

PHENOTYPIC ABNORMALITIES OF EYELASHES AND EYEBROWS IN GENETIC SYNDROMES

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Abstract

The aim of my study is to analyze the phenotypic abnormalities of eyelashes and eyebrows in the case of some genetic syndromes while tracking the abnormalities type and the frequency with which they are encountered, and also comparing these data to the specialty literature. A number of children were studied found in the records of the Department of Medical Genetics in Oradea who were diagnosed with various genetic disorders. The incidence of some minor anomalies was tracked, such as those of eyelashes and eyebrows. The results were compared with those of different clinical studies. The incidence of eyelashes and eyebrows phenotypic abnormalities has increased in patients with genetic diseases, according to the data published by different authors. Many genetic syndromes are associated with specific hair abnormalities, that is why this investigation is considered a useful, simple and practical diagnostic tool.

INTRODUCTION

Human hair, although it is an ectodermal structure devoid of vital functions, may present a number of anomalies that may reflect a local condition, or, on the contrary, a general medical condition, sometimes serious, with strong impact on the physical or mental health of children. Therefore it requires careful consideration in terms of density, texture, colour, insertion in order to identify existent anomalies.

Phenotypic abnormalities of eyelashes and eyebrows are, with some exceptions, regarded as minor, they are often not registered in the records of patients observation.

Numerous studies attest to the fact that 15-20% of healthy newborns have a minor abnormality. It has been estimated that infants with a minor anomaly have a 3% risk of having major associated anomalies. This association risk of major anomalies increases to 10% in the presence of two minor abnormalities, a situation encountered in 0.8% of healthy children. A percentage of 0.5% of healthy infants have three or more minor associated anomalies, for these infants the risk of having a major malformation increases to 20%. (Adam Margaret et. al., 2003)

According to the study published in 1999 by Garcia-Hernandez MJ and collaborators, the persons with various hair disorders, especially

alopecia, have higher levels of anxiety and depression, which exceed the ability to keep them under control. They also have a sense of loss of self-esteem, their quality of life is affected and have a depreciated image of their own body. Patients who lose their eyelashes and eyebrows may have identity issues, because such features define a person's face. (Garcia-Hernandez MJ et. al., 1999)

MATERIAL AND METHOD

To study the phenotypic abnormalities mentioned we conducted a retrospective study on a group of patients who were registered with the Department of Medical Genetics in Oradea. There were included in the study patients with complete and accessible medical documentation, as well as the related photo documentation.

14 cases were diagnosed with Cornelia de Lange syndrome, 4 cases with congenital hypertrichosis, 18 cases with anhidrotic ectodermal dysplasia, 34 cases of albinism (total albinism or partial albinism 2 patients), 1 case of congenital alopecia, 12 cases with Aarskog syndrome, 1 case with Waardenburg syndrome, 6 cases of Noonan syndrome, 15 cases with mucopolysaccharidosis type I, II and IV, 10 cases of Treacher-Collins syndrome and two cases with Dubowitz syndrome. (Figure 1)

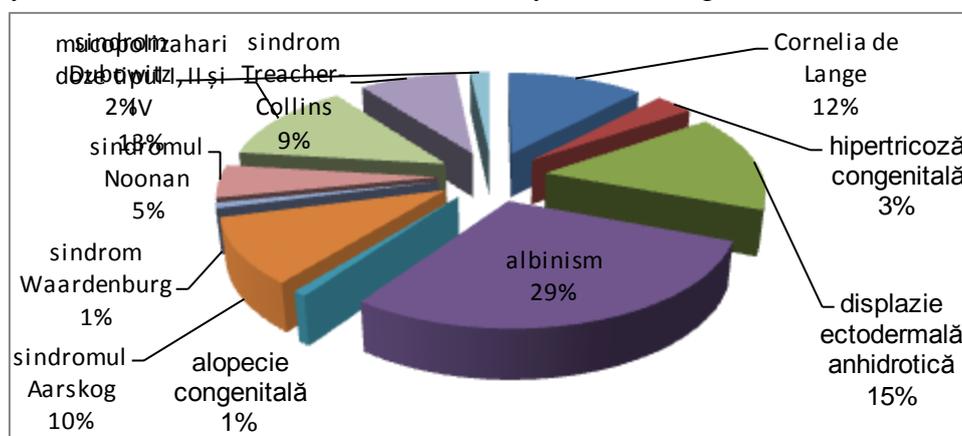


Fig. 1. Syndromes included in the study

Based on these criteria we included in the study group a total of 118 patients with genetic syndromes mentioned, out of which 72 male and 46 female.

A total number of 49 patients were from urban areas and 69 in rural areas.

The age of patients included in the study ranged between 0 and 34, inclusive, as follows: newborns: 10 cases, infants: 4 cases, 1 – 3 years: 9 cases, 4 – 6 years: 9 cases, 7 – 10 years: 16 cases, 11 – 14 years: 11 cases, 15 – 18 years: 8 cases, over 18 years: 51 cases. (Figure 2)

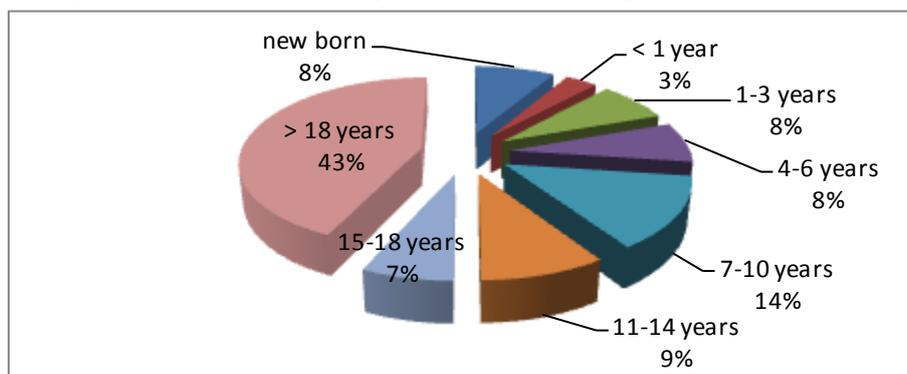


Fig. 2. Distribution of patients by age groups.

Phenotypic abnormalities classification was done according to the international classification published in London Dysmorphology Data Base, in which they are classified into anatomical variants, minor phenotypic abnormalities and proper malformations. (Winter RM, Baraitser M., 2001)

RESULTS AND DISCUSSIONS

In the case of patients included in the study were noted all types of anomalies: quantitative, of colour, of structure and of insertion. These were highlighted in a number of 87 patients in all the cases studied, these representing 74% of all patients. (Table 1, Table 2)

Table 1

Phenotypic abnormalities of eyelashes

Eyelashes anomalies	No. of cases
Absent upper/lower eyelashes	4
Scarce eyelashes	17
Long eyelashes	17
Bicolour eyelashes	1
Hypopigmented eyelashes	32

Table 2

Phenotypic abnormalities of eyebrows	
Eyebrows anomalies	No. of cases
Hypopigmented eyebrows	32
Absent eyebrows	2
Hypoplastic eyebrows	18
Scarce eyebrows	17
Rarefied eyebrows in medial/lateral side	1
Arched eyebrows	3
Synophrys	21
Thick/bushy eyebrows	25

Among the anomalies studied the pigmentation ones prevail, followed by quantitative and insertion anomalies.

In Cornelia de Lange syndrome the eyelashes and eyebrows abnormalities were noted in the following percentages: long eyelashes for 92% of patients, Synophrys for 78% of patients, thick eyebrows for 71% of patients. Reports in the literature mention the quantitative changes at the level of eyebrows and Synophrys as being present in 99% and 98% of the patients. (Jones KL, 2006 Hey MY et. al.2012, Ireland, M et. al. 1993, Allanson JE et. al. 1997, Hawley, PP et. al. 1985)

In the case of anhidrotic ectodermal dysplasia 94% of patients had anomalies of sparse and hypoplastic eyelashes and eyebrows type and in 6% of the cases we observed their absence, disorders mentioned in the specialty literature. (Kyle B. Jones et al, 2013)

Total or partial pigmentation abnormalities characterized the 34 cases of partial or total albinism. (Boissy, R. E et al 1996, Inagaki, K et al, 2004). Patients with Mucopolysaccharidosis showed quantitative and insertion anomalies, thick eyebrows and Synophrys types mentioned in 40% and respectively 6% of patients.

Dubowitz syndrome patients showed abnormalities similar to those described in several case studies: sparse eyelashes and eyebrows, rarefied eyebrows in medial/lateral side. (Moller et al 1985 KT, KR Swartz et al, 2003, Lee MK et al 2010, Huber et al 2011 RS)

As stated in numerous specialty literature articles, the absence of eyelashes and eyebrows characterizes from the point of view of anomalies studied, the patient with congenital alopecia. (Bennassar A et al, 2011)

Partial or total absence of eyelashes at the level of lower eyelid (encountered in 53% of cases) represent hair anomalies encountered in patients with mandibulofacial dysostosis (Treacher-Collins or Franceschetti-Zwahlen-Klein syndrome). (Robert J. Gorlin et al, 2001, Alison Mary, 2007)

In the present study the anomaly mentioned was found in 30% of patients with abnormalities of arched eyebrows, Synophrys, hypoplastic eyebrows types encountered in 10% of cases.

In the case of patients with Noonan syndrome, both in this study and the published data studied, there were no suggestive phenotypic abnormalities of eyelashes and eyebrows. (Romano et al, 2010)

According to literature data in Waardenburg syndrome the changes involving hair are the following: white strands of hair (partial albinism), premature greying (in 17-45% of cases), thicker eyebrows in the medial side (75% of patients), Synophrys. (Jones KL, 2006 Pardon, E et al, 2003)

In a study by da Silva E.O. (1991) on a sample of 73 patients, the frequency of hair anomalies was the following: thick eyebrows and Synophrys to 76% of patients, partial albinism in 29% of subjects and 44% presented premature greying.

In the present study it was included one patient with Waardenburg syndrome, with anomalies of bicolour eyelashes and thick eyebrows type.

Hair abnormalities frequently mentioned in various studies in patients with Aarskog syndrome are predominantly those referring to the front insertion, V-shaped, there have not been reported suggestive changes of eyelashes and eyebrows. (Porteous ME et al, 1991 KL Jones, 2006). In the study group these abnormalities were represented by thick eyebrows (in 8% of cases), arched eyebrows (in 8% of cases), Synophrys and long eyelashes to 16% of the patients.

CONCLUSIONS

Eyelash and eyebrow phenotypic abnormalities are minor anomalies that may be encountered frequently in some genetic syndromes.

Specific associations were observed between certain genetic disorders and various abnormalities of eyelashes or eyebrows, but in depth studies are needed on a significant number of patients.

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