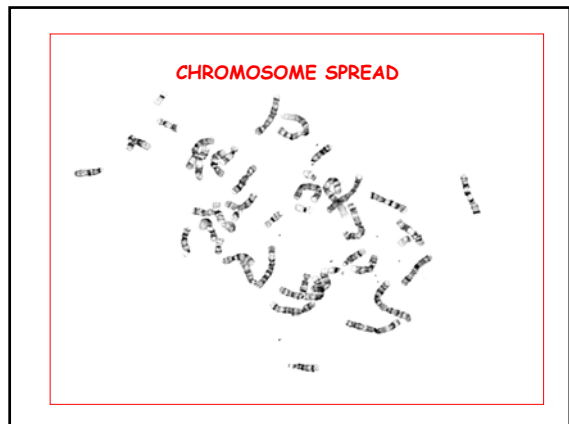
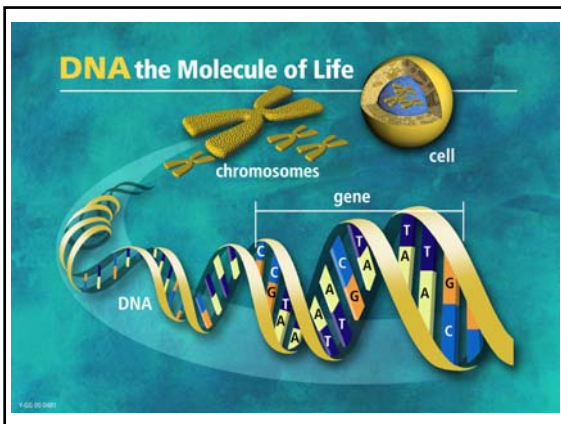
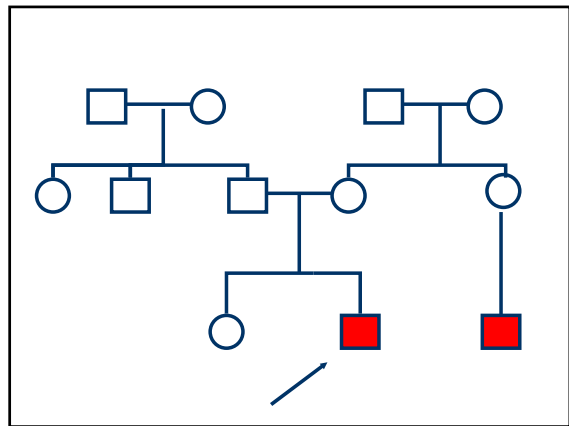
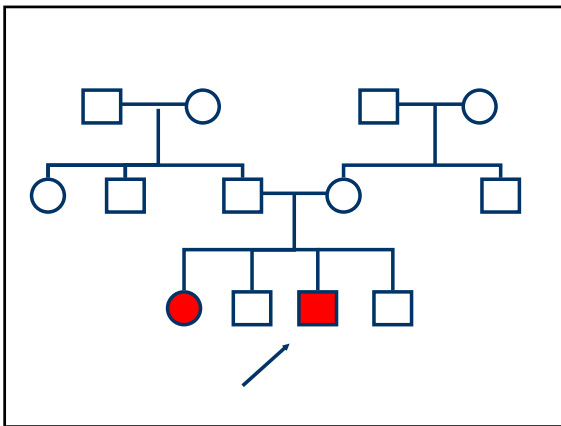
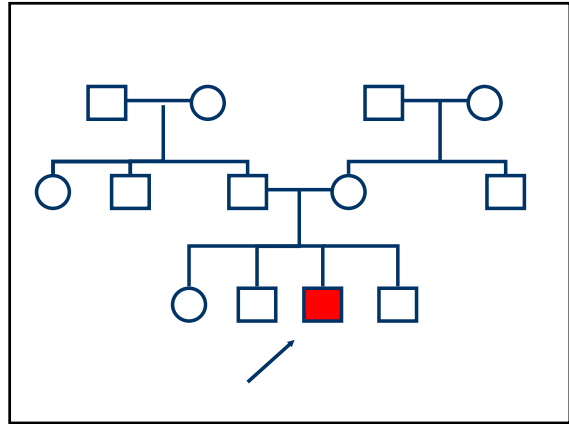
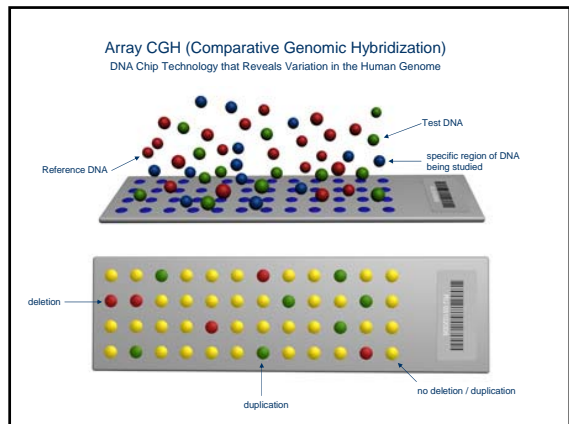
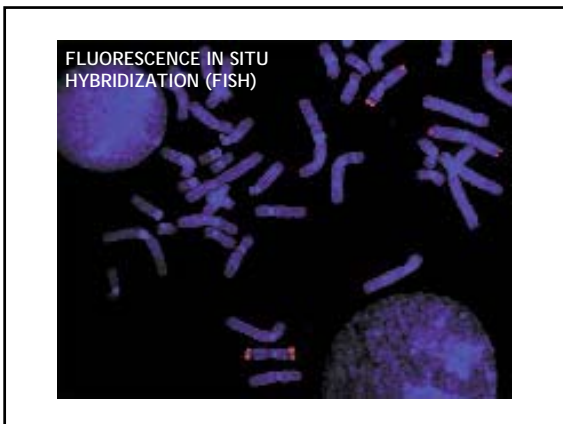
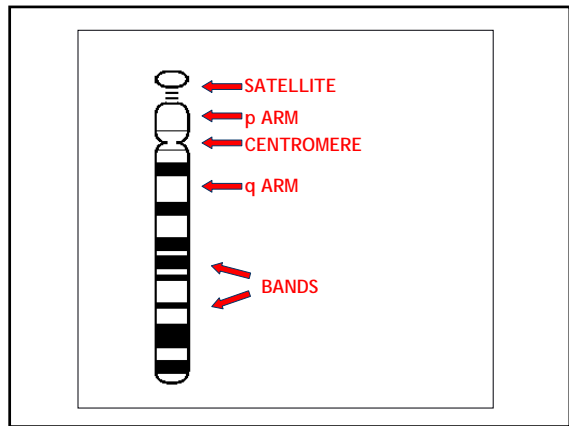
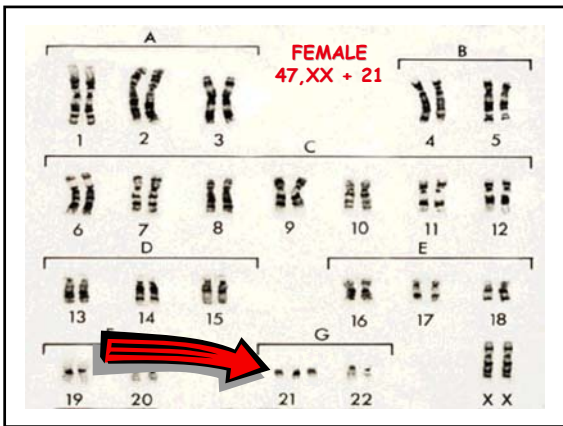
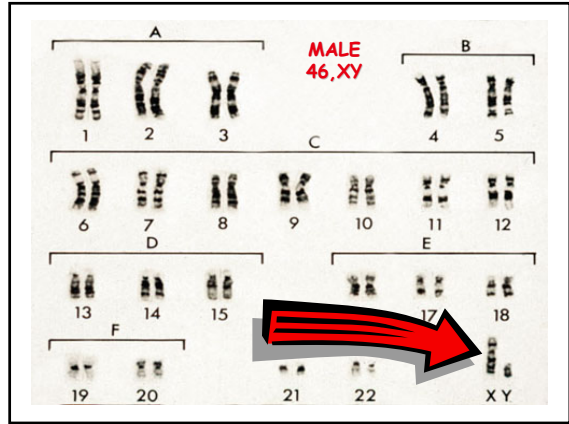
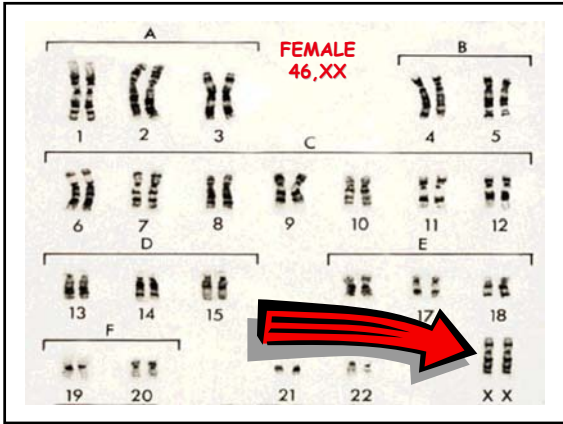
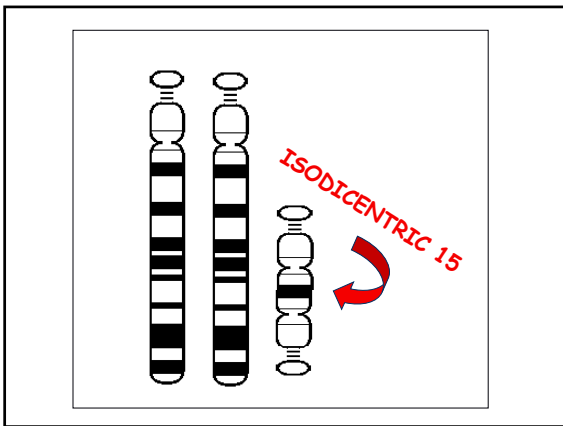
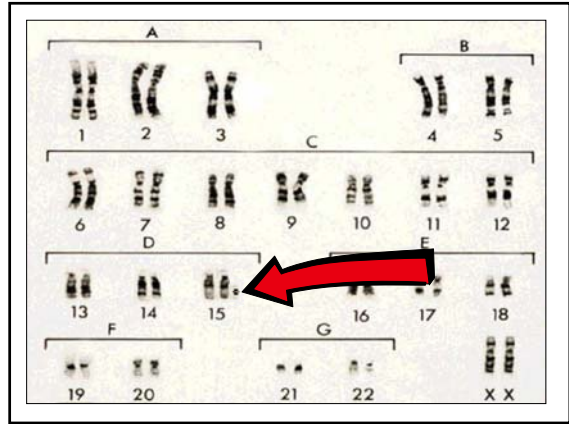
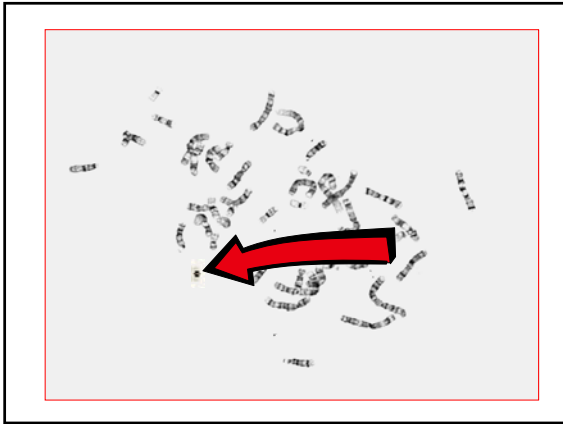


# Basic Concepts in Genetics and Diagnosis

Brenda Finucane, MS, CGC  
Genetic Services at Elwyn  
www.elwyngenetics.org








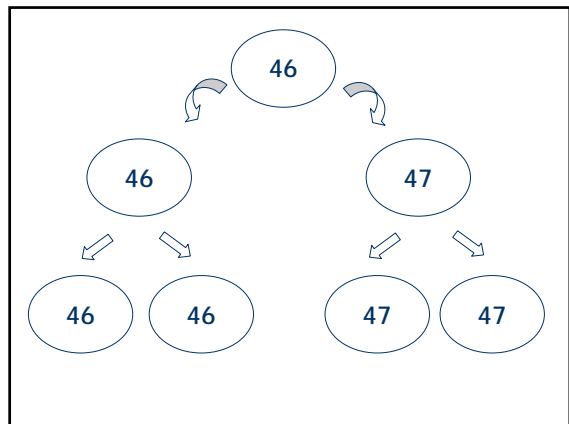
**ISODICENTRIC 15**

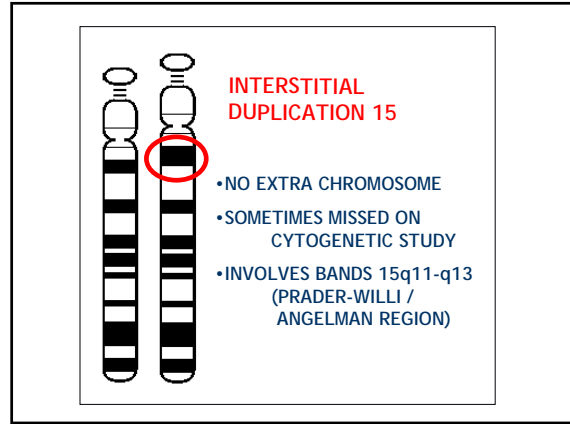
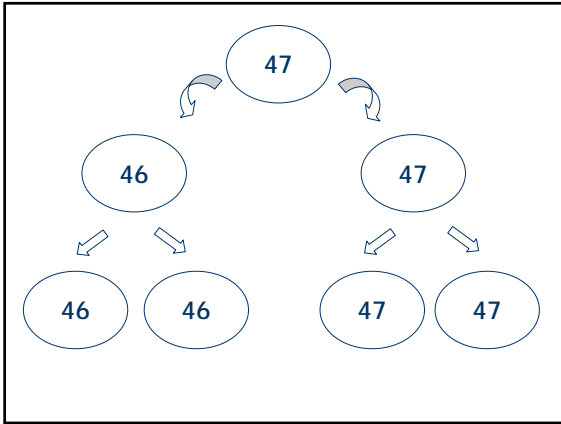
a.k.a.: idic(15)  
 inverted duplication 15  
 supernumerary marker  
 bisatellited supernumerary  
 tetrasomy 15  
 partial trisomy 15  
 related disorder: interstitial duplication 15

**ISODICENTRIC 15**



- EXTRA BISATELLITED CHROMOSOME
- DUPLICATION OF ALL OF THE p ARM
- DUPLICATION OF PART OF THE q ARM
- WITH TWO NORMAL 15s, RESULTS IN TETRASOMY OF 15p AND PARTIAL TETRASOMY OF 15q
- INVOLVES BANDS 15q11-q13 (PRADER-WILLI / ANGELMAN REGION)
- SOMETIMES INVOLVES MULTIPLE COPIES OF THE EXTRA MATERIAL
- SOMETIMES SEEN IN MOSAIC FORM





**VOCABULARY**

PHENOTYPE  
 CRITICAL REGION  
 MOSAICISM  
 MATERNAL / PATERNAL  
 SPORADIC  
 RECURRENCE

**Duplication 15q Syndrome**

FITTING GENETIC DIAGNOSES INTO CONTEXT IN A BEHAVIORAL WORLD!

**What is Etiology?**

- Developmental disorders are symptoms for which there are many different etiologies
- Genetic and / or medical factors play a role in the etiology of most developmental disorders

**Why is Etiology Important?**

- Genetic counseling for families
- Alleviates guilt, misconceptions
- Anticipation of medical needs
- Insight into behavior, learning styles
- Support groups

## Etiological Diagnoses

In the school setting:

- Etiological diagnoses often considered irrelevant
- Educational / psychiatric diagnoses determine services and treatment approaches

## Psychiatric Diagnoses

ADD ANXIETY DISORDER OCD  
ADHD OPPOSITIONAL DEFIANT DISORDER  
AUTISTIC DISORDER BIPOLAR DISORDER  
INTERMITTENT EXPLOSIVE DISORDER ID IMPULSE CONTROL DISORDER

## Diagnostic Alphabet Soup



Reaction of parent upon hearing that her child has yet another diagnosis

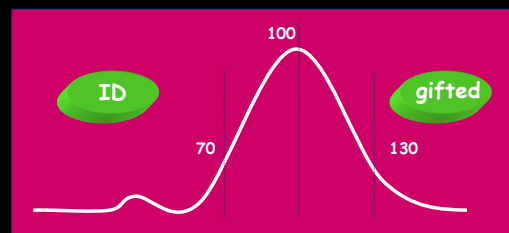


## Intellectual Disability (ID)\*

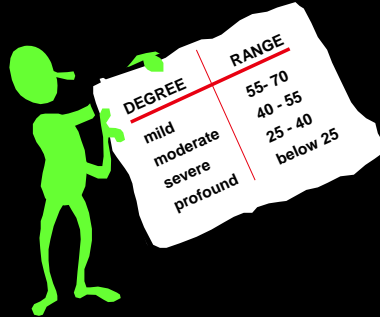
- Significantly subaverage intellectual functioning (IQ of 70 or below)
- Deficits in adaptive functioning
- Onset prior to age 18

\*formerly mental retardation

## Distribution of IQ scores



## Degree of Intellectual Disability



DEGREE	RANGE
mild	55-70
moderate	40 - 55
severe	25 - 40
profound	below 25

## Dup 15q in the School Setting

- Diagnostic confusion abounds!
- Psychiatric / behavioral symptoms:  
Found in association with many genetic disorders, including dup 15q syndrome
- Causes vs. Symptoms: Important for parents and professionals to understand distinction

## Dup 15q in the School Setting

- Educational and behavioral diagnoses, not genetic diagnoses, determine eligibility and services within the school setting
- Individuals with dup 15q syndrome often meet criteria for one or more behavioral / educational diagnoses
- Use these diagnoses for everything they're worth, realizing that the genetic condition is the underlying cause of the behavioral symptoms

## Dup 15q in the School Setting

- School districts and teachers are unlikely to be familiar with dup 15q syndrome
- This does NOT necessarily mean they are unable to provide excellent services
- An open mind, willingness to learn about dup 15q, and a creative approach to meeting a child's needs are just as important as experience with the syndrome