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Speech assessment in Down's Syndrome Children

Thesis
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In
Phoniatrics

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List of Abbreviations

AMRs	Alternate motion rates
ASSI	Stuttering Severity Instrument for Children and Adults-Arabic version
CAS	Childhood apraxia of speech
CD	Childhood dysarthria
DDK	Diadochokinetic
DS	Down's syndrome
HCG	Human chorionic gonadotropin
ID	Intellectual disability
MSD-NOS	Motor speech disorder–not otherwise specified
OMD	Orofacial myofunctional disorders
PAPP-A	Pregnancy associated plasma protein -A
SMRs	Sequential motion rates
TD	Typically developed
VPI	Velopharyngeal insufficiency

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Introduction

Down syndrome (DS) is the most prevalent chromosomal disorder appearing in 1 in 800 live births ***(Scully, 2014).*** The incidence rate is higher in mothers of aging above 35-years-old and increases with further advances in maternal age ***(Brás et al., 2018)***. It is named after John Langdon Down, the British doctor who fully described the syndrome in 1866 ***(Hickey et al., 2012)***.

Down syndrome is a genetic syndrome most commonly caused by a third copy of chromosome 21 (trisomy 21), although a small number of cases are due to mosaicism and translocation of material from chromosome 21 ***(Udwin and Kuczynski, 2010).***

The extra chromosome content can arise through several different ways. The most common cause (about 92–95% of cases) is a complete extra copy of chromosome 21, resulting in trisomy 21 ***(Reisner, 2013).*** In 1.0 to 2.5% of cases, some of the cells in the body are normal and others have trisomy 21, known as mosaic Down syndrome ***(Mandal, 2013).***

The presence of speech problems has been a cause of concern for both investigators and parents of children with Down syndrome as it can lead to persistent deficits in speech intelligibility. Over the years, investigators have listed a number of articulatory and phonological problems found in these children ***(Kumin, 2004).***

Differences in structure and size of the tongue, weakness of orofacial musculature and general hypotonicity can influence motor movements associated with speech, and negatively impact the articulatory and phonatory abilities of children with Down syndrome ***(Stoel-Gammon, 2001)***.

Probably no single causal factor can account for the high level of unintelligible speech in children with Down's syndrome. **A variety of factors may contribute:**

- A protruding tongue caused by a small buccal cavity impedes articulation ***(Olbrisch, 1985)***. However, surgically reducing tongue size has no effect on speech intelligibility.
- Hearing problems, mainly in the mild to moderately impaired range, have a particularly high prevalence. Even mild fluctuating hearing loss can affect speech intelligibility, and children may learn to rely more on visual information and fail to develop adequate auditory attention ***(Evenhuis et al., 1992).***
- Hypotonia affects the speed and precision of speech movement.
- Chronic upper respiratory tract infection is associated with blockage of the nasal cavity by mucus, leading to mouth breathing and lack of nasal resonance ***(Rondal and Edwards, 1997).***

Speech intelligibility is determined by individual factors and also by the interaction of these specific factors as they affect intelligibility for an individual ***(Kumin, 2001).***

Speech characteristics in Down may be in the form of:

- Lack of articulatory precision and appropriate pausing and phrasing
- Vowel errors due to anatomical and/or motor limitations of the tongue ***(Bunton and Leddy, 2011).***
- Reduction of consonant clusters and final consonant deletion are the most frequent errors
- Errors are typically inconsistent
- Speech sound errors frequently result in reduced intelligibility in conversation ***(Kent and Vorperian, 2013).***

Furthermore, children with Down syndrome continue to use phonological processes (systematic sound error and simplification patterns) for longer periods than typically developing children would do ***(Roberts et al., 2005)***.

Dysfluency can add to the lack of intelligibility in speakers and adds another level of difficulty in the production of speech by children with Down syndrome. Berstein Ratner suggests that, in typically developing children, dysfluency is related to language processing ***(Bernstein, 2012).***

Another factor that affects speech intelligibility for children with Down syndrome is difficulty with voluntarily programming, combining, organising, and sequencing the movements necessary for speech, this difficulty, childhood verbal apraxia ***(Kumin, 2006).***

Aim of the Work

The aim of this thesis is to analyze the speech of children with Down's syndrome, in order to detect types and significance of speech disorders affecting speech intelligibility; this will help in putting the best strategy for therapy.

Down's Syndrome

Down's syndrome (DS), also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21***(Brás et al., 2018)***. It is typically associated with physical growth delays, characteristic facial features and mild to moderate intellectual disability ***(Malt et al., 2013).***

The possibility of its occurance increases from less than 0.1% in 20-year-old mothers to 3% in those age 45. There is no known behavioral activity or environmental factor that changes the possibility. Down syndrome can be identified during pregnancy by prenatal screening followed by diagnostic testing, or after birth by direct observation and genetic testing ***(Morris et al., 2002).***

The parents of the affected individual are typically genetically normal. The extra chromosome occurs by chance ***(Stephen and Gary, 2010).***

There is no cure for Down syndrome. Education and proper care have been shown to improve quality of life. Some children with Down syndrome are educated in typical school classes, while others require more specialized education ***(National Association for Down Syndrome, 2012).***

Epidemiology:

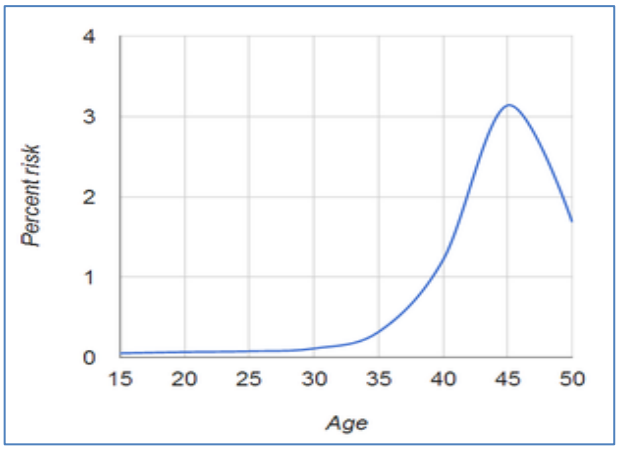

Fig. (1): The risk of having a Down syndrome pregnancy in relation to a mother's age ***(Dey and Ghosh, 2011).***

Maternal age affects the chances of having a pregnancy with Down syndrome. At age 20, the chance is one in 1441; at age 30, it is one in 959; at age 40, it is one in 84; and at age 50 it is one in 44. Although the probability increases with maternal age, 70% of children with Down syndrome are born to women 35 years of age and younger, because younger people have more children ***(Morris et al., 2002)***.

The father's older age is also a risk factor in women older than 35, but not in women younger than 35, and may partly explain the increase in risk as women age ***(Levitas and Reid, 2003).***

Genetics of Down's syndrome:

Down syndrome is caused by having three copies of the genes on chromosome 21, rather than the usual two ***(Stephen and Gary, 2010).*** The parents of the affected

individual are typically genetically normal. Those who have one child with Down syndrome have about a 1% risk of having a second child with the syndrome, if both parents are found to have normal karyotypes ***(Reisner, 2013)***.

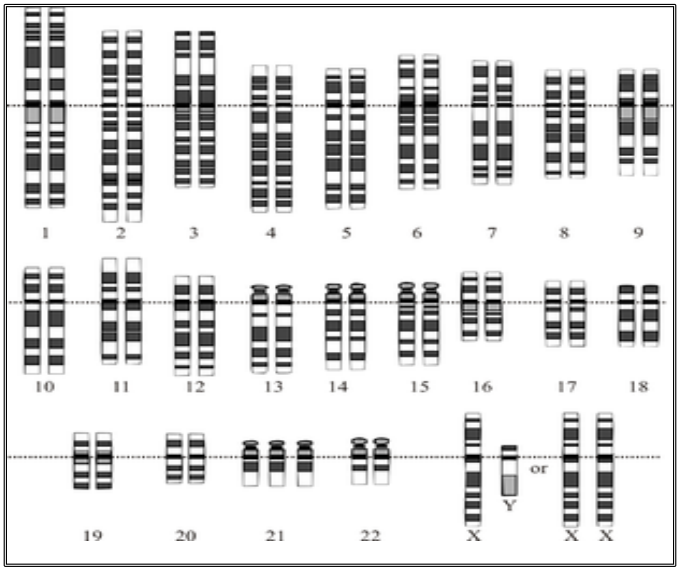

Fig. (2): Karyotype for trisomy Down syndrome.
from; ***National Human Genome Research Institute (2010)***

The extra chromosome content can arise through several different ways. The most common type (about 92–95% of cases) is a complete extra copy of chromosome 21, resulting in trisomy 21 ***(Mandal, 2013)***. In 1 to 2.5% of cases, some of the cells in the body are normal and others have trisomy 21, known as mosaic Down syndrome. The other causes (2– 4% of cases) can be Robertsonian translocation and isochromosomal or ring chromosome. Ischromosome is a term used to describe a condition in which two long arms of chromosome separate together rather than the long and short arm separating together during egg sperm development ***(Asim et al., 2015).***

The probability of this type of Down syndrome is not related to the mother's age (***Cummings, 2013).*** Some children without Down syndrome may inherit the translocation and have a higher probability of having children of their own with Down syndrome. In this case it is sometimes known as familial Down syndrome ***(Shaffer et al., 2012)***.

Mosaicism

Mosaic Down syndrome is when some of the cells in the body are normal and some cells have trisomy 21, an arrangement called a mosaic (46, XX/47, XX, +21) ***(Reisner, 2013).***

There is evidence that mosaic Down syndrome may produce less developmental delay, on average, than full trisomy 21 ***(De A. Moreira et al., 2000).***

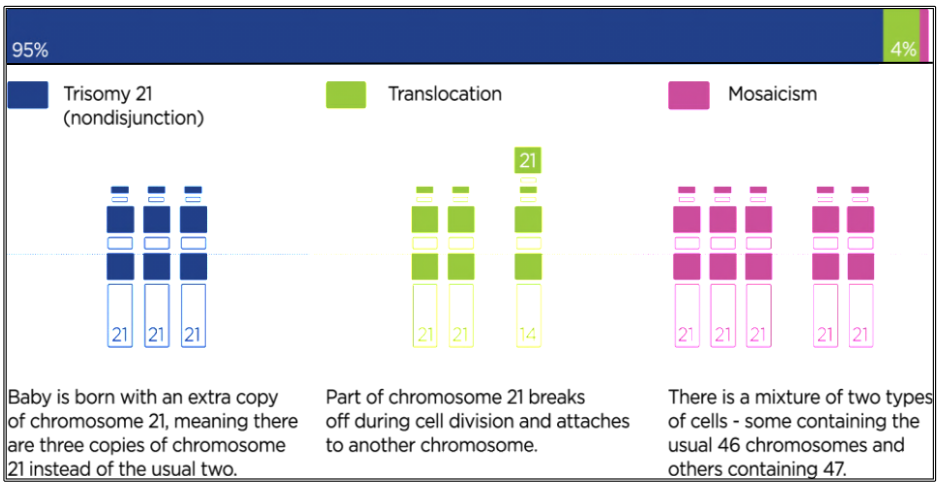

Fig. (3): Genetic Types of Down syndrome: after ***(National Down Syndrome Society, 2017)***