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Original Research Article

Biochemical Profile of People with Down syndrome: About 25 Cases

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Abstract: Down syndrome or trisomy 21 is a genetic disease resulting from an additional gain of chromosome 21. It predisposes to multiple pathologies including obesity, infections, diabetes, or hypothyroidism, responsible for various biological abnormalities. This work aimed to determine biochemical profile of congolese persons with Down syndrome, living in Brazzaville (Congo). The study was carried out on 25 persons with Down syndrome. They are cared for in four specialized centers for disabled children. The methodology was based on the determination of glycaemia, proteins and lipids by spectrophotometric and electrophoretic methods. Globally, normal values of total cholesterol, triglycerides, and glucose (except for one person who had moderate hyperglycaemia), were observed. We noted an hyperlipidemia associated with low levels of HDL, LDL, albumin, and alpha 1 globulin. *In Conclusion:* there is a disturbance in biochemical profile in subjects with Down syndrome, which predispose to diabetes, cardiovascular diseases and opportunistic infections.
Keywords: Down Syndrome, Trisomy 21, Biochemical analysis, Glycaemia, Lipids, Proteins.

INTRODUCTION

Down syndrome also called trisomy 21 is the most common chromosomal disorder with the incidence estimated between 1/700 to 1/1000 live births, with a high risk in strong correlation with advanced maternal age over 35 years and sometimes in younger women (Pazarbasi *et al.*, 2013, Dechamps *et al.*, 1998, Sghir, 2016).

A previous reported study at the teaching hospital of Brazzaville showed that DS is at the head of congenital malformation with a rate prevalence of 19.3% (Poaty *et al.*, 2018).

The disease is compatible with long survival but, it poses handicap problems mainly related to the intellectual disability and diverse pathologies such as diabetes, hypothyroidism, respiratory and cardiovacular diseases, cancers or Alzheimer (Turleau *et al.*, 2010, Mouko *et al.,* 2004, Vekemens 2003).

The predisposition of individuals with DS, to these pathologies, can lead to a disturbance of blood parameters mainly, lipids and glucose, the data of which have not yet been published in the Congo.

The aim of this study is to determine biochemical profile of a series of congolese individuals with DS.

PATIENTS AND METHODS

It was a cross sectional and descriptive study carried out over six months within four specialized centers in Brazzaville (Congo), whose essential mission is the educational, psychological and sometimes medical care of children living mainly with a congenital disability. It is: Medico-Psychopedagogical (MEDIPSYP) Center of château d'eau, Case Dominique of Poto-poto, Case Saint Vincent of Moungali, Integrated Health Center of Jane Vialle (Ouenzé).

PATIENTS

The study involved 25 subjects with DS, classified into 4 groups (Table 1). Informed consent from the parents and the institutions has been obtained beforehand.

METHODS

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The study was based on epidemiological research and biochemical analysis, and it obtained the IRSSA (Institute of Research on Health Sciences) reading committee's approval.

Epidemiological Research

Data collection was based on interview guidelines and medical records. The study variables were as follows: age, gender, weight and height. The body mass index (BMI), which is the ratio of weight (kg) to height squared (m²) was calculated.

Biochemical Analysis

The collection of venous blood (5ml / dry tube) allowed biochemical assays (glycaemia, lipids and proteins).

Biological analyzes and data processing were carried out in the laboratories of the teaching hospital of Brazzaville, the Faculty of Health Sciences of Brazzaville, and the Institute of Research in Health Sciences.

The samples (including control) were analyzed by two spectrophotometric methods and one electrophoretic technic: i) Trinder method (Trinder, 1969), it is a colorimetric and enzymatic technic which allowed determination glucose level in blood serum, total lipids, total cholesterol, triglycerides, high density lipid (HDL), and to calculate low density lipid (LDL). ii) Biuret (Lubran, 1978) for the determination of total proteins level. iii) Electrophoresis for the determination of other proteins on cellulose acetate.

RESULTS AND DISCUSSION

The distribution according to the centers shows that 80% (20/25) of the subjects come from the MEDIPSYP center and the Dominique case which are the two largest centers and specialized in disabled children in Brazzaville.

Epidemiologic and Anthropometric Data
Age and Gender

The predominant age group in the series was 6-10 years old, i.e. 32% (8/25) of individuals with trisomy 21 (Table 1). The mean age of DS was 12.18 years with extremes ranging from 01 to 32 years.

The mean age found in our series is in accordance with that published by Belmokthar, 2014, in Algeria, which is 11.2 years. The oldest group with a 32-year-old subject, represented 24% of individuals. These data show that DS is compatible with long survival. Indeed, the life expectancy reported in the literature is 50 to 60 years or more in some cases (Penrose, 1933, Piedra *et al.*, 2017).

The male sex was prevalent, 60% (15/25) with a sex ratio of 1.5. It should be noted that the male gender is often reported as being the most frequent in DS (Poaty *et al.*, 2018, Alao *et al.*, 2017).

Anthropometric Data

The mean weight was 46.4 kg with extremes of 9 and 93.3 kg. The mean body mass index was 23.1 kg/m2 with extremes of 15.2 kg/m² and 44.4 kg/m². We noted excess weight in groups 3 and 4. Overweight has been observed in 16% (4/25) of cases and obesity in 12% (3/25) of cases.

The results of the body mass index showed that DS is a pathology linked to a weight imbalance which concerns more overweight than obesity, main risk factor for diabetes. The excess weight is frequently observed in people with DS (Wernio *et al.*, 2022, Piedra *et al.*, 2017, Havercamp *et al.*, 2017, Foerste *et al.*, 2016, Belmokhtar, 2014). A mean short height height of 1.30 m was noted in the oldest group 4. Usual fact, because it exists a statural delay in DS subjects. Indeed, there is 20 cm reduction of final height in comparison with normal statural growth (Myrelid, 2009).

Biochemical Parameters

The means of all the biochemical parameters are reported in table 2. The lipids and proteins variations according to the age groups are respectively illustrated in figures 1 and 2.

Glycaemia

Concerning the blood glucose level, 99% of the subjects had a normal glycaemia rate except for a 16-year-old teenager (group 4) who presented a moderate hyperglycaemia of1.20g/l.

We note that 1 to 2% of people with DS have hyperglycaemia in adulthood, which lead to diabetes (Amani, *et al.*, 2014). Therefore, medical management of DS people must also be accompanied by a nutritional monitoring.

Lipids

Globally, triglycerides and total cholesterol were normal. Only one individual, belonging to group 4 (up 15 years) presented hypercholesterolemia. We observed in all groups, the low mean values of total cholesterol, HDL and LDL cholesterol fractions, already reported in other studies (Piedra *et al.*, 2017, Adelkan *et al.,* en 2012, Krzesińska *et al.*, 2022). However, the total lipids (Figure 2) were high in all groups with a mean value of 23. 38 g/l with extremes of 6.8g/l and 46.9g/l (Table 2). The highest values were noted in the group 3 (11-15 years) and 4.

Note that, hyperlipidemia increasing with age, is also reported by several authors (Piedra *et al.*, 2017, Adelkan *et al.,* en 2012, Pueschel *et al.*, 1992, Zamorano *et al.*, 1991). It predisposes highly to obesity, to cardiovascular diseases, and the risk would be four times higher than in the normal population.

Proteins

Protein analysis revealed a hyperproteinemia in six persons (i.e. 24 % of the series) of the group 4. The mean value of total proteins was 76.6 g/l with extremes of 66.94g/l and 90.7g/l (Table 2). We also observed decreased values of albumin (detected in 40% of DS) in young individuals and also alpha 1 mainly in older subjects (Figure 2). Those low levels would potentiate the occurrence of recurrent infections, particularly respiratory and otorhinolaryngeal infections.

All these data showed that individuals with DS have currentlty disturbance in the blood parameters (Wernio *et al.*, 2022). At the base, some authors incriminate: genetic factors due to the additional chromosome 21, low level of physical activity, abnormal dietary habits and low metabolism that lead to high calories intake (Wernio *et al.*, 2022, Foerste *et al.*, 2016).

Table 1: Distribution of the subjects according to age groups

Groups	Age range (years)	Number	Percentage (%)
1	0-5	4	16
2	6-10	8	32
3	11-15	7	28
4	>15	6	24
Total		**25**	**100**

Table 2: Glyceamia, lipids and proteins values

Parameters (g/l)*	Minimum	Mean	Maximum
Glycaemia (0.6-1.10l)	0.54	0.88 ± 0.13	1.20
Total cholesterol (1.40-2)	0.64	1.05 ± 0.25	1.69
Triglycerides (0.40-1.65)	0.47	0.89 ± 0.21	1.65
HDL (>0.65)	0.13	0.41 ± 0.25	1.20
LDL (0.40-1.30)	0.01	0.45 ± 0.28	1.28
Total lipids (2-8)	6.83	23.38 ± 10.13	46.9
Total proteins (62-80)	66.94	76.68	90.7
Albumin (40-50)	31.13	42.5	60.8
Alpha1 (1.20-3.50)	0.67	2.95	16.2
Alpha 2 (4.30-8,50)	4.53	7.85	14.2
Beta globulin (6-10.50)	6,32	8.96	13.05
Gamma globulin (7-14.60)	7.76	15.79	26.8

Parameters* (with normal values), HDL: high density lipid, LDL: low density lipid, Alpha: alpha globulin
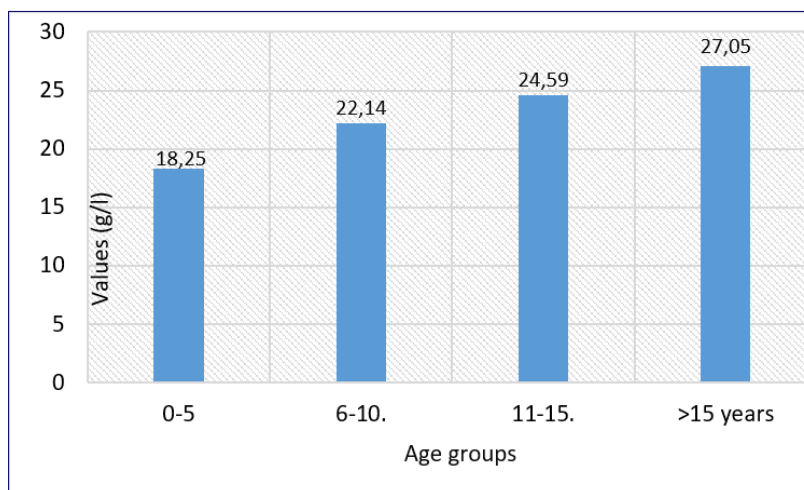

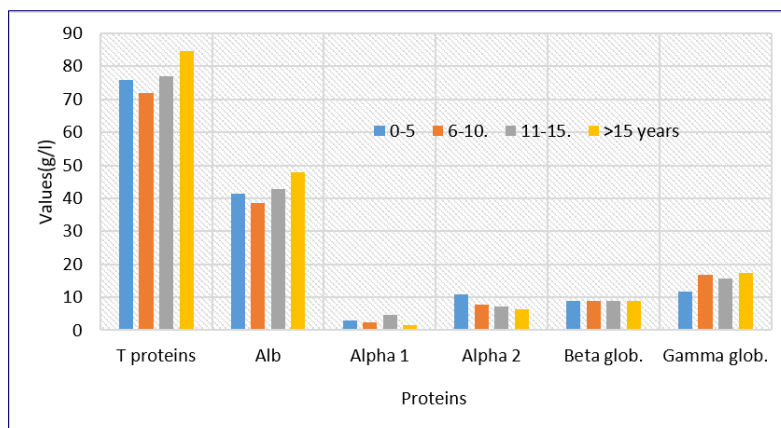**Figure 1: Mean Lipids values according the age groups**

Figure 2: Mean Protein values according the age groups

SUMMARY AND CONCLUSION

Trisomy 21, which almost constantly leads to intellectual disability, may be associated with biochemical disorders. In our series, 12% of the subjects were obese and the biochemical profile revealed hyperlipidemia increasing with age and moderate hyperglycaemia in one individual. On the other hand, decrease albuminemia and alpha-globulin 1 (predominant in older persons) have been observed. The biochemical disturbances predispose people with DS to diabetes, acquired cardiovascular diseases and opportunistic infections. Thus, a balanced diet with low-fat diet associated with physical activity, regular clinical and biological monitoring are needed.

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