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Epidemiological Profile of Trisomy 21 in Senegal

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Abstract

Introduction: Trisomy 21 (T21) is the most common genetic cause of learning disabilities and congenital malformations in humans. The genetic background is well established and maternal age is the main risk factor implicated. The aim of this study was to describe the epidemiological profile of T21 in the Senegalese population.

Patients and method: This was a 7-year retrospective observational study, from November 2016 to October 2023. We considered all cases of T21 in the cohort of the genetic consultation at the Albert Royer Children's Hospital. The data collected were analysed using SPSS 21.

Results: We collected 218 cases of T21 among the 378 patients seen at the consultation. According to the inclusion criteria, 205 cases of patients with T21 were selected. The sex ratio was 0.99. Consanguinity was found in 17.1% of patients and T21 in 2.9%. At least one spontaneous abortion was found in 27.8% of the mothers. The median age of the mothers at conception was 37 years. The 35-39 age group was the most represented (35.6%). A deep organ malformation was found in 70.9% of cases, dominated by cardiopathies (60.9%), the most frequent of which were: atrioventricular canal (44%), atrial septal defect (21.1%), persistent ductus arteriosus (18.3%) and interventricular septal defect (15.6%). **Conclusion:** Maternal age, the main risk factor identified for T21, seems to present a different profile, notably younger in our series.

Keywords: Trisomy 21; Epidemiological profile; Maternal age; Senegal

Introduction

Trisomy 21 (T21) or Down's syndrome is the most common genetic cause of intellectual disability and congenital malformations in the human population. It is the most common chromosomal anomaly, with an incidence of around 1/600 newborns [1,2]. It is now more than 60 years since the genetic context was identified. The study of genetic markers has shown that T21 results in 90% of cases from an error during maternal meiosis [2,3]. Two risk factors for non-disjunction have been observed in young women: the absence of genetic recombination between homologous chromosomes and the presence of an exchange in the telomeric position [4,5]. As maternal age increases, non-disjunctions linked to pericentromeric exchanges appear [5-8]. In addition, it is well known that recurrence is greater in young mothers and that the incidence increases considerably with advanced maternal age. However, the biological basis of the effect of maternal age remains largely unknown. The biological reasons underlying this spectrum of clinical and experimental observations are still unknown and constitute major biological

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technical platform leave little room for certain aspects of medical practice. Antenatal screening for accessible genetic DRPDOLHVLFOGLJDGPDJHPHWRIGHYHORSPHWDO delay are among the aspects that are not yet taken into account in health priorities.

Our series represents more than half of the medical JHHWLFVFRVOWDWLRFRKRUWLWKRIDVHVOWKRJK known to be the most common viable chromosomal anomaly LKPDWWKLVLUHHFDORHGRHVRWVHHPVvFLHW to assess the extent of the problem in our setting. In fact, we note on the one hand the existence of other specialised consultations in the facility, the absence of a single, FHWUDOLVHGSDWLHWOHGWKHRVWRIDUHRUHDRHQLHVL result, many patients are only seen in the organ speciality or specialities concerned, except in the event of a therapeutic impasse associated with developmental delay, or in the event of a family case. Furthermore, our series, compiled from the cohort of programmed outpatients, does not take into account FDVHVRI7KRGLHGGULJKRVSLWDOLDWLRUHIHUUHGIU transferred to our facility. Although little is known about its IUHHFLRUFRWULHVLWUHPDLPFKKLJKUWKDWWKH JUHVLWKHHVWOHHWRWKHWRWKHWLPLGLWRIDWHDWDO screening, which is not systematic, and also to the non-existence of abortion, which is illegal in our practice except where there is a vital maternal risk.

In our series, we noted a balanced sex ratio of 0.99, whereas the main trend in the literature is towards an imbalance in favour of boys [8,9], with the exception of the IRHWRSDWKRORJLFDOVHULHVIROORLQJDERUWLRMIR a slight female predominance was found [10]. In fact, meta-analysis of 55 publications providing the sex ratio, this male predominance was found in all studies with a high rate of FWRJHHWLFFRUPDWLRRIPRUHWKDOLNHRUVHULHV [9].

1HDUORHSUHJDFLVL[LRUVHULHVDV the result of a consanguineous union, but no case of trisomy DVIRG LWKHSDWLHWVVLEOLJ7KLVLVSSRUWHGE FWRJHHWLFVKLFKIRGWKHDFFLGHWDOIRUPRI7R hereditary) in all patients tested, as widely reported in the literature [3,6,7].

0RUHWKDDDUWHUIWKHPRWKHUVKDGKDG DWOHDVWRHVSRWDHRVDERUWLRSULRUWRFEHS median age of the mothers at the time of conception was 37 years, which is close to the average in our series (36 years). 7KHDJURSWKHPRVWUHSUHVHWHGEDRWEH correlated with fertility in this population. In fact, the Senegal Demographic and Health Survey for 2023 reports a peak in fertility in the 25-29 age group [117KLVGLVFUHSDFKHOSV to maintain advanced maternal age as a strong predisposing IDFWRUIRUWKHRFFUUHFHRI7RHYHUDVERUHODWHG

Hetch and Hook (Figure 3) [3,12], this theoretical exponential evolution observed in large samples of Caucasian populations does not maintain the same speed in Figure 4, which is UHSUHVHWDWLYHRIRUVHULHV7KLVLVSUREDEOHGHWRWK drop in the birth rate over the age of 39 in Senegal as a whole [11], whereas in France, for example, the overall prevalence has risen from 14/10,000 in 1978 to 23/10,000 live births in 2005, at the same time as the average age of mothers has increased from 26 to 30 over the study period [3].

6LPLODUOLWKDVDOUHDGEHHVKRWKDWRIDU RI7DUH UHODWHG WR PDWHUDO DJH PRGHOOLJWKH prevalence in the general population solely on the risk associated with maternal age, an approximation bias is introduced by applying a risk model to the entire population LRO RIEDVHV7KLVDSSUR[LPDWLRLVPDGH DOO existing publications on the subject using this methodology [3, 12].

In the search for organ malformations (cardiac, digestive, ULDUQ17DWOHDVWRHDVIRGLRIEDVHVM reported in the literature [13-15], cardiac disease was the PRVWIUHHWLRUVHULHVVLPLODUOLWHUPVRIWKHKHD

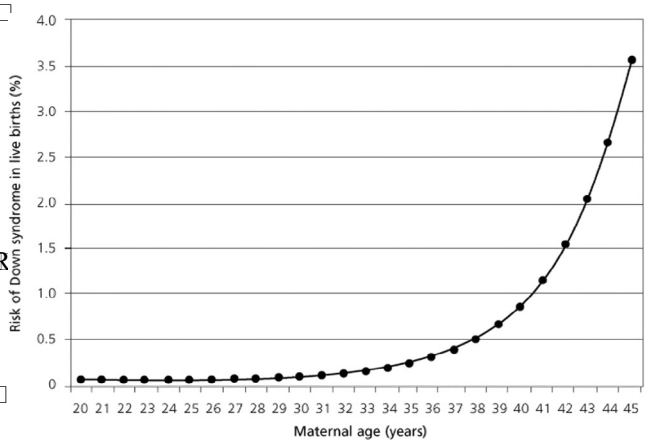

Figure 3: Expected prevalence of trisomy 21 at birth as a function RIPDWHUDODJHDFFRUGLQJWRHFWDGRRN0DWHUDODJHVSHFLF live birth prevalence of Down's syndrome - Hecht and Hook) [12].

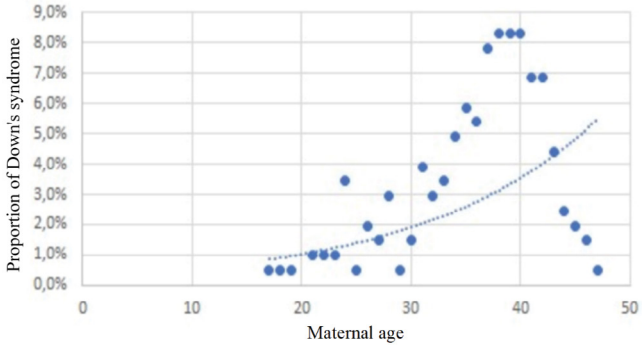

Figure 4:)UHHRI7ULVRPDFFRUGLQJWRWKHPRWKHUVDJH at conception.

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Biigé DIENG YJK, et al., Fortune J Health Sci 2024
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atrioventricular canal remained the most frequent cardiac defect in our series (44%), where we also reported a high proportion of persistent ductus arteriosus (18.3%), as found in the large American series [9], but not in Europe [14,15].

Conclusion

Maternal age, the main risk factor identified for T21, seems to present a different profile, notably younger in our series drawn from a black African population.

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