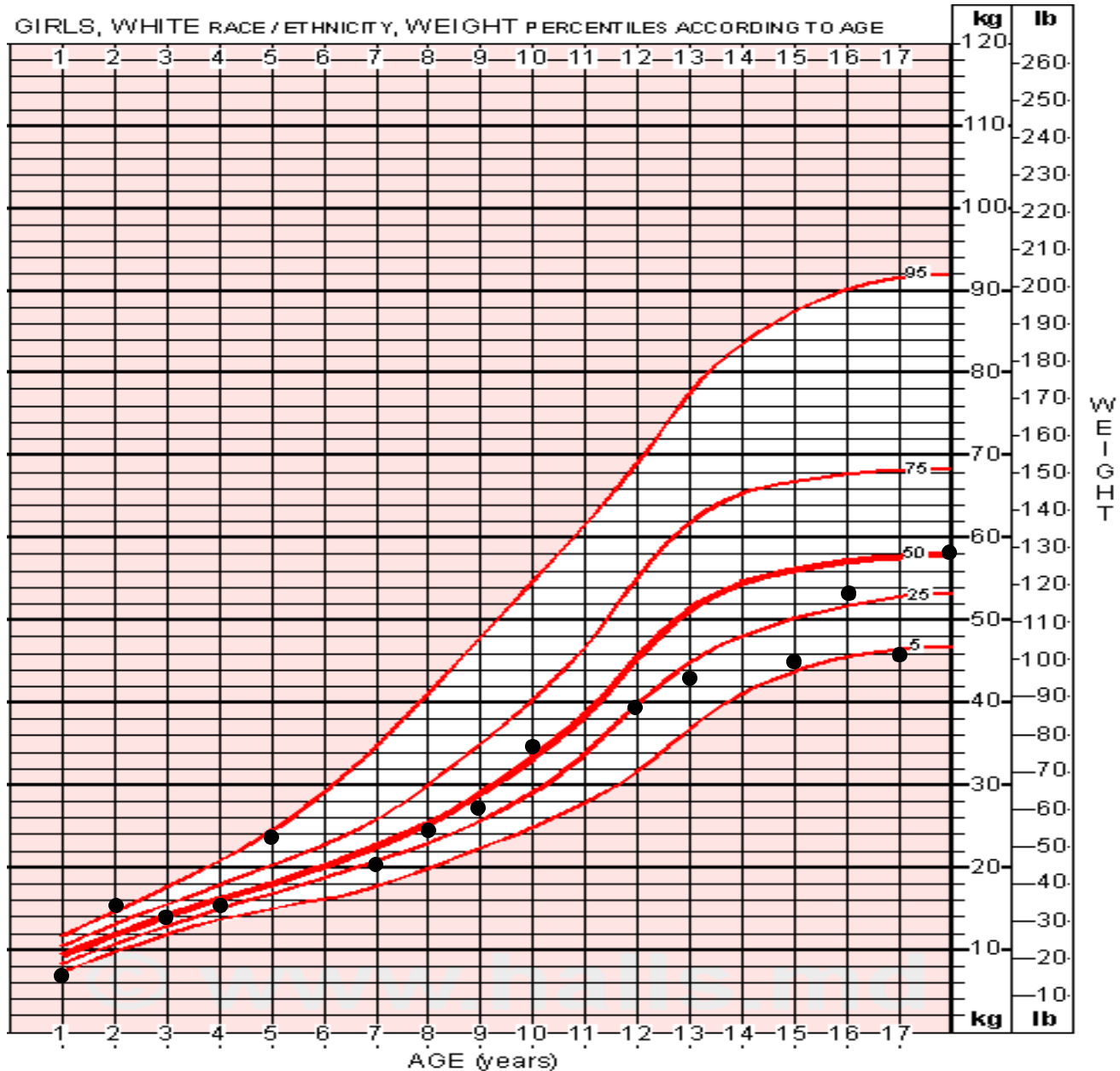
● = average weight for galactosemic boys

Classic Galactosemic Girls Average Height



● = average height for galactosemic girls

Classic Galactosemic Girls Average Weight



● = average weight for galactosemic girls

Classic Galactosemic Children - Height & Weight

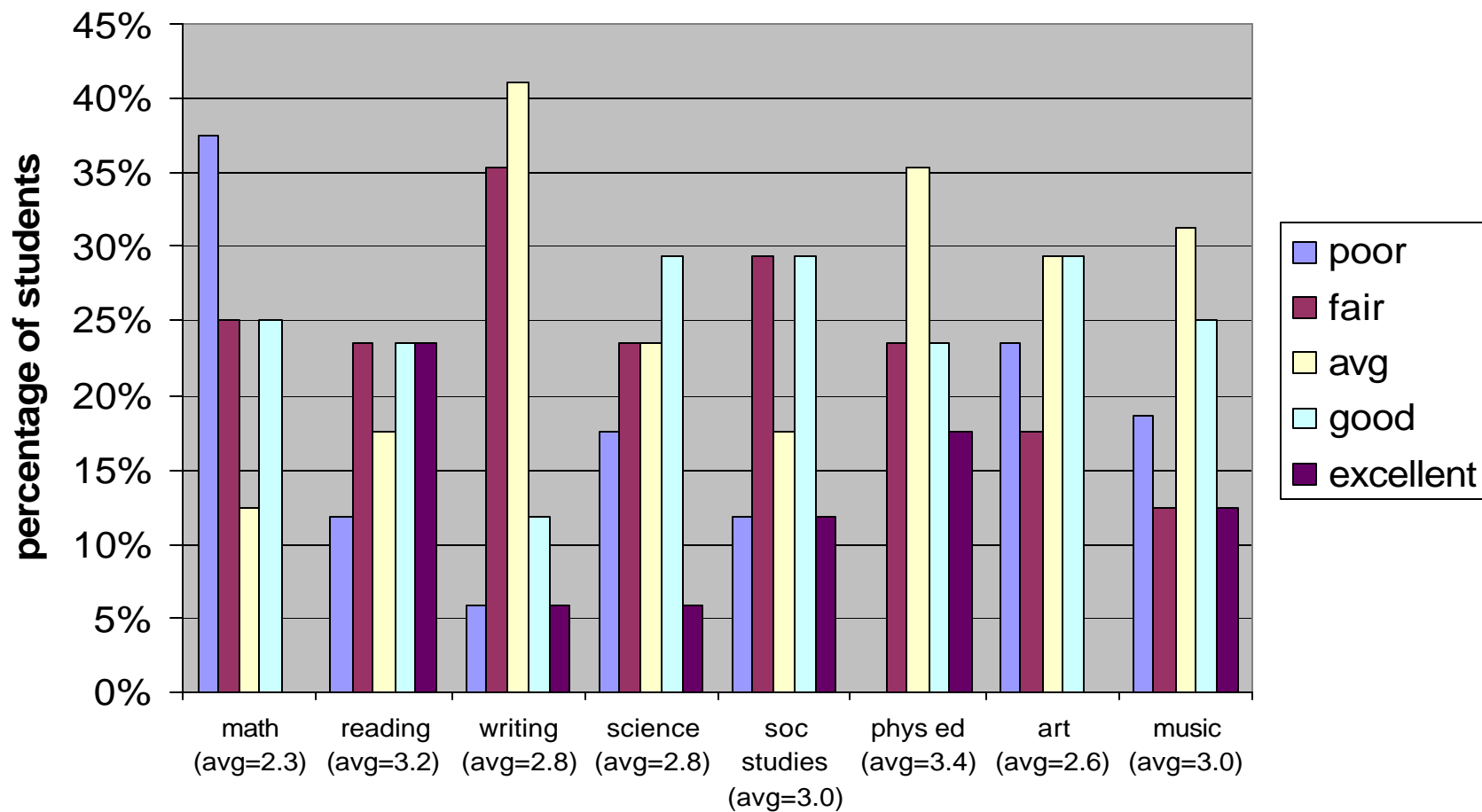
- The data source for this chart is the NHANES III survey, conducted in America during 1988 to 1994. The CDC used this dataset, as well as datasets from several older surveys, to construct the CDC standard pediatric growth charts.

Classic Galactosemic Adults – Average Height & Weight

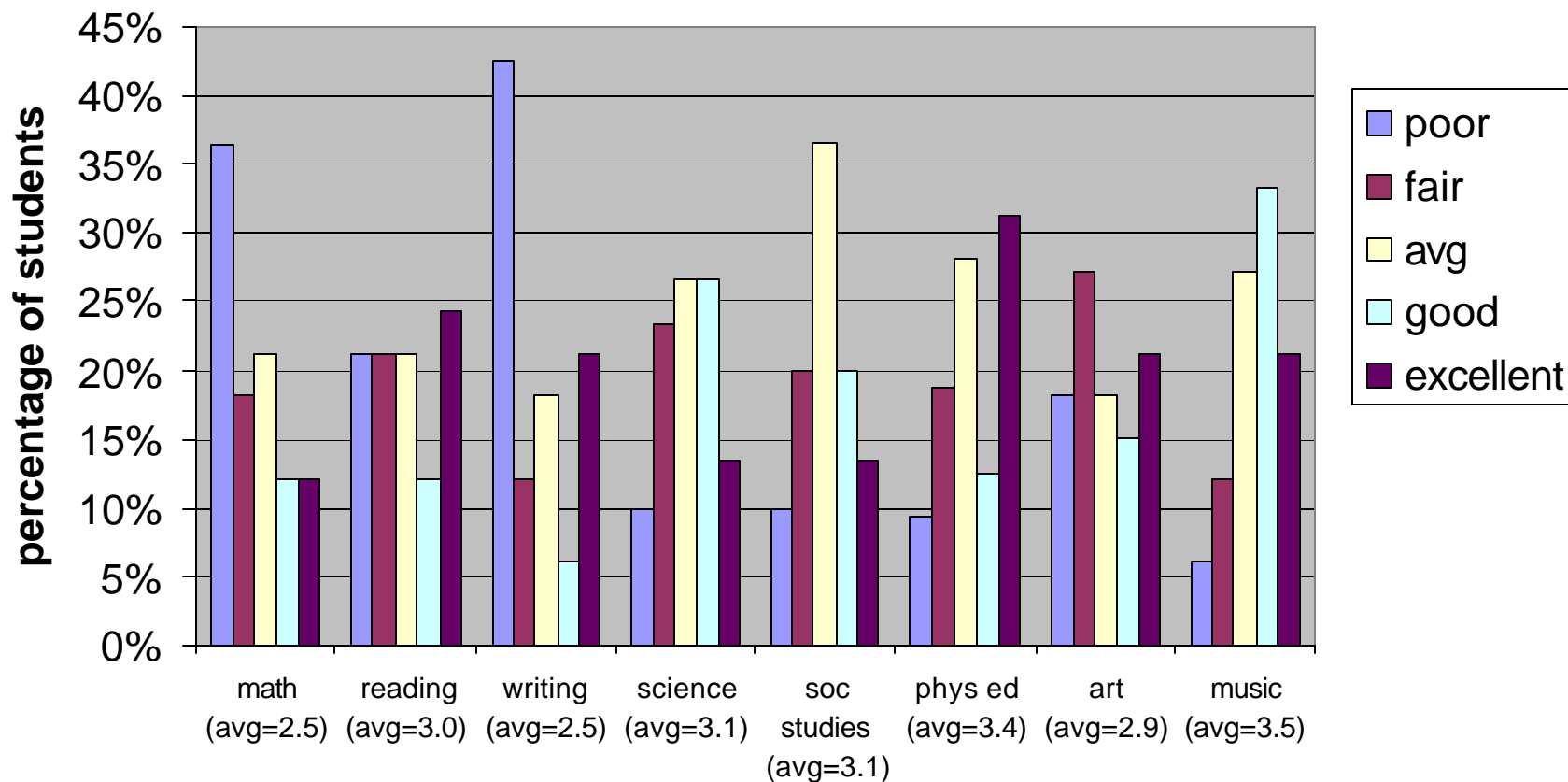
	Average Height	Average Weight
Males <i>Born before 1984</i>	5' 10"	160 lbs
Females <i>Born before 1984</i>	5' 4"	129 lbs

School Items

report card for students age 13-18



report card for students age 6-12



Services Received - Children

age group	occupational therapy	speech therapy	resource room or spec ed	tutoring	1-1 aid	assistive technology
3-5	4 / 19%	10 / 48%	1 / 5%	0 / 0%	0 / 0%	1 / 5%
6-12	12 / 34%	16 / 46%	13 / 37%	9 / 26%	6 / 17%	4 / 11%
13-18	6 / 33%	12 / 67%	7 / 39%	5 / 28%	1 / 6%	2 / 11%

Social Issues - Children

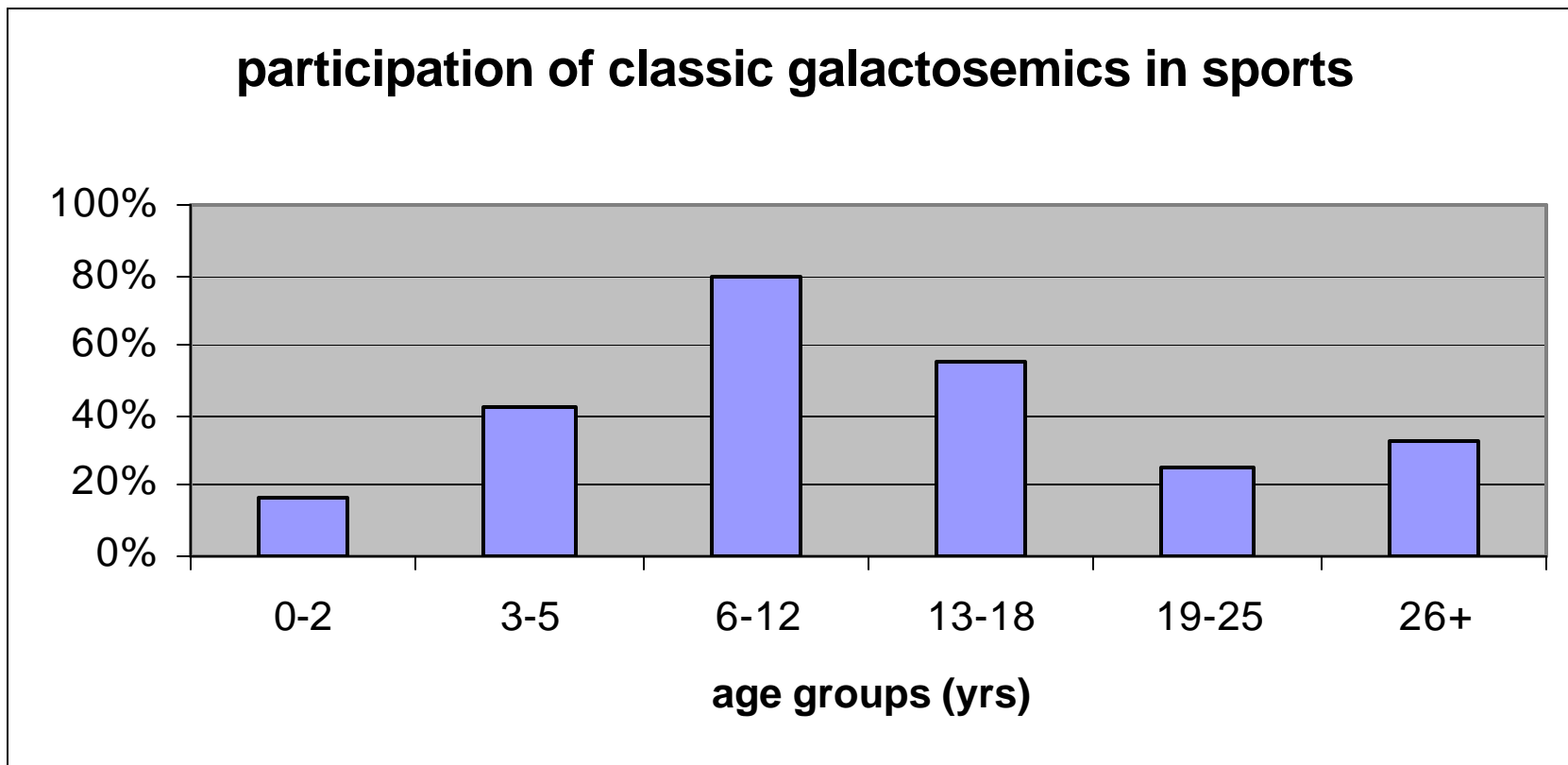
Age Group	Get along w/others?	Have Friends?	Loner?	Activities?
0-2	17%	17%	0%	17%
3-5	67%	67%	0%	48%
6-12	91%	91%	17%	89%
13-18	83%	94%	39%	83%

School and Beyond (age 19+)

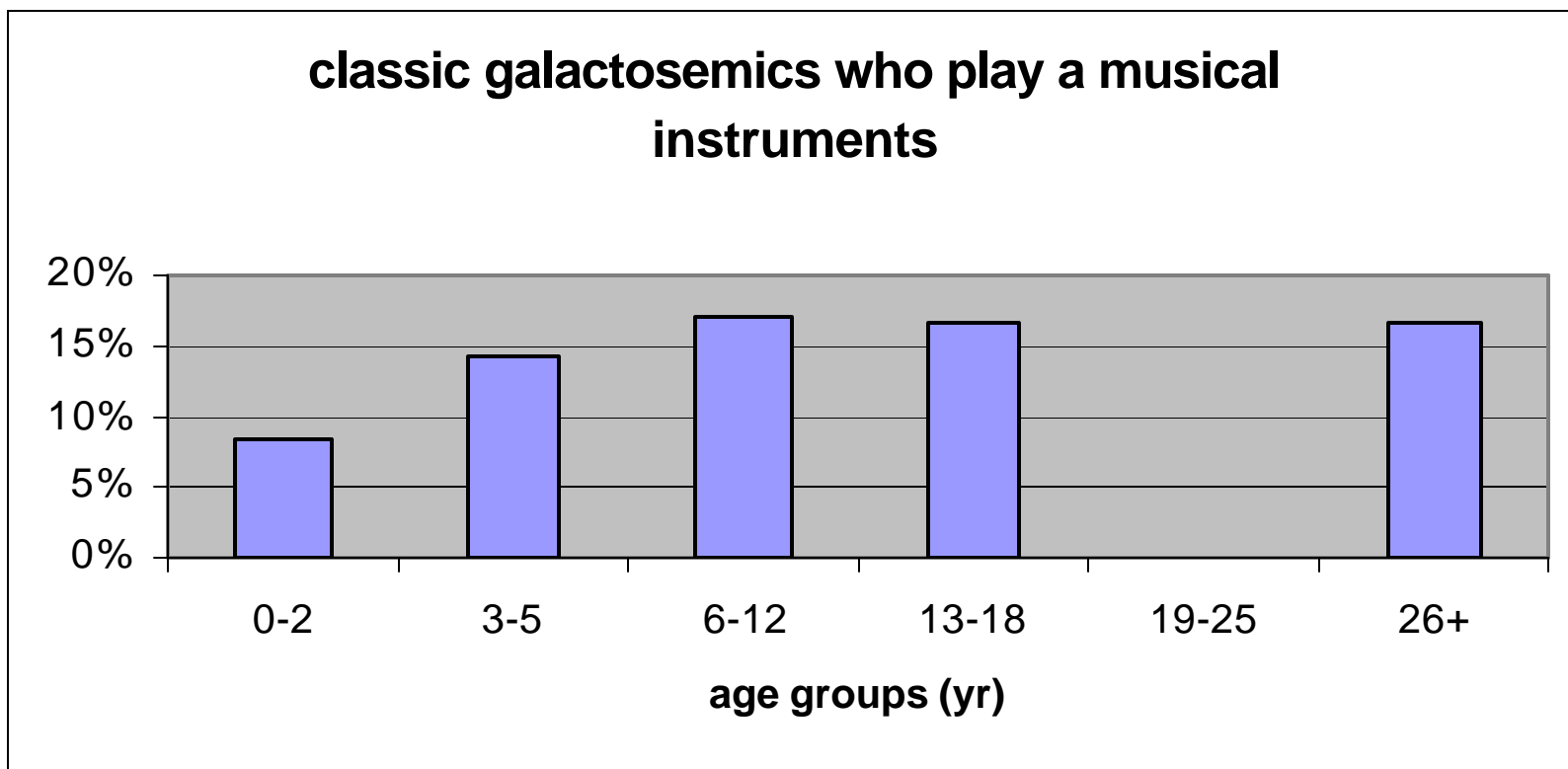
- 17 out of 19 had school trouble
- Average highest level of education ~12 yrs
- 4 have post high school education including college grads
- About 25% drive
- About 50% are employed
 - Restaurant worker, stone mason, dock worker, trucker
 - Dept. Store, Office worker, housekeeper, database analyst

Lifestyle

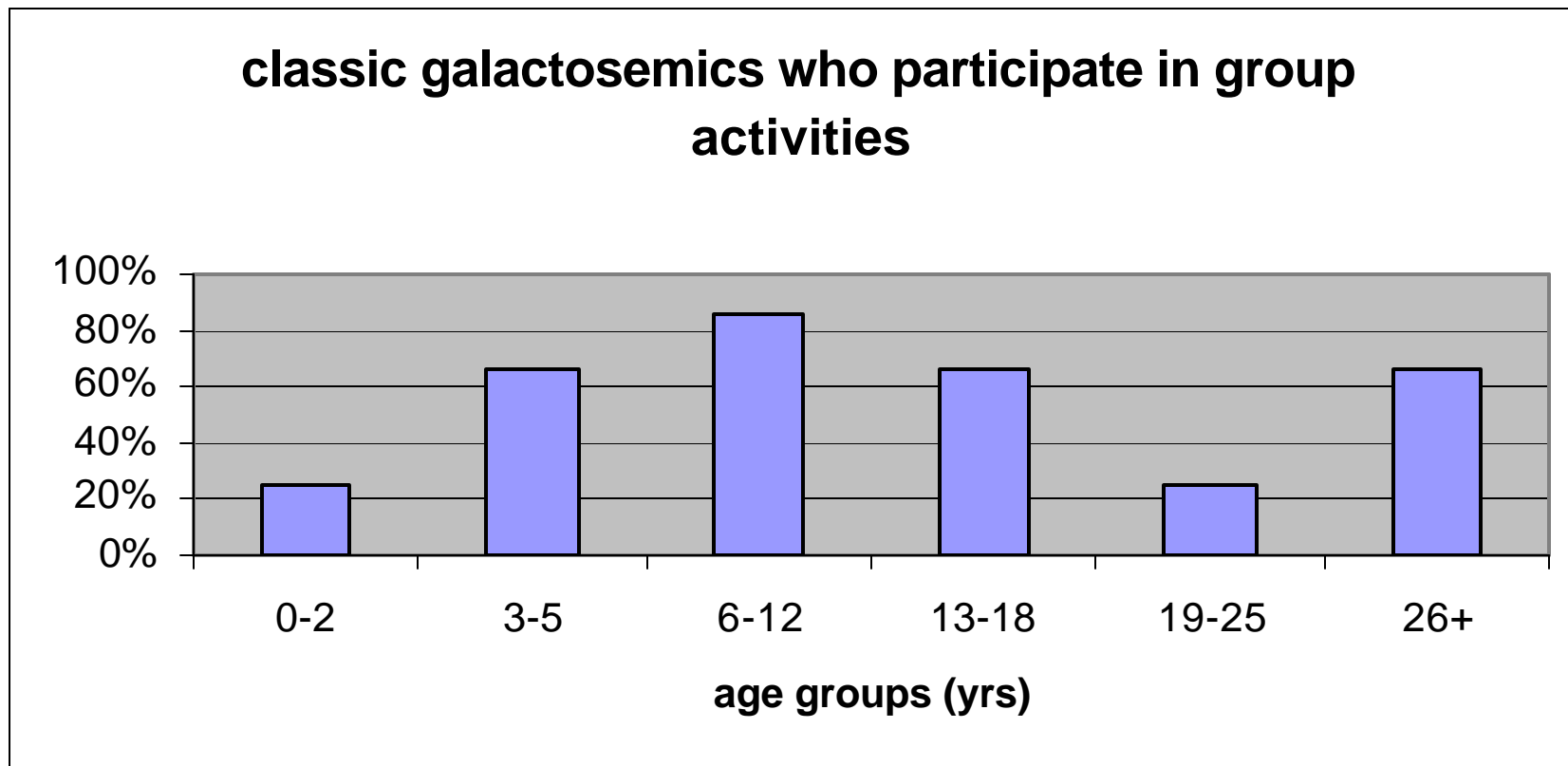
Lifestyle



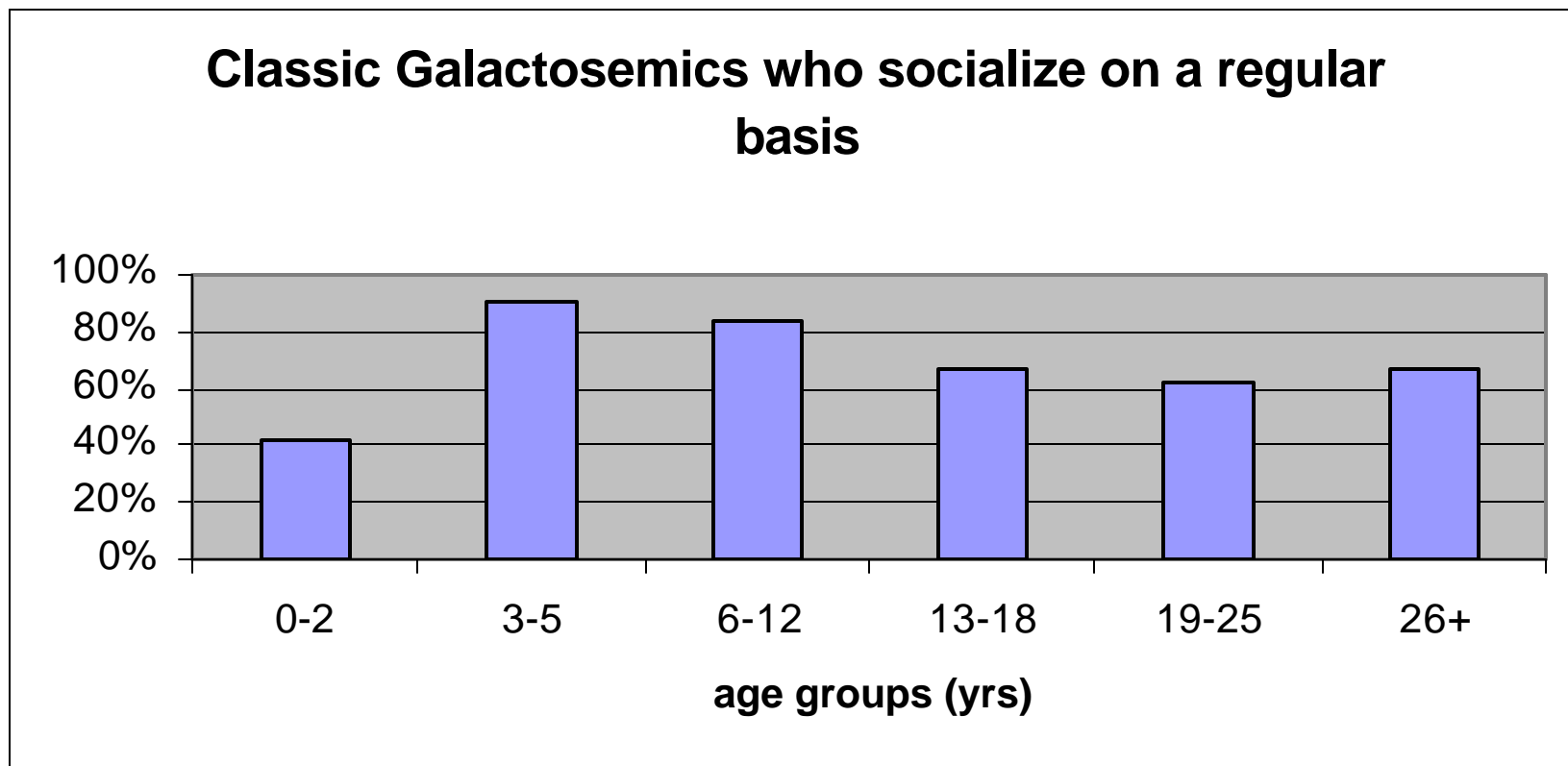
Lifestyle



Lifestyle

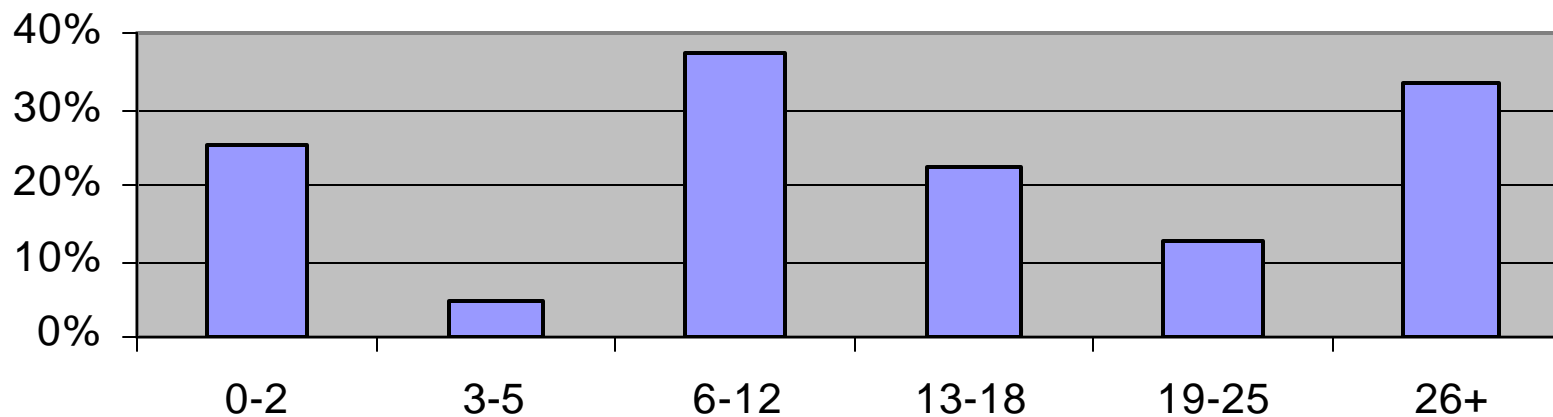


Lifestyle

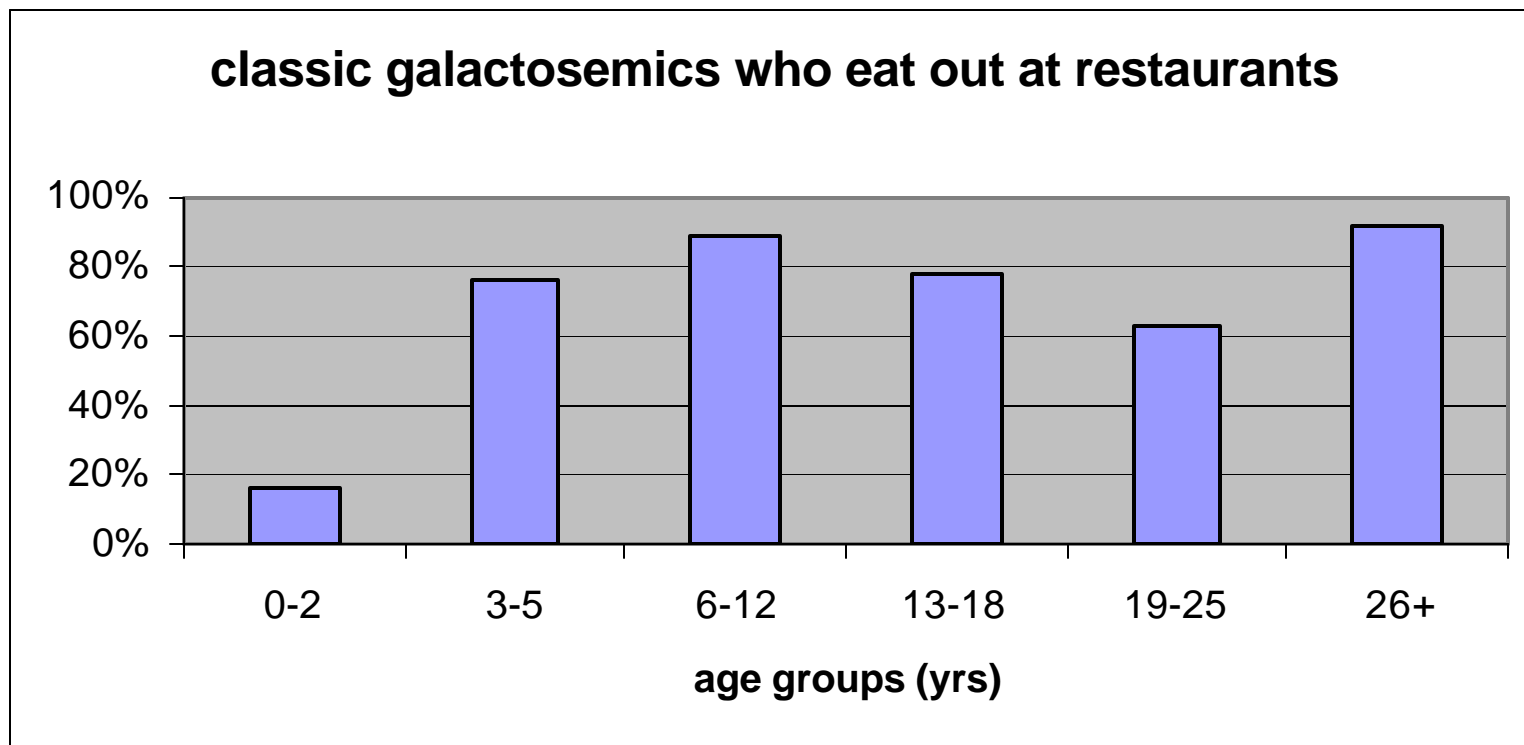


Lifestyle

percentage of respondents who believe that diet is limiting to person's ability to participate in events, activities, or social engagements



Lifestyle



Comments

“My daughter is amazing. She participates in everything she wants to (sports, band, etc.). She has gone on sleep-overs, camping trips ... ” – parent of a 6-12 y.o. daughter

“Our son has no complications from galactosemia. He is an excellent student and quite well adjusted.” – parent of 6-12 y.o. son

Comments

“So far, she has done very well. She is like any other girl her age. If you didn’t know it, you could never tell she has galactosemia”
– parent of 13-18 y.o. daughter

“ We have always celebrated the foods our son can have instead of feeling badly about those he cannot have. Our son is a strong, healthy and active young man. He works very hard for average grades but he enjoys life” – parent of a 13-18 y.o. son

Comments

“ Our daughter is very well liked and always has a great smile for everyone. She is the light of our lives. Galactosemia has not ruled us. We have taken the bull by the horns and made it a part of our everyday lives” – parent of a 13-18 y.o. daughter

“ I worried so much about what my son would do when he grew up, but he has a job that carries his health insurance and he is getting married. He has overcome many of the problems he had. He is a special person.” – parent of 19-25 y.o. son