

A Study on the Incidence of Various Genetic Syndromes in Kozhikode District, Kerala, India with special reference to Down Syndrome

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Abstract

Data of Kozhikode district was studied with respect to the occurrence of various genetic syndromes for the time period 2010-2011. A total of 63 syndromes have been reported in Kozhikode in the above time period as per the data collected from major hospitals of the study area. The most common conditions were Down Syndrome and Duchenne Muscular Dystrophy. The percentage of occurrence was found to be more than double for DS and about seven times higher for DMD when compared with previously published data. In case of DS, both parents were below 32 years in 41% of cases, contrary to the previously published data.

Keywords: Genetic syndromes, Kozhikode, Down syndrome, Duchenne Muscular Dystrophy

1. Introduction

Genetic disorders are among the leading causes of infant morbidity and mortality (Kaur, Singh, 2010). Currently around 4,000 genetic disorders are known, with more being discovered. They can vary in severity, from being fatal before birth to requiring continuous management; their onset covers all life stages from infancy to old age.

Common genetic disorders in Indian population are chromosomal disorders such as Down syndrome, Thalassemia, Hemophilia and Duchenne muscular dystrophy (Smith et al., 2005).

The present study found that a total of sixty four genetic syndromes with varying frequencies apart from skeletal deformities and developmental delays have been reported in Kozhikode district in the time period of 2010-2011

2. Collection of data

The following data were collected from the major hospitals of Kozhikode district:

- Diagnosis of genetic syndrome
- Details of karyotyping done
- Parental age
- Family history of the disease

The time period of the study was 2010-2011.

3. Results and Discussion

Two hundred and seventy four cases were analyzed and the following results were obtained.

The most common conditions which bring people to genetic clinics are developmental delays and skeletal deformities. They form about 11.314 and 16.058 % of the total patients. Apart from these, there were sixty four genetic syndromes found out of which most common among the inhabitants of Kozhikode are Down syndrome (24.088 %) and Duchenne muscular Dystrophy (5.474 %). The syndromes are listed in Appendix.

As per a study conducted in 2010 by Kaur and Singh, Down syndrome comprised of about 11.4% of the total cases whereas in the case of Kozhikode, the figure rises to 24.088%, more than double the former. Also, Kaur and Singh reported that muscular dystrophies comprised of 0.8 % whereas in our case, DMD and Gower syndrome together constitute 7.299% of the cases. Turner syndrome comprised of 2.3% in the previous study but in our study it turned out only to be 1.46%. Another difference is in the case of Fragile X syndrome which formed a good 9.3 % in the mentioned study, whereas in our study, it counts only less than 1%.

Of all the 274 cases analyzed, a total of 23.5% were born in families which had some history of the disease or out of consanguineous unions. Of this, 13% constituted the cases which showed family history and 10.5 % constituted those born out of consanguineous marriages. It has been known that parental consanguinity is often associated with a higher frequency of genetic disorders and congenital malformations in their progeny (Verma and Mathew, 1983; Mukherjee, 1994). It is well supported by similar studies carried out focusing on individual malformations reported in closely related couples (Kesavan et al., 1978; Karimi-Nejad, 1995).

Also, among these 23.5% cases, 17.02% patients had an affected sibling. It has been proven earlier that couples who already have an affected child are 13 times more likely to have another affected child (Verma and Mathew, 1983; Mukherjee, 1994).

3.1 Down syndrome

Down syndrome was diagnosed in 66 out of the 274 cases analyzed and it leads the list as the most common.

Birth order of DS showed a higher number of first and second born (Jyothy et al., 2000). But in the present study, only 3.175% of the patients were first born children, 28.57% were only

children, 30.159% were second borns and 38.095% had two or more elder siblings.

When the maternal and paternal age were analyzed, an interesting yet appalling pattern was observed. The mean maternal age was reported to be 30.34 years and mean paternal age was 31.04 years (Jyothy et al., 2000). But in the present study, the mean maternal age was found to be 28.44 and the mean paternal age was 34.23. In 41.176 % of the cases, both parents were aged below 32. In 32.353%, only one parent was aged above 32, in all cases the father and only in 29.412 % cases, both parents were aged above 32. Contrary to the accepted fact that the chances of occurrences of trisomies increases with increased maternal age, here the result obtained shows that there is only very less relation, if not any, between maternal age and occurrence of Down syndrome.

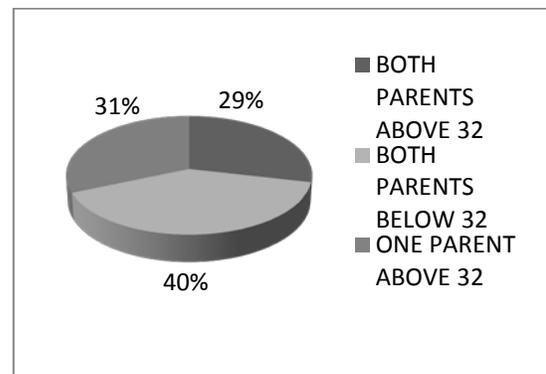


Fig.1 Comparison of incidence of Down syndrome and Parental age

In the case of maternal age, largest number of patients (35.294%) belonged to the age groups 21-25 and 26-30. Contrary to the observed results, the incidence of Down syndrome has been associated with the advanced maternal age and has been confirmed in most of the cases (Roizen and Patterson, 2003).

In all of the 66 Down syndrome cases analyzed, only 4.545% of cases had any previous family history of the disease. All other cases showed no trace of the disease even in extended pedigree charts.

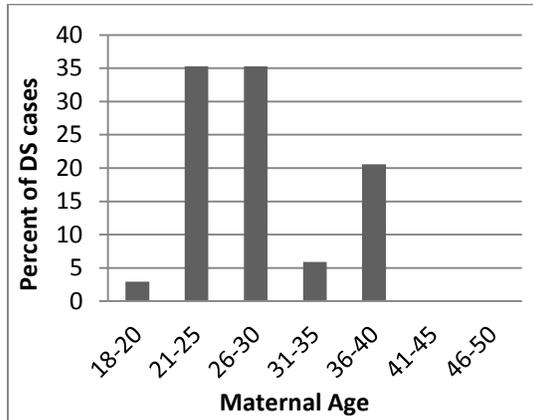


Fig.2 Comparison of Incidence of Down syndrome and Maternal age.

3.2 Duchenne Muscular Dystrophy:

DMD is the second most prevalent genetic syndrome (5.474 %) in Kozhikode.

In the case of DMD patients, family history seems to be a very important factor in determining the occurrence of the disease. In about 33.33 % of the cases, a family history of the disease, either paternal or maternal lineage, was present. Consanguineous marriage was seen in 6.66 % of cases.

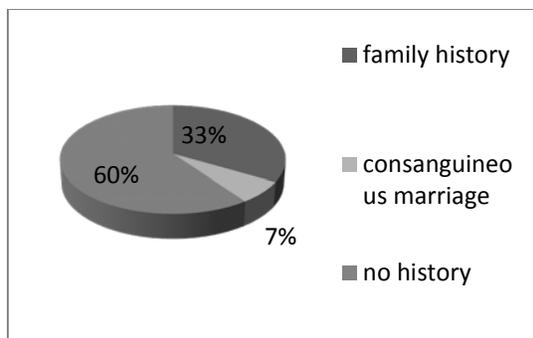


Fig.3 Family history and Consanguineous marriage among DMD patients

The pedigree chart in Fig.4 shows the family history of DMD in the maternal side, giving evidence that it is an X- linked recessive disorder.

In the case of Gower’s syndrome also, three of the five cases showed maternal lineage family

history and one showed consanguineous marriage.

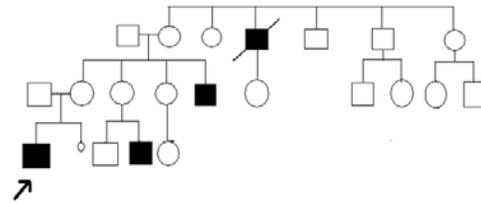


Fig.4 Pedigree chart of a DMD male

Other prevalent diseases were William syndrome (2.920%), Di George syndrome (2.190%) and Edward syndrome (1.825%). The only Phenylketonuria case was born out of a consanguineous marriage. Cystic fibrosis, which is usually common in people of Caucasian origin (Ratjen, Doring, 2003), has been found to have a prevalence of 1.095% of the total cases, out of which, one patient had three late siblings, all of whom had died of CF.

The major sex linked anomalies were Klinefelter syndrome (1.095%) and Turner syndrome (1.460%).

4. Conclusion

The study had concentrated on the occurrence of genetic syndromes among the inhabitants of Kozhikode district. The study revealed some results, which, else would have remained unnoticed.

The occurrence of more than sixty five genetic syndromes is a very strong indicator of the district’s genetic disease burden. In the case of Down syndrome, the finding that in most of the cases, parental age is below 32 is alarming. Due to small sample size, it can’t be determined whether the result is significant or not. Further research need to be done in the area to investigate it. DMD has proven to be the second most prevalent genetic disease. Being an X-linked recessive disorder it shows strong correlation

with familial history and consanguineous marriage. Various other diseases have also been observed including cystic fibrosis which is most prevalent among Caucasians.

Appendix

The following are the genetic syndromes that have been found among the population of Kozhikode district:

1. Achalasia Cardia
2. Achondroplasia
3. Acute disseminated encephalomyelitis (ADEM)
4. Arthrochalasia
5. Bannayan-Ruvalcaba-Riley Syndrome (BRRS)
6. Beckwith Wiedemann Syndrome
7. Brachydactyly
8. Bruton's Syndrome
9. Caffey Disease
10. Cartilage Hypoplasia
11. Cleft Palate And /Or Lip
12. Club Foot
13. Congenital Bilateral Squint
14. Cri Du Chat Syndrome
15. Cystic Fibrosis
16. Di George Syndrome
17. Down Syndrome
18. Duchenne Muscular Dystrophy
19. Edward Syndrome
20. Familial Hypercholesterolemia
21. Fragile X
22. Gower Syndrome
23. Hemolytic Anemia
24. Hemophilia A
25. Horseshoe Kidney
26. Hunter Syndrome
27. Hypertelorism
28. Hypogammaglobulinemia
29. Hypomelanosis
30. Hypotonia
31. Joubert's Syndrome
32. Kabuki Makeup Syndrome
33. Klinefelter Syndrome
34. Klippel Feil Syndrome
35. Klippel Trenaunay Weber Syndrome
36. Krabbes Disease Demyelinating Disorder
37. Marfan Syndrome
38. Morquio Syndrome
39. Multiple Hemivertebra
40. Nager Acrofaciodysostosis
41. Noonan Syndrome
42. Parry Romberg Syndrome
43. Phenylketonuria
44. Pierre Robin Sequence
45. Poland Syndrome
46. Prader Willi Syndrome
47. Proteus Syndrome
48. Prune Belly Syndrome
49. Pseudohypoparathyroidism
50. Rubinstein Taybi Syndrome
51. San Filippo Syndrome
52. Sotos Syndrome
53. Spinal Muscular Atrophy Type I
54. Spondylothoracic Dysostosis
55. Stickler Syndrome
56. Syndactyly
57. Tar Syndrome
58. Tay Sach's Disease
59. Treacher Collins Syndrome
60. Turner Syndrome
61. Waardenburg Syndrome
62. Werdnig Hoffman Syndrome
63. William Syndrome
64. Wiskott Aldrich Syndrome

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