UNIVERSITY of RWANDA

# "HEALTH RELATED QUALITY OF LIFE ASSESSMENT IN CHILDREN WITH DOWN SYNDROME IN RWANDA"


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Master of Medicine in General Pediatrics


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A dissertation submitted in partial fulfilment of the requirements for the Degree of

MASTER OF MEDICINE IN GENERAL PAEDIATRICS

In the college of Medicine and Health Sciences

**Supervisors:**

Assoc. Prof. Annette UWINEZA

Dr. Aimable KANYAMUHUNGA

**September, 2022**

## DECLARATION

I declare that this dissertation is the result of my own work except where specifically acknowledged and has not been submitted for any other degree at the University of Rwanda or any other institution.

Dr. Janvière UWIZERA

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Dr. Janvière UWIZERA

September 1st, 2022

I hereby declare that this dissertation "**HEALTH RELATED QUALITY OF LIFE ASSESSMENT IN CHILDREN WITH DOWN SYNDROME IN RWANDA"** has been conducted under my supervision and is submitted by Dr. UWIZERA Janvière.

Assoc. Prof. Annette UWINEZA

September 1st, 2022

## DEDICATION

I dedicate this work

To the Almighty God

To my lovely husband Jean Luc MUTAGOMA

To our sons MUTAGOMA HIGA Delight and MUTAGOMA ESA Hansel

To my mother, my late father and my in- law parents

## ACKNOWLEDGEMENT

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I express my sincere gratitude to my husband MUTAGOMA Jean Luc for his daily support and encouragement and to our sons Delight and Hansel

I recognize advice and support from my family members and friends

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I appreciate the role of all pediatricians and my fellow residents for their contribution in my training

God bless you.

## ABSTRACT

**Background**

Down syndrome is a genetic disease associated with delay in development, mental retardation and many congenital malformations which impact the quality of life of affected individuals. It is the most prevalent genetic disease in Rwanda however the associated problems and QOL of children with DS has not yet been studied.

**Objectives:** The aim of this study is to examine the associated health problems and QOL of children with Down syndrome

**Methods:** We conducted a qualitative study at CHUK/ clinical genetic outpatient department. We involved 20 participants. Data collection was done through semi structured interviews and thematic analysis was used.

**Results:** All twenty participants were Rwandans and were from almost all provinces; only western province was not represented. Children mean age was 43 months. Caretakers mean age was 40.5 years. Four domains of quality of life were assessed including physical health in which delayed gross motor development was a common problem for all, emotions and socialization which were reported to be good for all, communication in which delayed speech was a common problem but no hearing impairment reported, daily living skills and school performance in which the parents reported inability to perform daily basic skills as performed by others of the same age and poor school performance.

**Conclusion:** Apart from emotional function and socialization that all our respondents reported to be good, other domains of life are impaired in children with DS. Having impaired QOL implies a need for support in the involved domains. In Rwanda we have limited rehabilitation services to help these children. Our study forms a base of literature to expand the research and propose necessary support and services to assist children with Down syndrome and their families.



**Key words:** *children, Down syndrome, health problems, quality of life, Rwanda*

## ABBREVIATIONS AND ACRONYMS

**WHO:** World Health Organization

**UR:** University of Rwanda

**CMHS:** College of Medicine and Health Sciences

**QoL:** Quality of life

**HRQoL:** Health related quality of life

**DS:** Down syndrome

**OSA**: Obstructive Sleep Apnea

**OPD:** Outpatient department

**CHUK:** University teaching Hospital of Kigali

**UK:** United Kingdom

**IQ:** Intellectual Quotient

**PI**: Principal Investigator

**RSSB**: Rwanda Social Security Board.

**IRB**: Institutional Review Board.

**CBHI**: Community Based Health Insurance.

**SVD**: Spontaneous Vaginal Deliver

**C/S**: Cesarean Section

AAP: American Academy of Pediatric

## TABLE OF CONTENT

# CHAPTER I. INTRODUCTION

## 1.1. Background

Down syndrome (DS) is the most common genetic disorder that cause developmental delay and intellectual disabilities in children(1). It is an inherited disorder due to trisomy of whole or segment of chromosome 21(2). About 95% of Down syndrome cases have an additional chromosome 21 that makes the chromosomes to be 47 instead of normal count of 46. The other 5% present other chromosomal abnormalities which are translocation (3%) and mosaicism (2%) or partial trisomy(3).

Down syndrome is associated with a lot of dysmorphic features and slowed psychomotor development associated with birth defects including heart defects, gastrointestinal anomalies, weak neuromuscular tone, hearing and visual problems, hematopoietic disorders and an increased occurrence of other medical disorders(2).

Congenital malformations associated with DS predispose children to need extra care physically, psychologically and financially, which affect their well-being and that of their entire families hence affecting their quality of life (4).Over the last 50 years, life expectancy of individuals with DS increased to 50 years and older, especially in industrialized nations. Because of this lengthening life-span, research on the QOL of people with DS is necessary to appropriately provide care and services (5).

Quality of life (QOL) refers to feeling fulfilled in all domains of life including physical, material, social and emotional wellbeing(6). HRQOL takes into account disabilities and limitations caused by disease including physical, mental and social aspects related to disease symptoms(7). Studies have shown that children with DS have significantly lower physical health, lower psychosocial health and lower school functioning but with conserved emotional functioning(8,9).

Quality of life assessment has been introduced in health care by WHO with the aim of prioritizing not only the disease but the overall patient's well-being (10).

Quality of life covers physical wellbeing, material wellbeing, social wellbeing, emotional wellbeing, development and activity(11); whereas health related quality of life (HRQOL) is defined as the personal judgement of the impact of disease and its management throughout the psychological, physical, social and somatic domains of functioning and well-being(12). In this study we will focus on HRQOL.

Since medical genetics has been introduced in Rwanda in 2006, children with DS have been diagnosed and regularly followed up in different services but no study yet done to assess their quality of life and that is the aim of this study.

## 1.2. Problem Statement

Down syndrome is associated with intellectual disability that is manifested by a lower IQ (intellectual quotient) scores and deficits in adaptive behavior, they also frequently have speech problems, hyperactivity and concentration problems, that affect their communication and social relationship; it is associated as well with various medical conditions some being congenital others developing later in life(8,13).

Owing to the successful early surgical and medical treatment of congenital heart disease and congenital anomalies of the gastrointestinal tract together with preventive health care programs; the general outcome and quality of life of children with Down syndrome has improved and their life expectancy has been increasing (14).

Their increasing life expectancy, goes with an increase in people living with chronic diseases and disability, one of the greatest challenges of public health being how to improve their quality of life (7).

In addition, in Rwanda, there are only two geneticists and no genetic counselor; children with DS are monitored by pediatricians or even general practitioners in District Hospitals, with varying knowledge and comfort in care for these children(1). Therefore in Rwanda like in many African countries there is a lack of data about quality of life of children with DS.

This brings to our attention the need to conduct this study which aims to assess the quality of life of children with DS , to better understand the needs of children with DS and to improve the overall care they receive.

## 1.3. Research Aims and Objectives

### 1.3.1 Research Aim

This study aims to examine the QOL of children with DS

### 1.3.2 Specific Objectives

1. To measure the socio-demographic characteristics of children living with DS
2. To assess the health-related quality of life of children with DS
3. To identify the factors that influence the QOL of children with DS

# CHAPTER II. LITERATURE REVIEW

## 2.1. Epidemiology of Down syndrome

Down syndrome can be diagnosed based on its craniofacial characteristic features mostly in developing countries where prenatal diagnostic tests are not yet available (15). In developed countries where prenatal screening is carried out and which allow pregnancy termination, about 50% of fetuses with Down syndrome are voluntarily aborted ,which results in a slight decrease birthrate of children with Down syndrome making the incidence of DS lower in high income countries than in the developing countries (16). However the real prevalence of DS may be higher in the developed countries due to the improvement in the survival of children with DS observed as a result of advanced management particularly of cardiovascular malformations; the prevalence differs from each country but World Health Organization has predicted a global incidence between 1 and 10/1000 live births worldwide (17).

The prevalence of DS in African populations is not accurately estimated because in most countries no studies have been done and where they have been done, most studies are inaccurate. In a study in southwestern Nigeria, the incidence was 1 in 865 live births compared to 1.49 per 1000 live births in Cape Town, South Africa, between 1974 and 1993(5,17).

In a survey of genetic diseases done in Rwanda, DS was the most common karyotype syndrome observed accounting 18,26 % of all assessed genetic diseases(19).

## 2.2. Associated health problems and health related quality of life

DS is associated with systemic malformations but some affected people can present with no major malformations as it was shown in registries review in Europe done to determine major anomalies occurring in fetuses and infants with DS ; 36% of studied children had no associated major malformations and the most frequent associated malformations were cardiac defects accounting 44%, followed by digestive system abnormalities in 6%, musculoskeletal system abnormalities 5%, urinary system abnormalities 4%, respiratory system abnormalities 2%, and other system abnormalities (3.6%); In the aforementioned study, AVSD (30%) followed by ASD (25%) and VSD (22%) were the most common associated congenital heart defects whereas duodenal atresia(67%), hirschprung disease(14%) and tracheoesophageal malformations(10%) were the most common gastrointestinal abnormalities , and among musculoskeletal anomalies there were syndactyly, club foot, polydactyly and cleft palate (20). Another study done previously in 1996 from registries of France, Italy and Sweden had shown similarities with the above mentioned study; where cardiac defects were the most common malformations accounting for 26% on average, the most common cardiac defect was endocardial cushion defects (39%) followed by ventricular septum defect (28%) and atrial septal defect (7%), Tetralogy of Fallot or patent ductus were the 3% and 4% respectively; cardiac defects were followed by gastrointestinal malformations including duodenal atresia, anorectal atresia, esophageal atresia, small gut atresia, megacolon and annular pancreas by order of the commonly encountered respectively. The above registry review also showed an increased risk for cleft palate, cleft lip, and limb deficiencies in children with DS(21). DS is also associated with impaired immunity and increased risk of hematological malignancy (16). Christopher J. Chin et al have done a literature review of the otolaryngologic manifestations of DS and found that around 50% of children with DS have a stenosed external ear auditory canal and a small pinna making their ear exam difficult leading to missed ear infections and later to hearing loss, they also found an increased risk of chronic rhinosinusitis together with their head and neck anatomy, they are predisposed to have OSA(22).

In addition to systemic malformations, DS has been found to be associated with other problems including delays in gross motor skill development, cognitive impairment, neurological disorders, delays in speech and language development that result in behavior, emotional and psychosocial problems (20,21).

Difficulty to produce speech is thought to be caused by low muscle tone which affects the muscles of the mouth and the tongue, hence delays in speech and expressive language are greater than delays in receptive language, making it difficult for them to communicate their message(25).

Children with DS have also delayed motor development, it has been showed that children with DS need twice the time needed for achieving developmental milestone when compared with normal children(26). An eminent example is that children with DS were able to walk independently a year later than normally developing children(26,27). In a study done in Poland to assess gross motor development it was found that the standing position was achieved by 10% of children <3 years old and 95% of children between 3–6 years. As well, the walking ability was assessed and was performed by 10% of children under 3 years old and by 95% of children between 3–6 years (29).

In a research done in Australia that assessed functional status of school aged children with DS, they found that walking skills were fully developed by school age, and had many challenges in self-care skills performance like in achieving full independence in bathing, dressing and grooming(30).

As a consequence of craniofacial and upper airway anomalies, increased risk of obesity, tonsil and adenoid enlargement, and generalized hypotonia, almost all individuals with DS encounter sleep disturbances because of OSA, which is believed to affect up to 80% of people with DS(29,30). Obstructive sleep apnea in children is associated with problems in concentration, difficulties in learning and has been reported to be associated with lower cognitive function, developmental delay and poor school performance (33).

A study done in UK to investigate the relationship between sleep and attention in children with DS and William Syndrome, Children with DS had the most night waking and disturbed sleep and they were found to have the greatest attention difficulties (34).

A third to a half of children with DS are overweight, the increased prevalence of overweight and obesity in DS are related to the presence of co-occurring disorders such as hypothyroidism which predisposes to increased body weight and is frequent in children with DS(11,13,35). In low and middle income countries overweight and obesity are increasing in normal population because of different factors including nutrition transition, cultural perception and genetics(36).

Children with DS are often labeled as being socially strong, they show strong social capabilities including the skills and behavior necessary in daily life situations compared to others with mental disability but also show weaknesses in other aspects of social capabilities as planning, making responsible decision and playing(37).

Children with DS need to be followed up for medical management, home environment, early intervention (including physiotherapy, speech therapy and occupational therapy), education and vocational training. According to American academy of pediatrics the follow up starts in prenatal visit with counselling of the family and continues until adulthood. AAP recommends a full physical examination at birth to identify if the child has features of DS, and to consider ophthalmologist evaluation, otolaryngologist evaluation and screening of thyroid disease within the first six months then continue evaluation every year as some diseases can be developed with age (38). A study done in Australia describing HRQOL of children and adolescent with DS reported that children and adolescents with Down syndrome were within the normal range for psychological well-being but autonomy , parent relation , Physical well-being and social support scores were significantly lower(4).

According to Kitiwan Rojnueangnit et al(1), who assessed QoL of children with DS in Thailand, mean QoL was lower in children with Down syndrome compared to children without DS; children with DS scored high in emotional functioning and had the lowest score in cognitive functioning. Their average quality of life score was lower than children without Down syndrome $67.9 \pm 14.5$ and $79 \pm 12.8$ points respectively.

Some children with DS are able to talk, read and do daily living activities, but others are not able to speak and need to be supported in their daily activities, this affects the family in many ways psychosocially by feeling isolated from the society and financially by being overwhelmed with responsibility of taking care of a child with disability (35,36).

Recently patient management has moved away from relying only on clinical and laboratory indicators of disease, towards measures that take into account the patient's point of view; WHO has developed a quality of life assessment concept to help health care providers measure the impact of disease and impairment on daily activities and behavior of affected people(10).

Health and illness are processes that could be understood as a continuity, as they are directly related to economic and socio-cultural aspects, as well as to life styles(6).

# CHAPTER III. MATERIALS AND METHODS

## 3.1 STUDY DESIGN

This study is a qualitative study that evaluated the QoL of children with Down syndrome and its influencing factors as reported by their caregivers.

## 3.2. STUDY POPULATION

### 3.2.1. Inclusion criteria

Children with confirmed Down syndrome by karyotype

Children aged 2 to 15 years

### 3.2.2. Exclusion criteria

Caregivers who don't live with the child on daily basis

## 3.3. STUDY SITE

This study was conducted at CHUK (University teaching hospital of Kigali). CHUK is a tertiary and teaching hospital located in Kigali city, Rwanda. It receives patients who need specialized care, referred from district hospitals. This hospital, as it has a medical geneticist, receives children with different genetic disorders, including Down syndrome, from all over the country. This study was done specifically in clinical genetic outpatient department.

## 3.4. SAMPLE SIZE CALCULATION

The study used purposive sampling and there was no fixed number of participants. Interviews continued until we reached saturation. Based on the literature, saturation was found to occur within the first twelve interviews, even though basic elements for metathemes were present earlier after six interviews (40). Data saturation for this study was reached after participation of 20 participants determined by parallel coding of data during the interview period to confirm that new themes were not emerging.

## 3.5. DATA COLLECTION PROCEDURE

We enrolled in this study twenty children between 2-15 years of age with confirmed DS by karyotype. Their information and address were found from medical records in CHUK patients electronic records from which we identified participants who meet inclusion criteria; then we called them to attend the clinic. The caretakers were explained the purpose of the study by the PI; the ones who accepted to participate in this study have signed a consent form.

The sociodemographic information of each participant was obtained from parents using a designed clinical demographic questionnaire: age of parents and children, sex of children and caregiver, residence, relationship with guardians, Ubudehe category, type of insurance, level of education of the caregiver and occupation of the caregiver.

QOL was assessed through interviews with caretakers using an interview guide that was designed by the PI and supervisors. Since we didn't have a tested tool for our population we used WHO QOL tool to define domains of life and we adapted it to pediatric population. The WHOQOL tool is a quality of life assessment developed by the WHOQOL group with fifteen international field centers, with the intention to develop a quality of life assessment that would be applicable cross-culturally.

It assesses individuals' perceptions of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns. It assesses different life domains including physical capacity, psychological, level of independence, social relationships, environment and spirituality using Likert scales with higher scores indicating higher quality of life (41). For this study, Likert scales was not used as our respondents were too young and intellectually disabled, they were not able to respond to the questions, we opted to ask their caretakers living with them day to day assuming that they know problems their children have.

We conducted semi-structured interviews we generated both qualitative and quantitative results.

Interviews were conducted using the designed interview guide in a private room face to face with the participant, they were digitally recorded using a smartphone and stored using a password-protected laptop only accessed by the principle investigator and supervisors.

## 3.6. STATISTICAL ANALYSIS AND DATA MANAGEMENT

Thematic analysis was used for this study. Information and opinions collected from interviewees were recorded into transcripts in Kinyarwanda and later translated in English. A codebook has been developed to facilitate interpretation of responses provided by participants. The coding and analysis of the data has been done using "Atlas.ti" software version 7.1.4.

## 3.7. ETHICAL CONSIDERATION

### 3.7.1. Funding and sponsors

No funding has been sought for this project.

### 3.7.2. Potential conflict of interest

There is no conflict of interest

### 3.7.3. Confidentiality

To ensure confidentiality and anonymity of the participants, code names were used during data processing and reporting, and data was kept in a password -protected file with only access to PI and supervisors.

### 3.7.4. Informed consent

Caretakers have been explained clearly the purpose and importance of this study, they were given time to ask any question and they have been informed that participation is voluntary. If the participant accepted to participate in the study, she/he was asked to sign a written consent.

### 3.7.5. Incentives

No incentives were provided.

### 3.7.6. Risk to subjects

There are no physical, legal, financial and/or social risks to the subjects during this study. Anticipated risk was emotional distress for the caretaker because of talking about their daily life. The principal investigator conducted interviews in a sensitive manner using empathy and there was no problem encountered.

### 3.7.7. Ethical approval

The research proposal was reviewed and approved by the Pediatric departmental academic staff, then CMHS Institutional Review Board *(No 347/CMHS IRB/2022)* and CHUK ethics committee (*Ref: EC/CHUK/095/2022)* before starting data collection and proceeding with the research.

# CHAPTER IV. RESULTS

A total of twenty parents/guardians were recruited in the study and they were interviewed about socio-demographic characteristics of the children and of caretakers themselves, children living conditions and other questions about the health-related quality of life of the children diagnosed with Down syndrome and confirmed by karyotype.

## 4.1. Socio-demographic characteristics of the participants and of their caregivers

Then median age of recruited children was 43 months, the majority of the children (80%) were male and 95% of them were Christians. Of 20 children who were recruited in the study, 12 (60%) were in the third economic category of ubudehe. Ubudehe category is a socio-economic classification of Rwandan households based on household income and living conditions. There were six Ubudehe categories before, 02.3
6 which have been changed in 2015 to four categories graded from one to four from the poorest to the richest respectively.
Category 1 is very poor and vulnerable people who are homeless and unable to feed themselves if not assisted, category 2 are the ones who are able to pay for some form of rented or low class owned accommodation but who are not gainfully employed and could only afford to eat once or twice a day. Category 3 are citizens who are gainfully employed or even employees of labour, this include small farmers or owners of small and medium scale enterprises. Lastly in category 4 are chief executive officers of big businesses, employees who had full time employment with organisations, industries or companies, government employees, owners of shops or markets and owners of commercial transport vehicles or trucks.( https://rwandapedia.rw/hgs/ubudehe/poverty-level-categories)

The majority (85%) of the participants was using community-based health insurance and the reported mean birth weight for the children was 3.1 kg (table 1).

Table 1: Sociodemographic characteristics of the study participants

| Characteristics | Number | Percentage |
|---|---|---|
| **Age in months** | | |
| 24-48 | 11 | 55.0 |
| 49-72 | 3 | 15.0 |
| >72 | 6 | 30.0 |
| **Weight in kg** | | |
| Mean ± SD | 16.52 ± 9.47 | |
| **Gender** | | |
| Male | 16 | 80.0 |
| Female | 4 | 20.0 |
| **Province of origin** | | |
| North | 8 | 40.0 |
| Kigali | 7 | 35.0 |
| Est | 3 | 15.0 |
| South | 1 | 5.0 |
| **Religion** | | |
| Christian | 19 | 95.0 |
| Muslim | 1 | 5.0 |
| **Ubudehe category** | | |
| Category 1 | 1 | 5.0 |
| Category 2 | 7 | 35.0 |
| Category 3 | 12 | 60.0 |
| **Insurance** | | |
| CBHI | 17 | 85.0 |
| RSSB | 3 | 15.0 |
| **Mode of birth** | | |
| SVD | 12 | 60.0 |
| C/S | 8 | 40.0 |
| Birth weight in kg | | |
| Mean ± SD | 3.13 ± 0.55 | |

Considering the caretakers (parents or guardians), the median age was 40.5 years (with SD=5.86) ranging from 27 to 47 years, 15 of 20 caretakers (75%) were female and 50% of them at least attended secondary school. The majority (17 of 20) of caretakers was living with their partners, 55% were unemployed and 15 of 20 caretakers were the mothers of the children (Table 2).

Table 2: Sociodemographic characteristics of the participants' caretakers

| Characteristics | Number | Percentage |
|---|---|---|
| **Age of caretaker** | | |
| **Mean ± SD** | **40.55 ± 6.19** | |
| **Gender of caretaker** | | |
| Male | 5 | 25.0 |
| Female | 15 | 75.0 |
| **Education level of the caretaker** | | |
| Illiterate | 3 | 15.0 |
| Primary | 7 | 35.0 |
| Secondary | 4 | 20.0 |
| University | 6 | 30.0 |
| **Marital status of the caretaker** | | |
| Married | 17 | 85.0 |
| Single | 2 | 10.0 |
| Divorced | 1 | 5.0 |
| **Occupation of the caretaker** | | |
| Employed | 5 | 25.0 |
| Self-employed | 4 | 20.0 |
| Unemployed | 11 | 55.0 |
| **Relationship to the child** | | |
| Mother | 15 | 75.0 |
| Father | 4 | 20.0 |
| Other | 1 | 5.0 |

From the thematic analysis four main categories were identified with 13 themes summarized in the table below:

Table3. Table of themes

| Categories | Themes |
|---|---|
| Physical health | • Reason of first consultation and time of diagnosis<br>• Perception on weight<br>• Description of child's motor development<br>• Ability to do exercises<br>• Presence of chronic diseases and available rehabilitation services<br>• Sleeping status |
| Socialization and emotions | • Living with others<br>• Child's mood |
| communication | • Hearing ability<br>• Speaking |
| Daily living skills and school functioning | • Basic skills performance<br>• School attendance<br>• School performance |

Table 4. Frequency tables

Table 4. 1. Physical health (N=20)

| **Variable** | **n(%)** |
|---|---|
| Poor weight gain | 17(85) |
| Good weight gain | 2(10) |
| Appropriate Motor development | 1(5) |
| Physical activity | 17(85) |
| No physical activity | 3(15) |
| Delayed Motor development | 19(95) |
| Chronic disease( congenital heart disease) | 7(35) |
| Impaired sleep( upper airway obstruction) | 5(25) |

Table 4.2. Socialisation and communication (N=20)

| **Variable** | **n(%)** |
|---|---|
| Good relationship with others | 20(100) |
| Happy mood | 20(100) |
| Unusual anger | 10(50) |
| Hearing | 20(100) |
| Delayed speech | 20(100) |

Table 4.3. Living skills performance and school functioning (N=6)

| **Variable** | **n(%)** |
|---|---|
| Feeding themselves | 5(83.3) |
| Toilet trained | 3(50) |
| Bathing themselves | 1(16.66) |
| cooking | 1(16.66) |
| School attendance | 4(66.66) |
| Good school performance | 1(16.66) |
| Poor school perormance | 5(83.3) |

Data collected from children above 3 years of age

## 4.2. Health-related quality of life of children with Down syndrome reported by their caregivers

### 4.2.1. Physical health

All the caregivers were aware of their children's disease as they have been explained by the geneticist. Most of them knew the diagnosis of their children after six months of age after noticing that their children are lagging behind others in achieving developmental milestones.

*"Yes I know the disease of my child, I have been explained by the geneticist that she has trisomy 21 and the test has confirmed it"* [Participant 9]

> *"At 7 months of age I noticed that he was not growing well compared to his elder brothers, he was unable to roll over, was unable to hold his neck but also had cough I consulted HC then transferred to DH where he was treated for pneumonia but also they told me that he has a problem and he was put on regular follow ups"* [Participant 10]

> *"He was not growing well, he was bedridden, not able to move, I consulted HC they told me "that is how your child is" I consulted traditional healers without improvement, I kept him home until 2 years of age when I consulted again HC and asked for transfer to DH, upon reaching DH the doctor transferred him to CHUK but I didn't come, instead I went to Gahini hospital where they care for disabled people, they taught me to help him with some physiotherapy at home, I came in CHUK recently in ENT for ear infection and that is where they transferred him to a geneticist who confirmed trisomy 21"* [Participant 11, mother of 8years old child]

> *"He was very hypotonic, we noticed that he is very floppy at 2months of age, we consulted HC they told us that it will come with time, at 6months there was no change, we consulted again, they told us still to wait but he kept being very weak with head lag and unable to sit, at 2years we consulted again and we were transferred to geneticist who confirmed trisomy 21"* [Participant 12]

Other participants reported that they were accidentally announced that their children have the condition when they consulted the health facilities for other reasons.

> *"....I consulted DH because of cough they treated him but also told me to come at CHUK where the sample was taken for the test which confirmed trisomy, but even before the results the geneticist told me that it is obvious that he has trisomy 21*" [Participant 1]

> *"We didn't notice any problem, but one day he got sick and we consulted, arriving at hospital they told us that our child has trisomy that he will not live more than 2 years, then we decided to come to CHUK for consultation"* [Participant 8]

> *"At 3 months and 2 weeks of age he had fever, diarrhea and cough, we consulted a private clinic, and the Doctor we found there transferred us to CHUK as the cost was high in private clinic. At CHUK we met a geneticist who did karyotype and confirmed T21, she sent us to cardiologist and physiotherapy"* [Participant 3]

> *"I consulted because she used to have fever, I consulted several times health center receiving amoxicillin and paracetamol but they did not find the cause of fever, one day I consulted a private clinic the doctor I found there told me , this child has trisomy 21, it was my first time to hear that word. He recommended going to the District hospital to get a transfer of CHUK, I went to the DH but the 1$^{st}$ Doctor I met told that my child doesn't have T21, I kept consulting health center for recurrent fevers, they gave me a transfer to see a pediatrician at DH, the pediatrician sent me to geneticist where T21 was confirmed, at that time she was 1 year old"* [Participant 2]

The majority of caregivers reported poor weight gain for their children which they associate with the facts that their children have poor appetite and frequently fall sick and refuse to eat.

> *"...he does not gain weight because most of the time he is sick, like every month he is at hospital"* [Participant 7]

*"He has a poor weight gain because he does not like to eat, he has poor appetite; I think that if he was eating well he could gain weight" [Participant 3]*

*"About gaining weight, aaah he has poor weight gain and instead of gaining weight, he loses weight progressively"* [Participant 10]

Few participants (three participants) reported that their children have a good weight gain. For example some participants mentioned:

*"This girl has a good weight gain, she has been having much weight even when I bring her to hospital it is difficult for me to carry her"* [Participant 4]

*"The weight gain has been good during his growth"* [Participant 14]

One participant reported that his child lost weight progressively with age

"*shortly after birth he gained much weight, he looked obese but since 5 months of age he started losing weight progressively, it was even one of the reason of consultation"* [Participant 16]

Almost all participants (19 of 20 participants) revealed that their children had delayed developmental milestone, as they reported that their children hold their neck, sit, crawl and walk later than others of their same age. Some of them underlined the role of physiotherapy in helping their children to attain some milestones.

*"He has been hypotonic since birth and bed ridden, has been able to hold his neck beyond 1 year of age when he started physiotherapy, he is now able to sit supported, but not yet able to crawl, stand or walk"* [Participant 1]

*"It was very difficult I didn't even know that he will be able to sit, he started to hold his neck at 2 years, sat at 3 years and he is not yet able to walk but can crawl"* [Participant 10, 5years old child]

*"He reached 8 months of age without getting out of bed, he started physiotherapy and, he started to sit at 18 months, started to walk at 2 years"* [Participant 14]

*"He had a developmental delay, he managed to hold his neck and to sit at 1year 5months, he crawled at 2 years and he is now able to walk supported"* [Participant 7,

2years and 6months]

Only one participant perceived that the milestone development of his child is normal as that of other children.

> *"He seems to have normal developmental milestones because when we compare with others of his age he only has a delay of like 1-2 months"* [Participant 8]

The majority reported that their children are able to do exercises/physical activities without pain or easy fatigability compared to other children of their age.

> *"..., about doing exercises and playing with others, she plays with others with no pains, and he is tired when others also are tired"* [Participant 2]
>
> *"...he is very happy to play with other children*" [Participant 12]
>
> *"My child is able to play with other children and she is not easily tired and she doesn't have pains"* [Participant 9]

Three participants mentioned that their children are not able to do the exercises as other children due to their inability to move caused by severe hypotonia.

> *"He is not able to play with others as he does not move but when he is watching others playing he looks interested"* [Participant 12, 2years and 8months]
>
> *"... he is not able to do the exercises as he is not able to walk but when he is given a toy he can play with it"* [Participant 3]
>
> *"No, he cannot move, he is very weak but his siblings can play with him where he is sitting"* [Participant 1]

About sleeping pattern, the majority reported that their children sleep well and some of them had sleeping difficulties as a consequence of upper airway obstruction.

> *"He used to have troubles in sleeping when he was between 1 year and 2 years, he could pass the whole night without sleeping because of nasal obstruction but now it has decreased he is able to sleep"* [Participant 3]
>
> *"This child had trouble sleeping and noisy breathing but since he has been operated he is well sleeping"* [Participant 4]
>
> *"Yes, she uses to have nasal obstruction mainly in cold times and has snoring that impairs her sleep"* [Participant 2]

*"Yes, he can spend the whole day without sleeping or even the night, and we really don't know when he will be able to sleep well like others"* [Participant 10]

Seven of 20 participants reported that their children have other chronic diseases and all of them reported congenital heart diseases.

*"My child has a heart disease; the doctor told me that he has 2 holes in the heart"* [Participant 1]

*"Yes, I was told that she has a small hole on the heart, cardiologist told me that it can close itself, on 1st appointment it was not yet closed, he gave me appointment in 2 years again to see if it will be closed "* [Participant 2]

*"They told me that he has heart disease"* [Participant 10]

**"***Yes, congenital heart disease, the doctor told me that he has 2 holes in the heart but the two holes have now closed"*

Concerning hospital admissions, most of respondents reported that their children have never been admitted; some participants reported that their children have been hospitalized and the reason of admission for most of them was fever. One respondent reported that his child has been hospitalized once when he was going to undergo operation for adenotonsillar hypertrophy.

*"Not yet hospitalized since birth, he consults only outpatient department"* [Participant 3]

*"He has never been hospitalized; every time we consult we go home on the same day"* [Participant 5]

*"She has been hospitalized 2times, for the 1st time we spent 2 weeks for the 2nd one we spent like 10 days, the reason was high grade fever for the 2 admissions"* [Participant 2]

*"He has been hospitalized several times like every 1-2 months. He used to have high grade fever, but the recent hospitalization was 1 year ago"* [Participant 7]

*"Has been hospitalized many times, and most of time the reason was fever***"** [Participant 18]

*"Has been hospitalized only when she was going to be operated"* [Participant 4]

### 4.2.2. Socialization and emotions

Almost all parents mentioned that children with DS have a good relationship with others; they reported that children are happy and friendly when playing with other children

> *"When he is with his siblings he is very happy, he loves his siblings, they also like to play with him*" [Participant 9]
>
> *"He likes other children and he is excited when he sees others playing"* [Participant 13]
>
> *"My child is always happy and he loves other children but we also try to make him happy because we know that he has a problem. We cannot allow him to be alone we always make him busy playing with his siblings"*
>
> [Participant 5]
>
> *"Before starting physiotherapy he looked very sad and lonely but currently when he is with his siblings he is happy to play with them"* [Participant 1]

Only one participant reported his child to be most of time distracted and that he likes to isolate from others

> *He likes to be alone and doesn't like to play with other children, sometimes when I ask him to do something he can do it after like ten minutes or even he does not do it.*
>
> [Participant 19]

About half of the participants reported unusual anger, compared to their peers, as challenge children with DS face in their interaction with others said:

> *"When she is with other children she is happy to play with them, but she does what she wants, she is very frustrated when you try to deviate her"* [Participant 2]
>
> *"She uses to be happy, but when there is something for which she is not happy she shows an unusual anger, if you punish her for example she can remember that after 30 min and cry again."* [Participant 4]
>
> *"….. and he uses to be very happy, but when you give him something he doesn't want he becomes very frustrated than other children"* [Participant 7]

All the respondents reported that their children cannot do the things other children of their age are able to do, they are always behind in development and in acquiring skills.

"*Others of his age are now in primary second or third year, they can help their parents in some household activities but him, as you can see even if he is in primary one he does not perform well and "*. [Participant 11]

*"He is not yet able to walk and talk at two years of age while others of the same age are doing so, this is a problem for the whole family which has to care for him"* [Participant 12]

### 4.2.3. Communication

In communication we assessed parent's perception on hearing and speaking ability of their children. All the participants agreed that their children hear well and they are able to say some words but cannot speak in sentences. They reported the use of gestures in case they want to say something they don't know.

*"I think that she hears well because when I call her she responds"* [Participant 12]

*"My child has no problem with hearing when he is about to do bad things and I can call him and stop him"* [Participant4]

*"He hears well as he can respond to other children when they are playing and he responds to his name"* [Participant7]

*"He is able to say 1 word-papa and he is able to make gestures to communicate eg: He can communicate that he wantsbto poop by crying until he is facilitated."* [Participant 3]

*"She is able to say 3 words –mama, papa, amazi (water), and she is able to communicate with others with gestures, like when she wants to eat she can show you a plate"* [Participant 2]

*"He able to say 2 words: papa, mama and he is able to make gestures to show what he wants"* [Participant 5]

*"He is able to speak but cannot make a sentence"* [Participant 18]

*"She knows to speak most of words and people have started to understand what she says before, only people who live at home could understand what she says"* [Participant 20, mother of 12 year old girl]

### 4.2.4. Daily living skills and school performance

Parents were asked about the abilities of children in doing basic living skills at home. Our participants were asked about self-care tasks including feeding, dressing, undressing and toileting.

The majority reported that their children can feed themselves with solid foods, and about a half of them were able to communicate when they want to urinate or pass stool and most of them had difficulties in dressing or undressing themselves except the parent of 12years old child who reported that she is able to perform more than self-care skills.

> *"She can feed herself solid food, she is able to ask for potty chair when she wants to poop, not yet able to dress or bathe herself"* [Participant 2]
>
> *"He is able to feed him solid food, everything I do in his presence, he tries to imitate"* [Participant 3]
>
> *"He is able to feed himself solid food and he can dress and undress him with only simple clothes"* [Participant 5]
>
> *"She is able to go to the toilet herself, she is able to feed herself solid food"* [Participant 4]
>
> *"She can feed herself but not yet toilet trained we are still using pampers because she can't tell when she wants to pass urine or stool"* [Participant 9]
>
> *He can feed himself, he is toilet can dress and undress but not appropriately, he needs assistance* [Participant 19]
>
> *"She can cook and make sure she is not burnt, she can wash clothes, she can wash dishes, and she likes to imitate others"* [Participant 20, mother of 12 year old girl]

Few respondents reported that their children are not yet able to do self-care tasks.

> *"The child is not yet able to do anything for himself at home. We feed him, cannot tell you when he wants to pass urine or stool. We do everything for him; he is like a small baby"* [Participant 1]

School functioning was assessed for children above 3 years of age;

The majority of pre-school and school children reported that their children are not yet in school because they cannot afford the specific schools of disabled children which they describe to be very expensive

> *"She is not yet in school but we plan to put her in school this coming year, we want to put her in their specific school but they are very expensive"* [Participant 4, mother of a 9 years old girl]

*"This child is not able to walk and so he cannot go to school and we hope that he will go to school designed for disabled even though they are expensive"[Participant 15]*

> *"He is not yet in school, because of financial issue, we lost his father 2years ago, the situation is not good and the schools of disabled people are very expensive"* [Participant 19]

Most of our respondents reported poor school performance that they relate to the fact that they study with normal children whom they don't share problems.

> *"Yes, he is in P1, but he doesn't follow in class, he goes outside whenever he wants"* [Participant 11]
>
> "*He does not attend school because most of time he is sick and he doesn't know anything*" [Participant 10]
>
> *"She is studying in nursery 2 with her little brother, she knows only the songs"* [Participant 20, mother of a 12 years old girl]

Only one participant reported good performance compared to his child's classmates because he studies in school of disabled people

> *"He is able to do all things given at school because he studies in a school of disabled people"* [Participant 14]

## CHAPTER V. DISCUSSION

In recent years quality of life studies have emerged as a tool to assess the impact of chronic diseases and their treatment on patients in order to improve their overall wellbeing(10). We conducted this study, which is as far as we know the first study done to assess QoL of children with DS and its influencing factors in Rwanda, with the purpose of addressing a paucity of literature in our country.

### Demographics

This study involved 20 participants. All of them were Rwandans; the majority of them were from Northern Province (40.0%), 35.0% were from Kigali, 15.0% from eastern province and 5.0% from South province. 15 of 20 (75.0%) caretakers were mothers, 4 (20.0%) was a father and 1(5.0%) was a maternal aunt. The age of caretakers were aged ranges from 27-52 years with a mean age of 40.5 years, this agrees with Mohammed Al-Biltagi(17) in his review article he found that the risk of having a child with DS increases with the age of mother, he has found that the risk is less than 1 in 1,400 for a woman below the age 25 years, which increases to 1 in 1,000 for a mother less than 30 years, further increases to 1 in 350 for women who become pregnant at age 35. Families appear stable despite the challenges in caring children with Down syndrome 95.0% participants were married. Caretakers had financial challenge as 55.0% of them were unemployed despite having variable levels of education, 30% of caretakers had a university level. All of them had medical insurance most of them were using CBHI (85.0%) and had access to diagnosis as all children were confirmed to have DS by chromosomal analysis. Most of the children were male (80.0%) and were between 2-9 years of age, there was only 1 child with 12 years of age.

### Physical health

According to the results of this study, the diagnosis of DS with karyotype is made a bit late when caretakers noticed delayed developmental milestones. In a study done in Ireland 89.4% of patients were diagnosed in the early period which was defined as 7 first days of life(42).

Almost all our respondents reported delayed motor development for which physiotherapy strongly supported. Previous studies found as well delayed motor development in children with DS(16,28).

Another study done in Poland has confirmed in which Gross Motor Function Measure-88 was used to assess motor development in children with DS. They found delayed motor development because in that study only 10% of children below 3years of age were able to stand and to walk (29) . It also agrees with Hayes and Mark who declared that motor impairment predominates over cognitive function in infancy to reverse by school age(16).

Most of our respondents reported poor weight gain which is against previous studies in which an increased risk of obesity was noted in children with DS(30,36). Adadot Hayes and Mark 1. Batshaw (16) while discussing DS and its associated malformations stated that obesity is not a direct consequence of DS it may be due to eating too much, underactivity or unidentified hypothyroidism.

Previous studies being conducted in high income countries, the difference in weight gain in our study can be explained by low social economic status ,difference in environment and culture, as well as genetic as it was shown to play a role in obesity even in normal population(36).

High incidence of obesity has gone in pair with high incidence of obstructive sleep apnea (43) which is the main cause of sleep disturbances in children with DS; increased incidence of OSA is also due to their physical and anatomical characteristics together with other comorbidities such as congenital heart diseases, pulmonary hypertension (27,38); This is in line with our study in which all participants who had sleep disturbance was due to upper airway obstruction. Yet, in our study only four (10.0%) participants reported sleep disturbances which is different from a high prevalence of sleep disturbances in children with DS that was found by both Maris et al and Carter et al probably because of low incidence of obesity in our participants. (45,46)

All the participants reported that they are able to do physical exercise as their peers without DS, this can be explained by younger age as one study revealed that physical activity decreases with age in children with DS(28).

All participants with chronic diseases reported congenital heart diseases, it is in agreement with the literature where the most common congenital anomaly associated with DS is CHD (18,19); however DS is associated with many others malformations which were not found in our study.

The fact that we didn't find other malformations in our study can be because the study was conducted in outpatient department and did not involve all age groups as some anomalies present at birth and others present later in life(1).

**Emotion and socialization**

Many studies have shown that children with DS have good emotional response , by measuring the HRQOL in different studies the highest score was found in emotional functioning(1,4,8), and the children with DS have been found to be very social (37) this is the same for our study ,95% of the participants reported that their children are always happy ,they socialize and like to play with other children. Some of them reported unusual frustration manifested by their children in some circumstances and this is thought to be caused by their inability to communicate appropriately their message and they are frustrated because they are not well understood (25). Our study did not explore other aspects of social capabilities like handling challenges and decision making.

**Communication**

Children with DS have delayed expressive language than receptive language therefore they use gestures as a mean of communication(47) this agrees with the result of our study where most of the participants were not able to speak appropriately and were using gestures. There was no hearing problem reported in our study, which is different from other studies done previously that showed an increased incidence of hearing loss in children with DS as a consequence of recurrent ear infections in children with DS (22,42,43). This difference can be explained by the absence of ear infection in most of our participants as only one participant reported one episode of ear infection.

Sally R. Shott et al conducted a 5 year longitudinal study following otolaryngology problems seen in children with DS under 2 years of age, they found that since they started the research only 2 out of 48 children didn't had an ear infection(50), this highlights how frequent are ear infections in children with DS. However, absence of ear infections and hearing loss from this study are the results of parents' observation so it cannot be concluded screening programs have to be put in place.

**Daily living skills and school functioning**

As part of their developmental delay, children with DS need assistance in performing daily living skills at home but as most of our respondents reported, they can perform some basic skills like feeding themselves, dressing themselves and being toilet trained. there is no age correlation in our study, but the mother of the 12 years old child reported more independence in performing daily living skills and can do other household activities, more larger studies are required to prove that relationship; however also Lin et al(51) reported that the capacity to perform daily skills increases with age in children with DS.

Access to education is a problem because schools for disabled people are not available everywhere and where they are available are expensive. Few of our participants who decided to teach them in public schools reported poor school performance and they think that their children could be helped by specialized school where they learn according to their intellectual capacity. some of them linked poor performance with school absences because of recurrent sickness another one reported being distracted and not following regulations in class; the later agrees with Lisa A., Deborah J. and Elizabeth W., who found that students with DS have difficulties in self-regulation and behavior management that affect their school functioning(52).

## STUDY LIMITATION

Being a subjective assessment, the QoL is more reliable when it is reported by the patient him/herself. In our study given the young age and mental retardation of our participants, we used parent reports assuming that they know everything from their children as they live with them and know challenges they face every day.

This study has involved a small number of participants and has been conducted in one hospital thus cannot be generalized for the whole population; however this study, being conducted in a referral hospital where geneticists are based, enabled us to involve participants from across the country.

## CHAPTER VI. CONCLUSION AND RECOMMENDATIONS

The results of this study showed that quality of life of children with Down syndrome is impaired because of its associated different health problems. Parents are aged and most were unemployed. Unemployment makes them face financial constraint to afford school and other supportive measures needed for their children. In our study, all participants had medical insurance and access to diagnosis. They face gross motor developmental delay however they had limited rehabilitation services. Only physiotherapy can be accessed at every district hospital. Congenital heart diseases were present in 38.48% of participants and were the only chronic disease reported by the participants in our study. Emotional function and socialization have been reported to be the strongest side of children with DS. This is a first study done in Rwanda to assess the QOL of children with DS; it involved a small number of children, larger studies involving all age groups are needed to further assess the needs and ways of improving the QoL of children with DS. We recommend:

- The government:
  - to avail special public schools for children with mental disability
  - Avail rehabilitation services
- MOH
  - To establish a guideline for health supervision of children with DS

- Researchers
  - To conduct a larger study comparing child and parent report QOL

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# APPENDICES

## Appendix 1. Questionnaire

### Part 1: social demographic characteristics

| Characteristics | Variables | Labels |
|---|---|---|
| **Socio demographic characteristics** | Age | |
| | Gender | Male<br>Female |
| | Place of origin | Kigali<br>East<br>West<br>South<br>North |
| | Religion | christian<br>muslim<br>other |
| | Social category/ubudehe | category 1<br>category 2<br>category 3<br>category 4 |
| | Health insurance | CBHI<br>RSSB<br>MMI<br>Private<br>Others<br>None |
| **Care taker's characteristics** | age | |
| | Gender | |
| | Educational level | Illiterate |

| | | |
|---|---|---|
| | | Primary school<br>Secondary school<br>Diploma<br>Bachelor's degree<br>Master's degree |
| | Marital status (parents only) | Single<br>Married<br>Living together<br>Separated<br>Divorced |
| | Occupation of care taker | Employed<br>Self-employed<br>Unemployed |
| | Relationship to child | Father<br>Mother<br>Sister<br>Brother<br>other |

## Part 2: INTERVIEW GUIDE

### I. Physical health

1. How was your child born? What is the birth weight?
2. What was the reason of your first consultation?
3. Are you aware of the disease of your child? Tell me when you first knew your child diagnosis.
4. Please tell me about your child's developmental milestones. When did he/she started to hold the head, to sit, to crawl and to walk

5. Please tell me about your child's weight gain (you can refer to the weight he/she had at different vaccination visits or OPD follow ups)
6. Is he/she able to do exercises? If no, why?(getting tired, having pain)
7. Is your child having trouble sleeping?
8. Does your child have any chronic disease? If yes, which one?)
9. Tell me about the hospital services
10. How many hospital admissions did he/she had up to now?

**II. Socialization and emotion**

1. Please tell me about how your child lives with others.
2. Is your child able to do things that others of his/her age can do?
3. Please tell me about your child's feelings( happy, sad, afraid, angry)

**III. Communication**

1. Is your child able to hear?
2. Is your child able to speak, if not how does he/she communicate with others?
3. When did he/she start to talk?

**IV. Daily living skills and school performance**

1. Please tell me about what can your child do at home(ability to eat, dress, bathe, performing household chores, understanding issues related to personal safety)
2. Does your child go to school? If no what is the reason of not going to school? If yes, is he/she able to do same things as peers at school?
3. Does your child miss school? If yes, what do you think is the reason?

**INTERVIEW GUIDE-KINYARWANDA VERSION**

**I. Ibirebana n'ubuzima bw'umwana**

1. Umwana yavutse neza? Yavukanye ibiro bingahe?
2. Ni iki cyatumye umujyana kwa muganga?
3. Waba uzi indwara umwana wawe afite? Wamenye ryari uburwayi bw'umwana wawe?
4. Umwana yiyongera ibiro? Mwakwifashishaa ibiro yagiye agira igihe cyo kumukingiza

5. Umwana yakuze gute? (gufata ijosi, kwicara, kugenda,kuvuga)
6. Umwana wanyu ashobora gukina n'abandi? Niba atabishobora utekereza ko ari iki kibimutera?( kunanirwa, kubabara)
7. Umwana wanyu hari ikibazo cyo gusinzira afite?
8. Hari indi ndwara idakira yaba afite? Niba ayifite ni iyihe?
9. Mwambwira ubufasha umwana wanyu ahabwa kwa muganga?
10. Kugeza ubu amaze kujya mu bitaro inshuro zingahe?

**II. Ibirebana n'ibyiyumviro n'imibanire n'abandi**

1. Mwambwira uko umwana wanyu abana n'abandi?
2. Mbese ashobora gukora nk'ibyo abandi bana bangana bazi gukora?
3. Mwambwira uko mubona ibyiyumviro by'umwana wanyu? ( akunda kuba yishimye, ababaye, afite ubwoba)
4. Umwana wanyu yaba agira ibibazo mu gusinzira kwe?

**III. Ibirebana no kuvugana n'abandi**

1. Umwana wanyu arumva?
2. Mbese ashobora kuvuga? Niba atabishobora avugana n'abandi ate
3. Yatangiye kuvuga ryari?

**IV. Ibirebana n'ibyo akora n'ubuzima bw'ishuri**

1. Mwambwira ibyo umwana wanyu ashobora gukora mu rugo? (kwirisha, kwiyambika,kwikarabya, gukora indi mirimo yo mu rugo, kumenya ibyamwangiza)
2. Umwana wanyu yatangiye ishuri? Niba atiga ni iyihe mpamvu ibitera?

   Niba yiga ashobora gukora nk'ibyo abandi bana bakora ku ishuri?

3. Ajya asiba ishuri? Niba asiba ni iki kibimutera?

## Appendix 2. Consent form

**Study Title: "Health related quality of life assessment in children with Down syndrome in Rwanda"**

We are Dr Janviere UWIZERA, Dr Annette UWINEZA and Dr Aimable KANYAMUHUNGA . We are doing a research on health related quality of life assessment in children with Down syndrome in Rwanda**.** This study aims to assess quality of life of children with Down syndrome

in order to improve on their health supervision. Participants are caretakers of children with trisomy 21, two to eighteen years of age.

Participants in this study will not receive any incentive, you are free to accept or to refuse participation and can withdraw from this study at any time without any consequence to the rest of care given to your child.

Data for this study will be obtained from interview with participants, which will be recorded using a smart phone. Participants' information will be kept confidential and on condition of anonymity, the information obtained from these assessments shall be used for educational and research purposes only.

In case a clarification is needed, you can contact:

Dr Janviere UWIZERA 0786101715; Dr Annette UWINEZA 0788741577; Dr Aimable KANYAMUHUNGA 0788670200.

I,……..., have been fully informed about the purposes of this study and my questions have been answered satisfactorily. I hereby, fully consent to participate in this study on **Health related quality of life assessment in children with Down syndrome in Rwanda.**

| | | |
|---|---|---|
| …….. . | …… | …/…/… |
| Name of the participant | Signature of the participant | Date |
| …….. … | …… | …/…/…. |
| Name of researcher | Signature of researcher | Date |

**Amasezerano yo kwemera kugira uruhare mu bushakashatsi**

INYITO Y'UBUSHAKASHATSI: "Health related quality of life assessment in children with Down syndrome in Rwanda"

Nitwa Dogiteri Janviere UWIZERA, mfatanije na Dr Annette UWINEZA, na Dr Aimable KANYAMUHUNGA turimo gukora ubushakashatsi bugamije kureba uko abana bafite trisomie 21 babayeho mu buzima bwabo bwa buri munsi ndetse n'ingaruka baba baterwa no kuba bafite trisomy 21. Ubu bushakashatsi bugamije kumenya imibereho y'aba bana kugirango turebe uko twarushaho kubafasha kubona ubuvuzi bukenewe mu rwego rwo kubafasha kubaho ubuzima bwiza.

Abazinjira muri ubu bushakashatsi ni abarwaza baherekeje abana bafite trisomy21 bari hagati y'imyaka ibiri (2) na cumi n'itanu (15).

Kwitabira kwawe ni ku bushake wemerewe kwemera cyangwa guhakana kujya muri ubu bushakashatsi. Abazabwitabira nta kiguzi bazahabwa, kandi niba uhisemo kutitabira ubu bushakashatsi, nta ngaruka mbi bizakugiraho ku mivurirwe y'umwana wawe. Amakuru akenewe muri ubu bushakashatsi tuzayabona tubaza ibibazo kandi dufata amajwi dukoresheje telephone. Abazabujyamo bazagirirwa ibanga ku makuru yose bazatanga kandi ibizava muri ubu bushakashatsi bizakoreshwa mu rwego rwo kwigisha gusa.

Ufite ikibazo cyangwa ibindi bisobanuro waduhamagara kuri numero zikurikira:
Dr Janviere UWIZERA 0786101715; Dr Annette UWINEZA 0788741577; Dr Aimable KANYAMUHUNGA 0788670200.

Njyewe ,…….nyuma yo gusobanurirwa neza ubushakashatsi bwitwa "**Health Related Quality Of Life Assessment In Children With Down Syndrome In Rwanda,"** nemeye kubujyamo.

| | | |
|---|---|---|
| …….. | …… | …….. |
| Amazina y'umurwaza witabira ubushakashatsi | Umukono | Italiki |
| …… | …… | …… |
| Amazina y'umushakasatsi | Umukono w'umushakashatsi | Italiki |

## Appendix 3. CMHS IRB approval

UNIVERSITY of RWANDA

COLLEGE OF MEDICINE AND HEALTH SCIENCES

DIRECTORATE OF RESEARCH & INNOVATION

*CMHS INSTITUTIONAL REVIEW BOARD (IRB)*

Kigali, 6th /June /2022

**Dr. UWIZERA Janvière**
**School of Medicine and Pharmacy, CMHS, UR**

**Approval Notice: No 347/CMHS IRB/2022**

Your Project Title ***"Health Related Quality Of Life Assessment in Children with Down Syndrome in Rwanda"*** has been evaluated by CMHS Institutional Review Board.

| | | Involved in the decision | | |
|---|---|---|---|---|
| | | | No (Reason) | |
| **Name of Members** | **Institute** | Yes | **Absent** | **Withdrawn from the proceeding** |
| Prof Kato J. Njunwa | UR-CMHS | X | | |
| Prof Stefan Jansen | UR-CMHS | X | | |
| Dr Brenda Asiimwe-Kateera | UR-CMHS | X | | |
| Prof Ntaganira Joseph | UR-CMHS | X | | |
| Dr Tumusiime K. David | UR-CMHS | X | | |
| Dr Kayonga N. Egide | UR-CMHS | X | | |
| Mr Kanyoni Maurice | UR-CMHS | | X | |
| Prof Munyanshongore Cyprien | UR-CMHS | X | | |
| Mrs Ruzindana Landrine | Kicukiro district | | X | |
| Prof Gishoma Darius | UR-CMHS | X | | |
| Prof Donatilla Mukamana | UR-CMHS | X | | |
| Prof Kyamanywa Patrick | UR-CMHS | | X | |
| Prof Condo Umutesi Jeannine | UR-CMHS | | X | |
| Dr Nyirazinyoye Laetitia | UR-CMHS | X | | |
| Dr Nkeramihigo Emmanuel | UR-CMHS | | X | |
| Sr Maliboli Marie Josee | CHUK | X | | |
| Dr Mudenge Charles | Centre Psycho-Social | X | | |

After reviewing your protocol during the IRB meeting of where quorum was met and revisions made on the advice of the CMHS IRB submitted on 6th June 2022, **Approval has been granted to your study.**

Please note that approval of the protocol and consent form is valid for **12 months.**

Email: researchcenter@ur.ac.rw P.O Box 3286 Kigali, Rwanda www.ur.ac.rw

You are responsible for fulfilling the following requirements:

1. Changes, amendments, and addenda to the protocol or consent form must be submitted to the committee for review and approval, prior to activation of the changes.
2. Only approved consent forms are to be used in the enrolment of participants.
3. All consent forms signed by subjects should be retained on file. The IRB may conduct audits of all study records, and consent documentation may be part of such audits.
4. A continuing review application must be submitted to the IRB in a timely fashion and before expiry of this approval
5. Failure to submit a continuing review application will result in termination of the study
6. Notify the IRB committee once the study is finished

Sincerely,

Date of Approval: The 6th June 2022

Expiration date: The 6th June 2023

**Prof Stefan JANSEN**
**Ag. Chairperson Institutional Review Board,**
**College of Medicine and Health Sciences, UR**

**Cc:**

- Principal College of Medicine and Health Sciences, UR
- University Director of Research and Postgraduate Studies, UR

## Appendix 4. CHUK IRB approval

CENTRE HOSPITALIER UNIVERSITAIRE
UNIVERSITY TEACHING HOSPITAL

Quality Health Care
Training & Research

Ethics Committee / Comité d'éthique

24th Jun,2022 Ref.:EC/CHUK/095/2022

***Review Approval Notice***

**Dear Janviere Uwizera,**

*Your research project:* ***"HEALTH RELATED QUALITY OF LIFE ASSESSMENT IN CHILDREN WITH DOWN SYNDROME IN RWANDA "***

During the meeting of the Ethics Committee of University Teaching Hospital of Kigali (CHUK) that was held on 24th Jun,2022 to evaluate your request for ethical approval of the above mentioned research project, we are pleased to inform you that the Ethics Committee/CHUK has approved your research project.

You are required to present the results of your study to CHUK Ethics Committee before publication by using this link:www.chuk.rw/research/fullreport/?appid=622&&chuk.

PS: Please note that the present approval is valid for 12 months.

Yours sincerely,

**Dr Emmanuel Rusingiza Kamanzi**
The Chairperson, Ethics Committee,
University Teaching Hospital of Kigali

ETHICS COMMITTEE
CHUK

Scan code to verify.

*" University teaching hospital of Kigali Ethics committee operates according to standard operating procedures (Sops) which are updated on an annual basis and in compliance with GCP and Ethics guidelines and regulations "*