

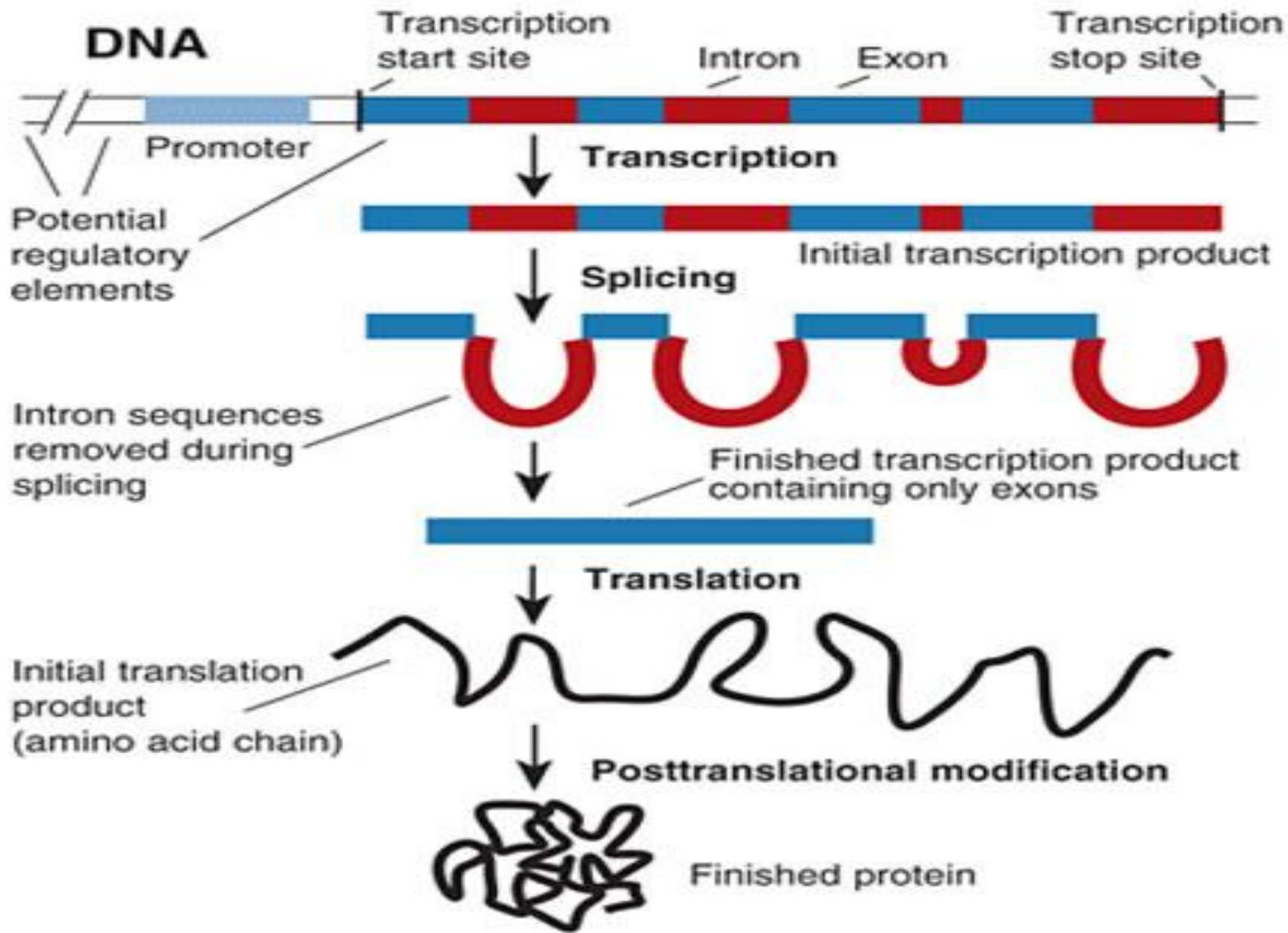
# Genetic counseling in child psychiatry



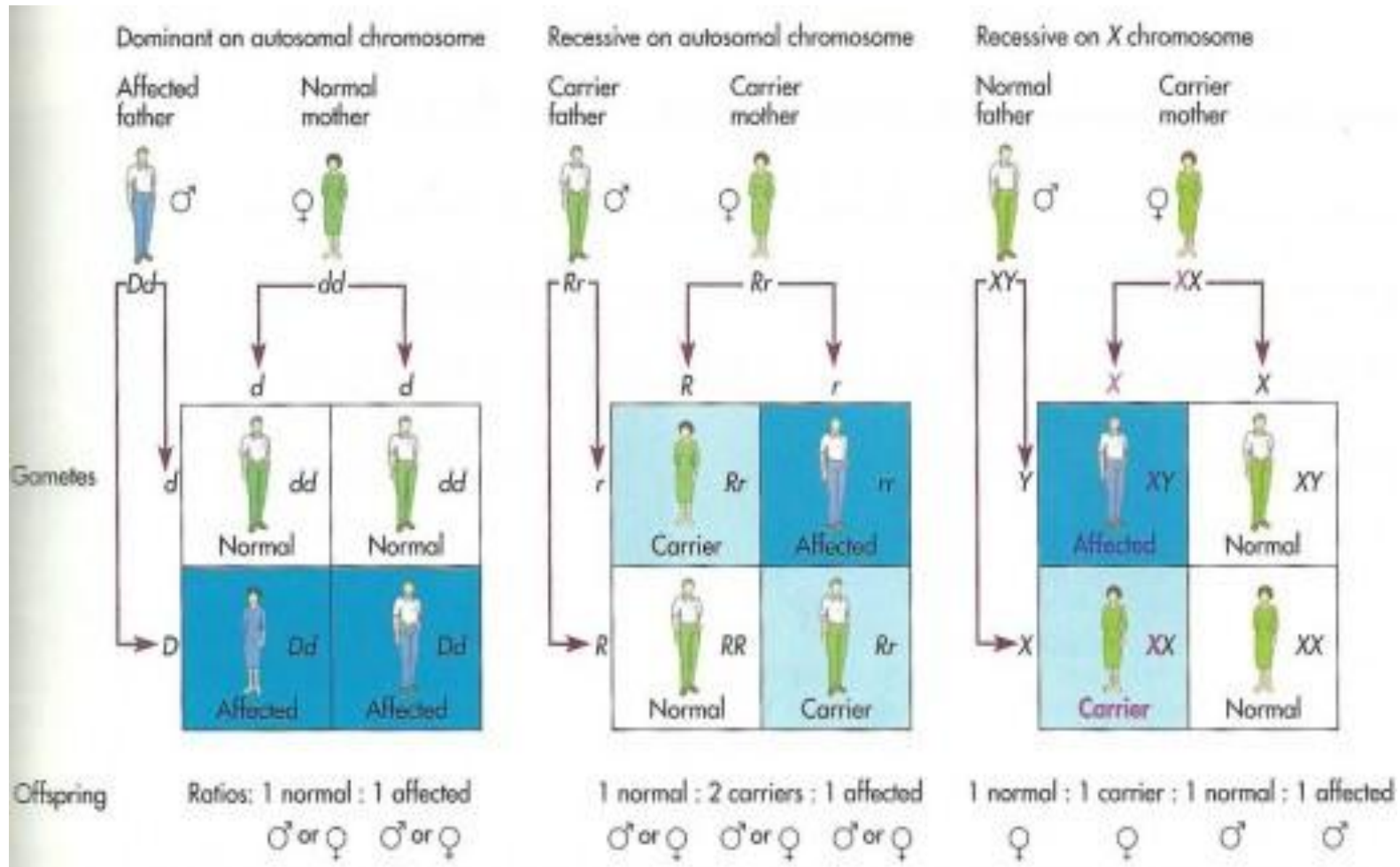
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# Mendelian inheritance



# Why genetic counseling?



# Overview

- ❑ Psychiatry - Genetics
- ❑ Genetic counseling for child psychiatry disorders
- ❑ Genetic counseling for risk of adult psychiatry disorders for children with family history of mental disorders

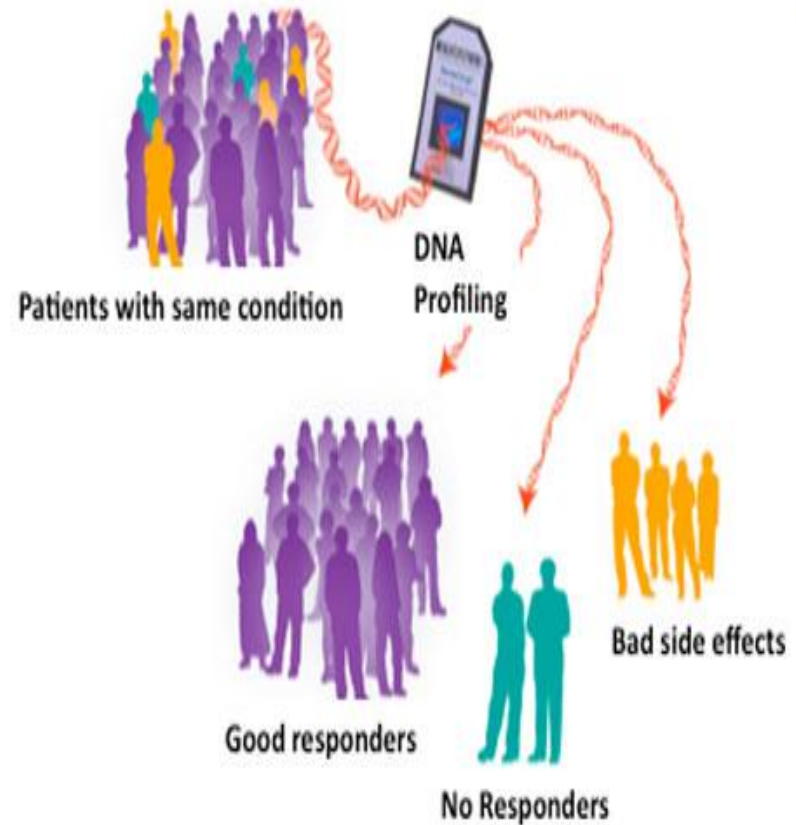
# Psychiatric genetics

- ❑ Many psychiatric disorders have a clear genetic component
- ❑ Multifactorial
- ❑ Mostly multilocus disease
- ❑ Complex inheritance - polygenetic
- ❑ Incomplete penetrance
- ❑ Importance of epigenetics



# Pharmacogenomics

- ❑ Slow, fast or normal metabolizers of CYP 450 enzyme system
- ❑ Predicts the dosage required and risk of side effects
- ❑ Could predict choice of drug treatment in future



# Genetic counseling in child psychiatry disorders

- ❑ ID, Microcephaly, CP
- ❑ Chromosomal disorders
  - Fragile X
  - Down syndrome
  - Prader-Willi syndrome
- ❑ Autosomal dominant – Phacomatosis
- ❑ Autosomal recessive – Inborn errors of metabolism
- ❑ Tourette disorder
- ❑ OCD
- ❑ ADHD
- ❑ SLD
- ❑ ASD



# Genetic Testing

- ❑ Diagnostic- Down syndrome
- ❑ Predictive- Huntington's
- ❑ Carrier Detection- Fragile X
- ❑ Adoption
- ❑ Prenuptial (e.g Tay-Sachs in Jewish Community)
- ❑ Preconception
- ❑ Prenatal

# Factors affecting the uptake of prenatal testing for different single gene condition

<b>Disease</b>	<b>Age of onset</b>	<b>Severity</b>	<b>Treatable?</b>	<b>Predictive certainty of test</b> (% of people developing condition with adverse result) <sup>39</sup>	<b>Uptake of test</b>
Tay-Sachs	From birth	Fatal	No	100%	High levels of uptake in counselled couples in certain communities; many do not seek counselling.
Huntington's	Middle age	Fatal	No	100% but some variation in age of onset (see Box 3.2)	18% uptake in counselled couples. <sup>40</sup> Very low overall.
Phenylketonuria	From birth	If not treated, severe mental handicap results.	Yes	100% (see Box 3.1)	All testing is neonatal. National screening programme.
Neuro-fibromatosis	Child-hood	Variable	Some complications treatable	100% for NF1 but severity varies	1/60 families <sup>41</sup>
Fragile X	From birth	Variable	No	Prognosis may depend on the mutation present	Some at least but epidemiological data unavailable. <sup>42</sup>
Early-onset Alzheimer's disease	Middle age	Severe	Potential treatments currently being explored.	100% for the APP or presenilin 1 or 2 genes (see Box 3.3)	Low uptake of predictive testing for early onset forms. <sup>43</sup>

# Stages of Genetic Counseling

- ❑ Confirm diagnosis
- ❑ Obtain family history
- ❑ Assess recurrence risk
- ❑ Evaluation of consultee
- ❑ Evaluation of potential burdens and benefits
- ❑ Forming a plan of action
- ❑ Follow up

# Issues in genetic counseling

AS YOU CAN SEE FROM YOUR GENETIC PRINTOUT YOU ONLY THINK YOU'RE DEPRESSED WHEREAS YOU ARE IN FACT A JOLLY, HAPPY FULL OF THE JOYS OF SPRING TYPE PERSON!

FRAN



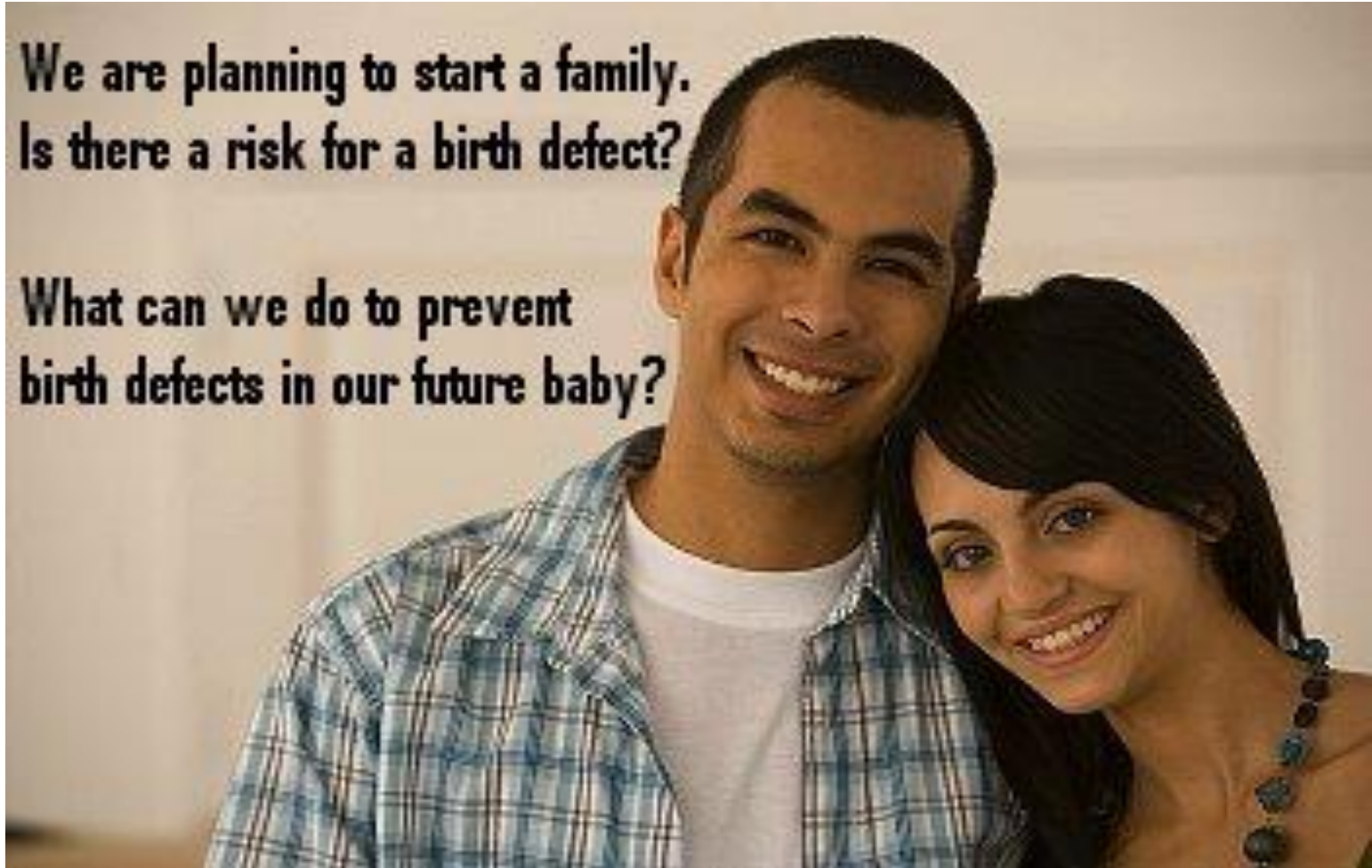
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# Genetic counseling for children with family history of mental disorders

**We are planning to start a family.  
Is there a risk for a birth defect?**

**What can we do to prevent  
birth defects in our future baby?**



# Questions to consider

- ❑ Is the condition genetic in nature
- ❑ Can one be 100% sure it is genetic?
- ❑ Which parent has contributed?
- ❑ Will it affect the unborn child?
- ❑ How can we prevent it?
- ❑ Genetic engineering and gene therapy
- ❑ Adoption
- ❑ Assisted pregnancies- donor sperm or eggs
- ❑ Termination of pregnancy



## 2 Empirical risks of developing schizophrenia for relatives of a person with schizophrenia<sup>17,18</sup>

Relationship to person with schizophrenia	Lifetime risk
General population	1%
First-degree relative	
Identical twin	40%–48%
Fraternal twin	10%–17%
Sibling	9%
Parent	6%–13%
Offspring	13%
Second-degree relative	
Aunt/uncle	2%
Niece/nephew	4%
Grandchild	5%
Third-degree relative	
First cousin	2%

### 3 Estimated lifetime risks for common adult psychiatric disorders

Psychiatric disorder	General population	First-degree relative
Schizophrenia	1%	5%–16%
Bipolar disorder	1%–5%	4%–18% (BPD) 9%–25% (UPD)
Major depression	5%–35% (females) 5%–15% (males)	10%–25%
Obsessive compulsive disorder	1%–3%	10%
Panic disorder	2%–6%	8%–31%

Adapted from: Finn CT, Smoller JW.<sup>36</sup>

BPD = bipolar disorder; UPD = unipolar depression. ◆

# Response and implications

- ❑ Guilt, shame and stigma
- ❑ Strain on family members
- ❑ Opportunity to discuss early signs
- ❑ Prevention strategies
- ❑ Family planning and reproductive decisions
- ❑ Discussion can be complicated in some

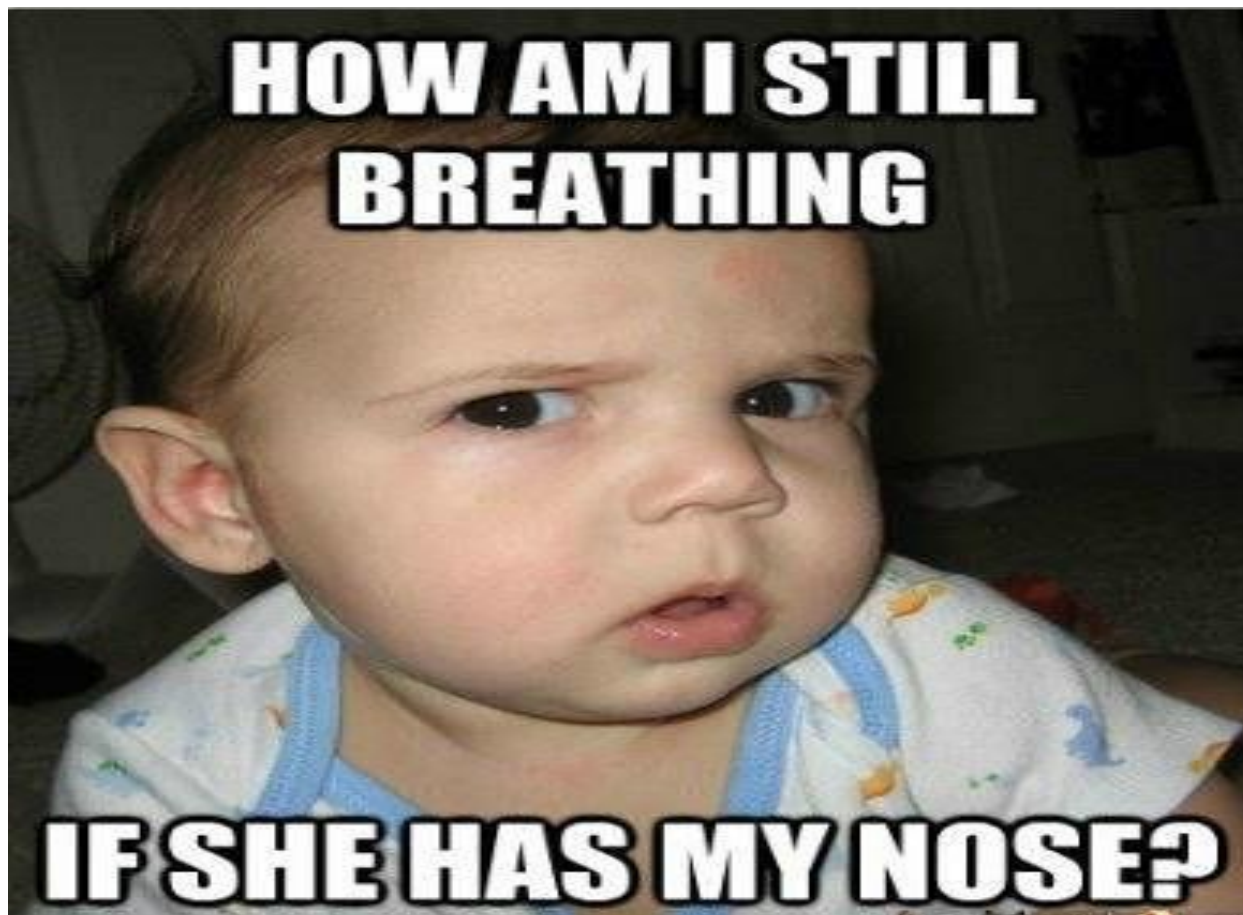
# Challenges in Huntington's

- ❑ Insurability
- ❑ Employability
- ❑ Psychological impact

# Conclusion

- ❑ Genetic factors - important in risk assessment
- ❑ Hope of preventive and Rx strategies in future
- ❑ Genetic counseling helps families understand the complexities of inheritance
- ❑ Facilitates informed decision making
- ❑ Important to keep the ethical, legal and practical issues in mind

Questions?





## Further reading

- ❑ Evaluation of Genomic Applications in Practice and Prevention (EGAPP). [www.egappreviews.org](http://www.egappreviews.org).
- ❑ Clinical application of genetic information about mental disorders. <http://nuffieldbioethics.org/wp-content/uploads/2014/07/Mental-disorders-and-genetics-the-ethical-context.pdf>
- ❑ Genetic counseling for psychiatric disorders. MJA:185(9);507-510.
- ❑ Kaplan & Sadock's comprehensive textbook of psychiatry. Chapten1.18. Population Genetics and Genetic epidemiology in Psychiatry