

GENODERMATOSES IN PAEDIATRIC AGE GROUP

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Pattern of genodermatoses in paediatric age group was studied. The relative incidence of genodermatoses in paediatric dermatology out patient department was 0.62% . The commonest genodermatoses observed was ichthyosis.

Key Words: Genodermatoses, Ichthyosis

Introduction

Genodermatoses can be broadly classified into three categories namely chromosomal disorders, single gene disorders and multi-factorial disorders. Most of the genodermatoses show single gene or Mendelian type of inheritance comprising of autosomal dominant, autosomal recessive and X-linked recessive type of inheritance.¹

Thus genodermatoses form an important group of disorders. The study of these disorders will be helpful in prenatal diagnosis of these disorders and in genetic counselling. Due to paucity of studies in this field we have undertaken this study in the paediatric age group to know the pattern of genodermatoses.

Materials and Methods

In the present study of twelve months from 1.7.92 to 30.6.93, 5464 paediatric patients of various skin ailments attended paediatric dermatology OPD of Kalawati Saran Hospital, New Delhi. Out of these 34 were diagnosed as cases of genodermatoses.

Detailed history including family pedigree analysis and clinical examination were carried out in all the patients. Blood examination including Hb, TLC, DLC, ESR, liver function tests and kidney function tests were done in all

cases. Urine was subjected to routine and microscopic examination. Radiological examination was done in specific cases. Histopathological and electron microscopic examination of skin was done in relevant cases. Karyotyping was also performed in specific cases.

Results

A total of 5,464 paediatric dermatological patients were seen out of which there were 34 (0.62%) patients of genodermatoses. The commonest genodermatoses was ichthyosis (19 cases) followed by acrodermatitis enteropathica (4 cases) and 3 cases each of palmoplantar keratoderma and epidermolysis bullosa. There were 2 cases of cutis laxa and one case each of tuberous sclerosis, pachyonychia congenita and hypomelanosis of Ito.

Mode of inheritance was autosomal dominant in 17 cases (50%), autosomal recessive in 11 cases (32.3%) and X-linked recessive in 6 cases (17.7%).

Family history was present in all (100%) cases of X-linked type of genodermatoses and 29.4% cases of autosomal dominant type of genodermatoses. As expected no case of autosomal recessive type had positive family history.

Out of 19 cases of ichthyosis, there were 10 (52.7%) cases of autosomal dominant type comprising of 9 cases of ichthyosis vulgaris and one case of bullous ichthyosiform

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erythroderma. There were 3 cases (15.7%) of autosomal recessive type consisting of one case each of lamellar ichthyosis, collodion baby and Sjogren Larsson syndrome. There were 6 cases (31.6%) of X-linked type of ichthyosis.

Discussion

In the present study the incidence of genodermatoses was 0.62%. The exact incidence of these disorders has not been reported in the literature but it is thought that at least 1% of all live births had disorder inherited in a simple Mendelian fashion.²

Fifty percent patient had autosomal dominant type of disorders. 32.3% had autosomal recessive type of disorders while 17.7% had X-linked type of disorder. Similar frequency of inheritance for such type of disorders had been reported by other workers.³

In cases of autosomal dominant disorders classical vertical transmission could be seen in 29.4% cases. Such low incidence can be explained on the basis of: 1) new mutation which is frequent in autosomal dominant traits, 2) low expressivity of gene in parent, and 3) extramarital paternity.⁴

In cases of X-linked recessive disorders characteristic pattern of inheritance was observed. Only males were affected and the trait was being transmitted from carrier mothers to their sons.⁴

In this study there were 19 cases (55.8%) of ichthyosis. Autosomal dominant ichthyosis (52.6%) was the commonest followed by X-linked (31.5%) and autosomal recessive (15.7%) ichthyosis. Such pattern of ichthyosis has been well documented.^{5,6}

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