1 st International Early Childhood Conference
Eurlyaid Annual Conference 2012

Braga, 14.09.2012

Early Intervention in Fragile X Syndrome

Ana Apolónio (ARSA) **& Vítor Franco** (U. Évora)

Projecto "PTDC/CPE-CED/115276/2009"

COMPETE
QUADRO DE REFERÊNCIA ESTRATÉGICO NACIONAL
UNIÃO EUROPEIA
Governo da República Portuguesa
FCT

Fragile X Syndrome

- Most common form of inherited developmental disability, although very often under-diagnosed;
- FXS was first documented in 1943 by the physician James Purdon Martin and the geneticist Julia Bell and was formerly known as the Martin-Bell syndrome.;
- It is a genetic condition caused by a change in the code of a single gene on the X chromosome;

- It may occur in both sexes and in all populations, regardless of ethnic group and socio-economic levels;

Fragile X Syndrome

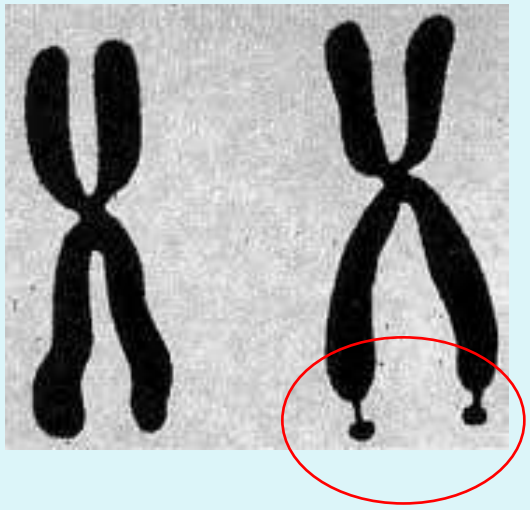
- It is more prevalent in males because females have another X chromosome to compensate to a varying degree for the one affected.

- Recent statistics indicate that 1 in 2500-4000 males and 1 in 4000-6000 females are affected and approximately 1 in 260 females and 1 in 300-800 males are carriers of a pre-mutation gene;

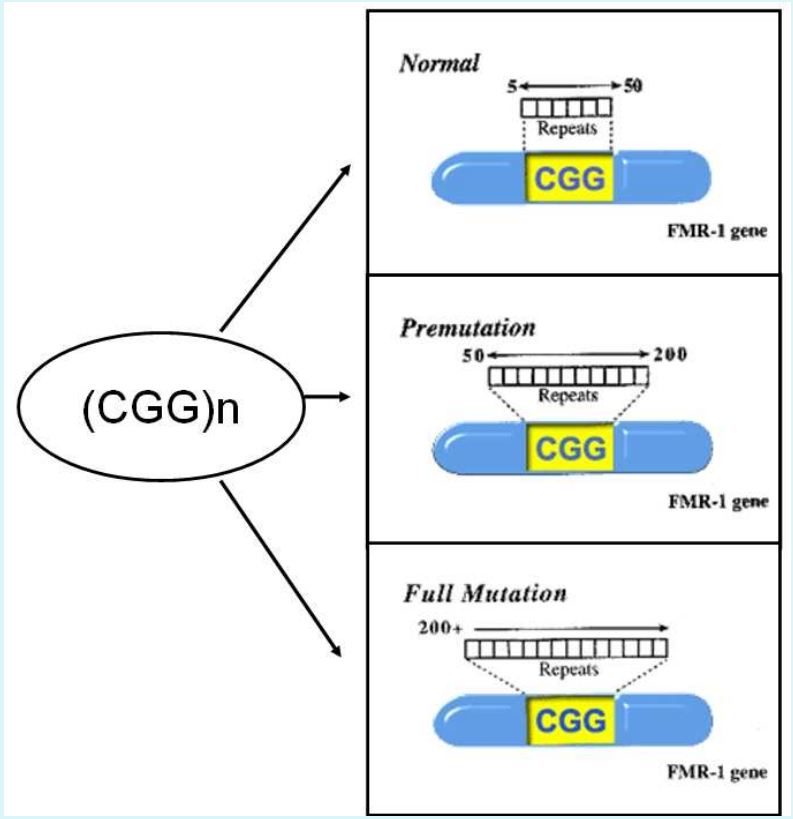
Fragile X Syndrome

- It is caused by a dynamic mutation in the gene FMR1 (Fragile Mental Retardation), located on the longer arm of **X chromosome**, that appears to be **broken**;

- This broken appearance is caused by an expansion of a small part of the sequence formed by the repetition of a trinucleotide CGG, in the beginning of the FMR1 gene.



The number of repeats of the sequence CGG determines the gene's ability to produce FMRP (Fragile Mental Retardation Protein)



Fragile X Syndrome

- An altered *FMR1* gene can be passed on without symptoms, so many people are unaware that they have it;
- As a result, a premutation form of the *FMR1* gene can be passed through a family for generations, without any symptoms;
- However, with each generation, it becomes more likely that the premutation gene will expand its number of repeats to become a full mutation gene, which would also increase the severity of symptoms.

Fragile Mental Retardation Protein

– Its exact role is not yet fully studied;

– Research suggests that is involved in forming pathways in the brain;

– People without enough FMRP have many malfunctioning neural connections.

Common features

– Developmental delay

–Learning disabilities

– Poor eye contact

– Hiperactivity

– Language delay

–Short attention span

– Physical features: broad forehead, prominent ears, macro-orchidism,...

–Autistic like features: gaze aversion, social anxiety, hand-flapping and hand-biting sensory defensiveness

–Fine and gross motor delay

–Coordination problems

Early childhood

–Great variability in developmental and behavioural features – there is no evident phenotype at birth ;

– Few research in the area, mostly with boys over 6;

– 3 dimensions:

–Social interaction
– Attention deficit hyperactivity disorder
– Autistic behaviours

– Diagnostic confirmation through genetic tests, based on detection of developmental and behavioural delays.

Early childhood

–Parents often raise concerns about the child's development before the first birthday;

–Usually formal diagnosis comes during the 2.º year of life, or later.

Importance of early detection

Importance of early diagnosis

- Genetic screening and counselling for informed decisions;
- Early intervention;
- Minimizes family uncertainty and frustration and provide information about FXS;
- Helps family to understand the child behaviour;

Impact on families

- Great impact on family system;
- Genetic implications of FXS causes increased family stress (especially mothers);
- Feelings of guilt associated with FXS;
- Needs of information, emotional support.

Meeting Johnny

– Referred to Early Intervention Program by the physiatrist with 18 months

– Developmental delay, especially in intellectual and language areas;

– Started EI less than a month after referral;

– Genetic screening confirmed FXS at the age of 2 (full mutation);

– Older sister also diagnosed with FXS, mother carrier;

– Family history of intellectual retardation.

Starting EI

Strengths

– Good social interaction;
– Liked to play with known children and with animals;
– Gentle and loving;
– Good eye and physical contact;
– Good reaction to visual and hearing estimulation;
– Good gross motor skills;
– Receptive verbal skills better than expressive skills

Weaknesses

– Poor everyday life skills
– Language delay (**parents main concern**)
– Sometimes has aggressive behaviour towards other children
– Attention and concentration problems
– Social ansiety

Starting EI

Family

– Good social support network ;

– Good relationship between family members;

– Uncertainty about the child’s problem and the future;

–Mother’s health problems;

– Concerns about the daughter's learning difficulties;

– Low income;

Early Intervention provided:

– Help to John's inclusion in kindergarten with educational support;

– Speech therapy, addressing language problems, through routine-based strategies and individual work;

– Psychological support for John's mother:

  – help to understand his behaviour and problems

  – information about FXS

  – emotional support

Ending EI

- Better adjustment and adaptation;
- Less agressive behaviours;
- Some improvement in the attention and concentration problems;
- Sphincter control (at 5);
- Independent in daily routines;
- Increased expressive language skills;
- Difficulties to understand complex orders and abstract concepts;
- Parents feel more confidence in John's capacities to learn, but increased concerns about his sister.

Conclusions:

–Increasing Importance of understanding group of children with chromosomopaties;

–Need to strengthen links between EI and genetic services for screening and genetic counseling;

–Defining specific intervention according to FXS variability characteristics

–Family support:

- Providing information about each child unique developmental and behavioral features
- FXS information
- Referral to genetic counselling
- Emotional support

Bibliographic references

Bailey, D. B., Hatton, D. H., & Skinner, M. (1998). Early developmental trajectories of males with fragile X syndrome. *American Journal on Mental Retardation, 103*, 29–39.

Bailey, D. B., Hatton, D. D., Mesibov, G., Ament, N., & Skinner, M. (2000). Early development, temperament, and functional impairment in autism and fragile X syndrome. *Journal of Autism and Developmental Disorders, 30*, 49–59.

Bailey, D. B., Skinner, D., & Sparkman, K. L. (2003). Discovering fragile X syndrome: family experiences and perceptions. *Pediatrics, 111* (2), 407-416.

Bailey, D.B., Skinner, D., Hatton, D. & Roberts, J. (2000). Family experiences and factors associated with the diagnosis of fragile X syndrome. *Developmental and Behavioral Pediatrics, 21*, 315-321.

Hagerman, R.J. (2002). *The Fragile X Syndrome: Diagnosis, treatment, and research*. Baltimore: The Johns Hopkins University Press.

Miranda, P. L., Bailey, D. B., Roberts, J. E., & Hatton, D. D. (2004). Developmental screening and detection of developmental delays in infants and toddlers with fragile X syndrome. *Journal of Developmental and Behavioral Pediatrics, 25* (1), 21-27.