STUDY ABOUT INCIDENCE OF CONGENITAL BONY ABNORMALITIES IN A POPULATION WITH MENTAL DEFICIENCY

IOANA MIHAELA TOMULESCU
University of Oradea, Faculty of Sciences, Department of Biology

ABSTRACT (online version)
This study is about the incidence of congenital bony abnormalities in a population with mental deficiency. It is known that genetic disorders cause mental disorders and malformative disorders, including bony abnormalities. The most often observed congenital abnormalities are: congenital hip sprain, flat foot, club foot, equin foot and congenital vertebral column disorders. We studied 596 children interned in Neurology and Psychiatry Clinical Hospital of Oradea between 1999 and 2001 period. In 596 children, 393 presented different types of mental deficiency. We observed that most common bony disorders in this population are congenital hip sprain, vertebral column abnormalities and club foot.

KEY WORDS: mental deficiency, congenital bony disorders.

INTRODUCTION
The most frequent congenital bony disorders observed in studied population are: congenital hip sprain, club foot, flat foot and vertebral column abnormalities. Speciality reference materials mention that congenital bony disorders possibly occur sporadically, isolated or associated in plurimalformative syndromes.

Speciality reference materials estimate a 1.2% frequency of congenital bony disorders in newborn children (Czeizel, 1988). Most common congenital bony disorders are: syndactylia, polydactylia, hemimelia, equin foot, flat foot, club foot, congenital hip sprain etc. These can occur isolated, too, but they can be an important component parts of clinical board of some chromosomal disorders, such as Down syndrome, Turner syndrome, Patau syndrome (Bembea, 2001). Some of these abnormalities can be surgical and/or orthopaedical solved. Many authors relate a 5/1000 frequency for clubfoot in newborn children. It seems that boys are affected twice often than girls. Congenital hip sprain has a 1-7/1000 frequency in newborn children, and hemimelia has a 1/5000 frequency in newborn children (Jones, 1997; Reed et al., 1989). It is important to mention the risk in phratria for club foot. It is about 1/20 for a female princeps case and 1/50
for a male princeps case. When the princeps case is a parent (is not important the sex of the parent), the recurrence risk is about 1/33. Sex ratio in congenital hip sprain is 6F:1M and the recurrence risk in phratria is about 1/20 for sisters and about 3% for brothers. In the case of descendents of an affected parent, the recurrence risk is about 1/8 (Bridge, 1997).

**MATERIAL AND METHODS**

We investigated 596 children interned in Neurology and Psychiatry Clinical Hospital of Oradea between 1999 and 2001 period. In 596 children, 393 associated different types of mental deficiency. The methods utilised were cytogenetic, clinical investigations, somatometrical, statistical, psychiatric investigation and hearing tests, too. There were realised family investigations and were constructed pedigrees.

**RESULTS AND DISCUSSIONS**

**General aspects.** We reported 81 cases of congenital bony disorders. These cases represent a frequency of 13,59% in studied population. In this total, 55 cases (67,9%) are associated with mental deficiency, which represents 14% in population with mental deficiency. In general, the frequency in rural area has a meaningful increase than in urban. In general, girls associated mental deficiency more often than boys did. In studied population, 13 cases (23,64%) associated mild mental deficiency, 10 cases (18,18%) associated moderate type of mental retardation and 32 cases (58,18%) associated severe mental deficiency. Most of cases proceed from rural area. The most often is observed the congenital vertebral column disorders (figure 1, 2 and 3). The frequency is meaningfully increased in children group with severe mental deficiency. This result can be explained because of the existence of congenital bony abnormalities, such as common traits in chromosomal syndromes or other syndroms which cause plurimalformative phenotypes associated with mental deficiency. Speciality reference materials related frequent association between nervous and bony disorders (Johnson, 1996). Also, there were observed in some families a concentration of abnormalities, frequent autosomal dominant, which possibly have a variable expression, such those related by Bembea et al. (2002) and Puiu & Vasile (2002). Sometimes, these abnormalities associate bony disorders of face (Belengeanu et al., 2002). Rodini et al. (2002) related the frequent association of bony disorders with nervous disorders and labial/

**FIG. 1.** Plotting the frequency of congenital bony cases associated with mental deficiency in studied population.

**FIG. 2.** Plotting the frequency of different types of congenital bony disorders associated with mental deficiency in studied population.
FIG. 3. Plotting the frequency of congenital bony disorders cases associated with different types of mental deficiency.

FIG. 4. Plotting the frequency of flat foot cases associated with mental deficiency in studied population.
**Flat foot.** In 81 case of congenital bony disorders, 28 (34.57%) are represented by flat foot (figure 4). In 28 cases, 16 cases associated different types of mental deficiency: 5 with mild, 5 with moderate and 6 with severe mental deficiency. The frequency of flat foot is increased in group with severe mental deficiency. The explanation of this result may be the plurimalformative syndromes who include mental deficiency and bony abnormalities. An important factor for bony abnormalities it is congenital hyposecretion of thyroid (Bucerzan et al., 2002; Schimke, 1995). There are bony disorders determined by autosomal genes with dominant inheritance, agglomerated in some families, genes who possibly to determine a certain mental deficiency. In general, when mental deficiency is associated with bony disorders, the severity of mental deficiency increase proportionally with number of congenital bony disorders.

In moderate and severe mental deficiency groups, we observed an increase frequency of bony disorders in boys group. The explanation could be the presence in some cases of X-fragil syndrome.

**Club foot.** In general, the incidence of this abnormality is 5/1000 living newborn children. In 1 of 1000 newborn children, the abnormality is like a very severe type. Sex ratio is 2M:1F and the risk in phratria is 1:50 for male case princeps and 1:20 for a female case princeps. In 81 cases of congenital bony disorders there are 3 cases of club foot. All of these 3 cases are boys and 2 of them proceed from rural area.

**Congenital hip sprain.** In 81 cases of congenital bony disorders, 14 cases are represented by congenital hip sprain. Ten cases of 14 are associated with different types of mental deficiency. Most of cases are girls and they proceed from rural area. Also, we observed an increased frequency in severe mental deficiency group (figure 5).

**Equin foot.** We observed 18 cases of this abnormality. Only one of 18 cases didn’t has associated mental deficiency. Two cases of 17, associated mild mental deficiency, 1 associated moderate mental deficiency and 14 associated severe mental deficiency. It seems that mutant gene determined the abnormality of bones (figure 6). Also, this disorder is common in children with alchoolic mothers during the pregnancy period and in plurimalformative syndromes (Ellhassone et al., 1996; Jones, 1997; Mitişor et al., 2002).
Vertebral column disorders and other congenital bony disorders. In 81 cases of congenital bony abnormalities, 35 cases (43.21%) are represented by congenital vertebral column abnormalities and other
congenital bony abnormalities (figure 7). These 35 cases represent a frequency of 5.87% in studied population. In 35 cases, 24 associated mental deficiency, which means a frequency of 68.57%. In population with mental deficiency, the frequency is of 6.11%.

**FIG. 7.** Plotting the frequency of congenital vertebral column disorders and other bony abnormalities cases associated with mental deficiency in studied population.

**CONCLUSIONS**
- We observed in 393 children with different types of mental deficiency a frequency of 25% cases with constitutional abnormalities which could be caused by the genetic disorders.
- Cranial and facial dysmorfism has a frequency of 13%. This frequency is increased in the group of severe mental deficiency. These abnormalities could be caused by some genetic disorders which cause bony abnormalities, too. The frequency is increased in severe mental deficiency group.
- Congenital bony disorders are often in the population with mental deficiency.
- The frequency of these abnormalities is meaningfully increased in the group of children with severe mental deficiency.
- In many cases the congenital bony disorders are associated with congenital nervous abnormalities.
• The explanation of these frequencies is the presence in the children group with associated mental deficiency of many genetic disorders which could cause congenital abnormalities.

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