Klinefelter Syndrome: A Small Sample Retrospective Analysis

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Introduction

Klinefelter Syndrome (KS) is characterized by the presence of supernumerary X-chromosome and thus a 47,XXY karyotype. Although it's the most common numerical chromosomal disorder in males (150/100.000♂)[1] this syndrome remains underdiagnosed, with only about 25% of patients being identified, and only 10% during childhood.

Methods

Retrospective analysis of 11 KS patients followed-up in the Endocrinology Department of Coimbra's Hospital and Universitary Center. The registered data included education and occupation, time and motif of diagnosis, co-morbidities and treatment. Patients were divided in two groups: A – diagnosis at pediatric age, n=8; B – diagnosis in adulthood, n=3.

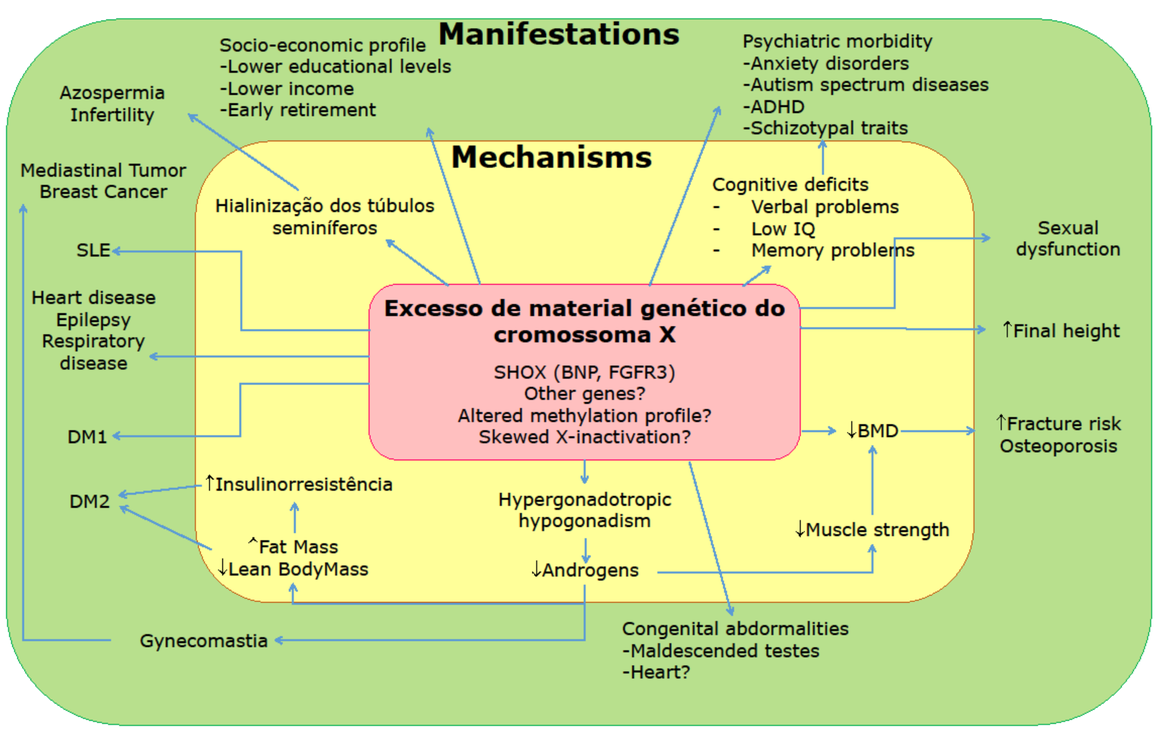

Figure 1 - Relation ship between etiopathogeny and possible clinical manifestations of KS. SLE – Systemic Lupus Erythematosus, DM – diabetes *mellitus*, IQ – intelligence quofeicient, ADHD, attention deficit/hyperactivity disorder. Adapted from Groth KA, et al.[3]

Resultados

11 patients were evaluated, with a mean age of 34,45±14,2 years, a mean age of diagnosis of 19,7±16,5 years and a mean education of 7,14 years (n=7).

Group A - Diagnosis at pediatric age

- **N = 8**
- Mean age of diagnosis : 10,5±3,1 years
- Diagnosis in the context of:
  - -Learning difficulties (n=8)
- Puberty induction therapy (n=8)
- Mean final stature:170,3±9,8cm

On the last appointment:
Mean age: 24,5 years
- Gynecomastia - 5 patients (62,5%)
- Osteopenia – 2 patients (25%)
- No documented changes in carbohydrate metabolism
- Cognitive deficit – 8 patients (100%)
- 4 patients finished highschool, 2 with primary education. 1 illeterate patient (n=7).

Group B - Diagnosis in adulthood

- **N = 3**
- Mean age of diagnosis: 42,7±14,2 years
- Diagnosis in the context of:
  - -Study of fertility (n=1)
  - -Hypogonadism hypogonadotropic workup (n=2)
- Mean final stature: 177,3±3,8cm

On the last appointment:
Mean age: 47 years
- Gynecomastia - 2 patients (66,7%)
- Osteopenia – 2 patients (66,7%)
- Osteoporosis – 1 patient(33,3%)
- DM2 – 2 patients (66,7%) – both with difficult metabolic control, proliferative retinopathy
- Psychiatric disorders– 2 patients (66,7%)
- Cognitive deficit– 2 doentes (66,7%)
- 1 patient finished highschool, now employed. 2 didn't finish primary education, unemployed.

Conclusion

Cognitive impairment was found in 90,9% of patients and was the most frequent comorbidity, with educational and professional impact. Gynecomastia and osteopenia were frequent as well. Timely diagnosis can result in a better care with proper follow-up and regular screening of possible comorbidities.

References:
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