

If a twentieth part of the cost and pains were spent in measures for the improvement of the human race that are spent in the improvement of the breed of horses and cattle, what a galaxy of genius might we not create! ...Men and women of the present day are, to those we might hope to bring into existence, what the pariah dogs of the streets of an Eastern town are to our own highly bred varieties (Galton 1865).

The man of the moment [was] J. Craig Venter, Ph.D., whose pioneering work to sequence the human genome — our essential code for life — had whetted public appetite for medical miracles in the diagnosis, treatment and prevention of even the most complex of common diseases. "Imagine a world where families leave the hospital with their newborns and take their baby's complete genetic profile with them on a CD-ROM," Venter told his audience. "And imagine a world where your physician has as part of your medical record your genetic code, which can be used to determine, for example, your risk profile for side effects from drugs or other medical treatments. These might be possible in a genomics-based medical system in the near future." (Massoglia 2003)

# PKU neonatal diagnosis

single gene mutation -> retardation & early death

# PKU neonatal diagnosis

single gene mutation

-> early diagnosis

-> effective dietary therapy

# PKU neonatal diagnosis

single gene mutation

-> early diagnosis

-> effective dietary therapy

genetic  $\neq$  unchangeable

# PKU neonatal diagnosis

single gene mutation

-> early biochem diagnosis

-> dietary amelioration

*plus*

ongoing struggle to

secure health insurance coverage for diet

enlist family & peer support for staying on

diet

# PKU neonatal diagnosis

single gene mutation

-> early biochem diagnosis

-> lifelong management of chronic condition

*including*

ongoing struggle

secure health insurance coverage for diet

enlist family & peer support for staying on  
diet

*and*

“maternal PKU”

P/H

PKU posed  
~~PKU~~ Consider  
~~diagnosis~~

low IQs & other deficits

teratogenic  
~~metabolic~~ ~~causes~~  
fetus

diagnosis & care

neo natal

diet

hard to maintain

improvement

decrease

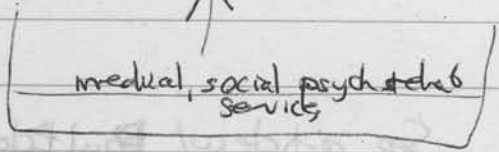
improvement

social support

mandated testing

subsidy + drug classification (till 1972)

religious + ethnic identity



wider social context

bio-medical vs psychological curing + care of change

0

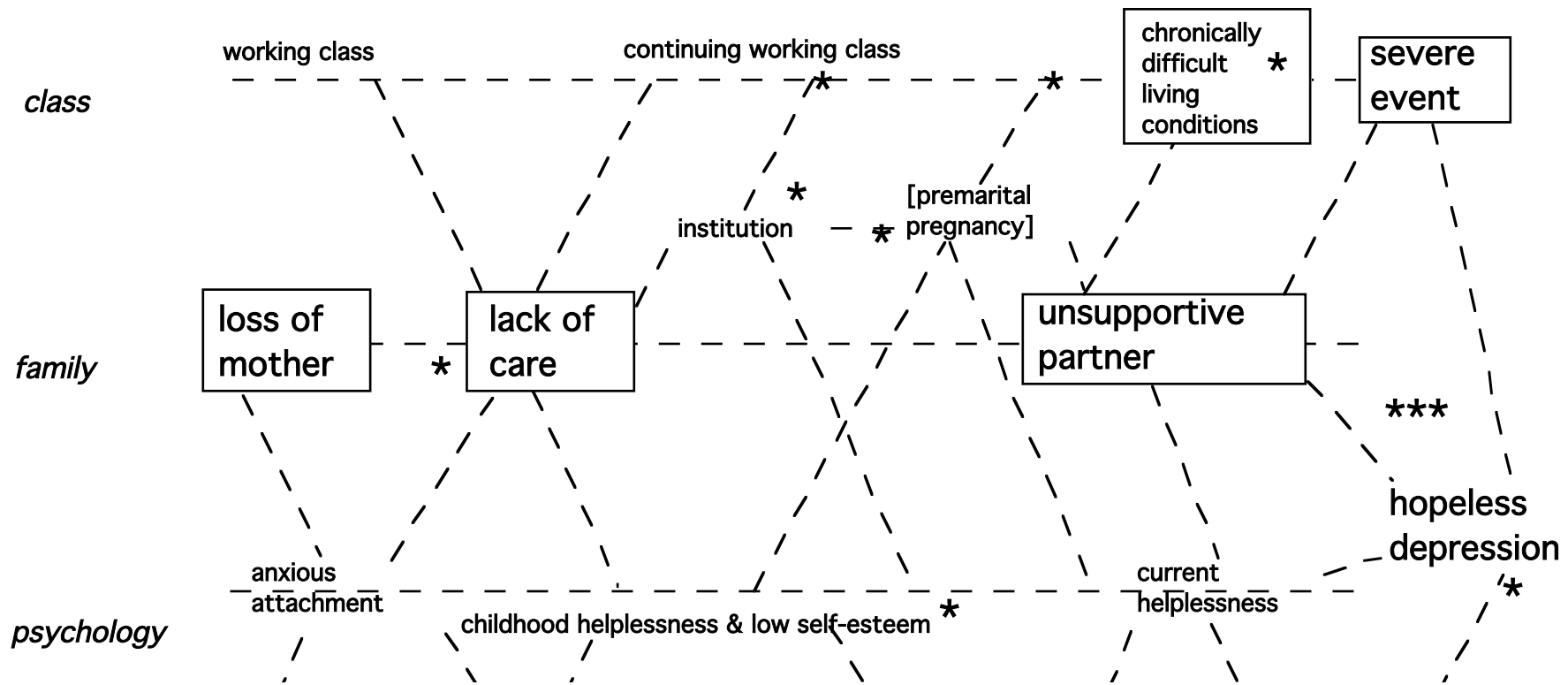
s/6

teenager

young adulthood



Open Question: Optimism re: genetic diagnosis?



*hypothetical  
genetics/  
biochemistry*

\* = point of  
possible  
intervention



