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Impacts Of Genetic Diseases On The Affected Children's Parents

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ABSTRACT

Globally, 1 out of 150 live births has a detectable chromosomal abnormality. Genetic diseases account for about 7 percent and 20–30 percent of birth defects in the industrialized and developing countries respectively. The aim of this study was to determine the impacts of genetic diseases on the affected children's parents in Africa. A scoping review of eleven electronic databases was done. A multistep selection process was carried out. Only original peer-reviewed qualitative research studies done in Africa and published in peer-reviewed journals between 2018 and 2022 were included. Out of 4765 identified articles, only 10 studies fully met the inclusion criteria. The study concluded that parents of children with genetic diseases face many challenges such as; low quality of life-physically, socially, psychologically and environmentally; lack of information; lack of specialized services; financial strain; negative coping mechanisms; and existential concerns. Recommendations include; counselling and social support for affected parents to help mitigate the impact of genetic diseases; raising awareness of genetic diseases among the affected parents and their families, as well as the public in general; international and national policies and programmes should be instituted to enhance screening and detection of genetic disorders, coordination of clinical management of genetic diseases, facilitation of training of medical genetics/genomics professionals, and the stimulation of research in genetic diseases; ensure affordability of treatment and healthcare services to parents with children with genetic diseases; promote resilience and positive adaptive coping mechanisms; and provision of trauma counselling to the affected parents.

Keywords: Genetic diseases, children, parents, Africa.

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INTRODUCTION

A genetic disease is a disorder caused in part or entirely by a change in the usual DNA sequence. Genetic diseases can be caused by a mutation in one gene (monogenic disorder), by mutations in multiple genes, or by a combination of gene mutations and environmental factors (multifactorial or complex inheritance disorder), or by damage to chromosomes (changes in the number or structure of entire chromosomes, the structures that carry genes). Almost all diseases have a genetic component. Some diseases are caused by mutations that are inherited from the parents and are present in an individual at birth, like sickle cell disease. Other diseases are caused by acquired mutations in a gene or group of genes that occur during a person's life. Such mutations are not inherited from a parent, but occur either randomly or due to some environmental exposure (such as cigarette smoke). These include many cancers, as well as some forms of neurofibromatosis¹.

Common monogenic disorders include: Sickle cell disease, Cystic fibrosis, deafness that's present at birth (congenital), Duchenne muscular dystrophy, Familial hypercholesterolemia (a type of high cholesterol disease), Hemochromatosis (iron overload), neurofibromatosis type 1 (NF1) and Tay-Sachs disease. Common chromosomal disorders include: Down syndrome (Trisomy 21), FragileX syndrome, Klinefelter syndrome, Triple-X syndrome, Turner syndrome, Trisomy 18 and Trisomy 13. Complex or multifactorial disorders stem from a combination of gene mutations and other factors such as environmental mutagens such as chemical exposure, radiation, tobacco smoke and UV exposure from the sun, diet, certain medications and alcohol use. Common multifactorial disorders include: late-onset Alzheimer's disease, Arthritis, Autism spectrum disorder (in most cases), Cancer (in most cases), Coronary artery disease, Diabetes, Migraine headaches, Spina bifida and Isolated congenital heart defects².

Genetic disorders may also cause rare diseases. According to experts, there may be as many as 7,000 of these diseases. Rare genetic disorders include: AA amyloidosis, Adrenoleukodystrophy (ALD), Ehlers-Danlos syndrome, Mitochondrial diseases and Usher syndrome. To identify genetic diseases, genetic counseling and testing is done. The genetic test reveals the gene mutation(s). In cases of family history of genetic disorders, DNA testing for genetic disorders is done. The options include: carrier testing, prenatal screening, prenatal diagnostic testing and newborn screening. Unfortunately, most genetic disorders do not have a cure. However, some have treatments that may slow disease progression or lessen their impact on life².

Globally, about 1 out of 150 live newborns has a detectable chromosomal abnormality. Unfortunately, this high incidence represents only a small fraction of chromosome mutations

since the vast majority are lethal and result in prenatal death or stillbirth. Reports show that 50 percent of all first-trimester miscarriages and 20 percent of all second-trimester miscarriages are estimated to involve a chromosomally abnormal fetus³. Reports from pediatric accountable care organizations in the United States show that children with genetic diseases account for a disproportionately large percentage (nearly 10–20%) of pediatric hospitalizations⁴. In industrialized countries, genetic disorders account for about 7 percent of birth defects, while it is much higher in the developing countries, estimated at 20–30 percent⁵. Wojcik and colleagues lament that, although genetic disorders are a leading cause of infant mortality, they are not accurately captured by vital statistics⁶.

Sickle cell disease (SCD), which is a monogenic disorder, is an inherited blood disorder and the most common inherited disease worldwide, affecting over 400,000 babies annually with the greatest burden of disease within sub-Saharan Africa. Early diagnosis by newborn screening and simple interventions can significantly reduce this high mortality rate, but these capacities are limited mostly to babies born in developed countries, which represent less than 5% of the global burden of disease. For example, the majority of children in Angola and across Africa with sickle cell disease die within the first 5 years of life, often before a diagnosis is even made⁷. In Nigeria, SCD is the single most common severe genetic disorder occurring in up to 2%–3% of newborns. The sickle cell trait affects up to a quarter of the population. Thus, given Nigeria's population of over 180 million, the country probably has the largest population anywhere of people affected by SCD or at risk of having children with SCD⁸.

Table 1: Genetic diseases

Classes of genetic diseases	Proportion of those affected	Most affected
Chromosomal disorders	1 in 150 live newborn babies	Estimated to affect 50% of all trimester miscarriages and 20% of all second trimester miscarriages
Numerical abnormalities	Down syndrome is the most commonly observed of the autosomal trisomies, being found in about 1 out of 800 live births, both trisomy 13 and trisomy 18 are also seen in the population, albeit at greatly reduced rates (1 out of 10,000 live births and 1 out of 6,000 live births, respectively). They are lost to miscarriage	Pregnant women over 45 years of age have 1:20 of giving birth to chromosomally abnormal child and 1:50 proportion of child being born with trisomy 21(down syndrome). Risk is 1:400 for those over 35 years old while it is 1:1000 for women under 30 years old.
Structural abnormalities	5% of all cases of Down syndrome result from the presence of excess chromosome 21 material attached to the end of another chromosome as the result of a translocation event	Couples who have one pregnancy with a structural chromosomal abnormality are generally at significant increased risk above the general population to repeat the experience
Abnormalities of the sex chromosomes	1 in 400 male and 1 in 650 female live births demonstrate some form of sex chromosome abnormality	Turner syndrome is a condition of females who, in the classic form, carry only a single X chromosome (45, X). Although the condition is seen in about 1 in 2,500 to 1 in 5,000 female live births, the 45,X karyotype accounts for 10 to 20 percent of the chromosomal abnormalities seen in spontaneously aborted fetuses, demonstrating that almost all 45,X conceptions are lost to miscarriage
Diseases associated with single-gene Mendelian inheritance	Disorders associated with single-gene Mendelian inheritance are typically categorized as autosomal dominant, autosomal recessive, or sex-linked. On average, half the children of an affected parent and a heterozygote are affected, and half are heterozygotes.	X-linked disorders are single gene disorders that result from the presence of a mutated gene on the X chromosome. Because females (XX) have two copies of the X chromosome but males (XY) only have one copy, X-linked disorders are more common in males.

Diseases associated with single-gene non-Mendelian inheritance	Disorders that result from triplet repeat expansions within or near specific genes (e.g., Huntington disease and fragile-X syndrome); a collection of neurodegenerative disorders, such as Leber hereditary optic neuropathy (LHON)	An apparently sporadic pattern of disease incidence is observed if virtually all cases arise as a result of new mutation. No parent to child inheritance of the phenotype is seen, generally because the affected individuals are consistently unable to reproduce through infertility or for physical or social reasons. Occasionally sibling recurrence is seen in such cases, if the parent in whom the new mutation arose has gonadal mosaicism, which is sometimes accompanied by somatic mosaicism.
Mitochondrial DNA mutations Diseases caused by multifactorial inheritance	These disorders involve the influence of multiple genes, generally acting in concert with environmental factors. Such common conditions as cancer, heart disease, and diabetes are now considered to be multifactorial disorders.	The disease can occur in isolation, with affected children born to unaffected parents. Although familial aggregation is also common (i.e., there may be multiple cases in the same family), there is no clear Mendelian pattern of inheritance.

Source: Britannica. (2022). *Human genetic disease*.

Due to the problem of genetic diseases in children, the researchers carried out this study to determine the impacts of these diseases on the sick children's parents in the African region.

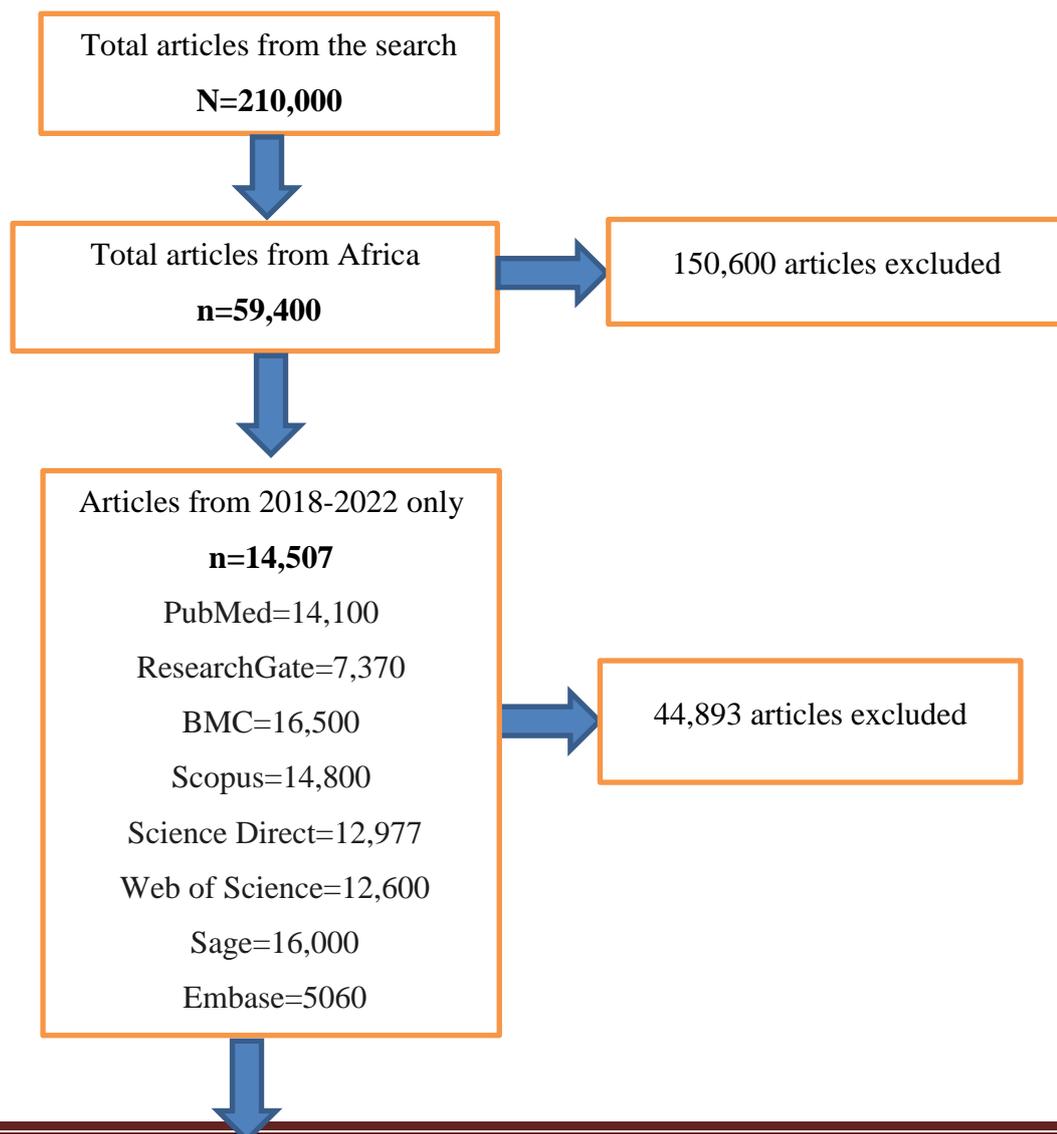
MATERIALS AND METHODS

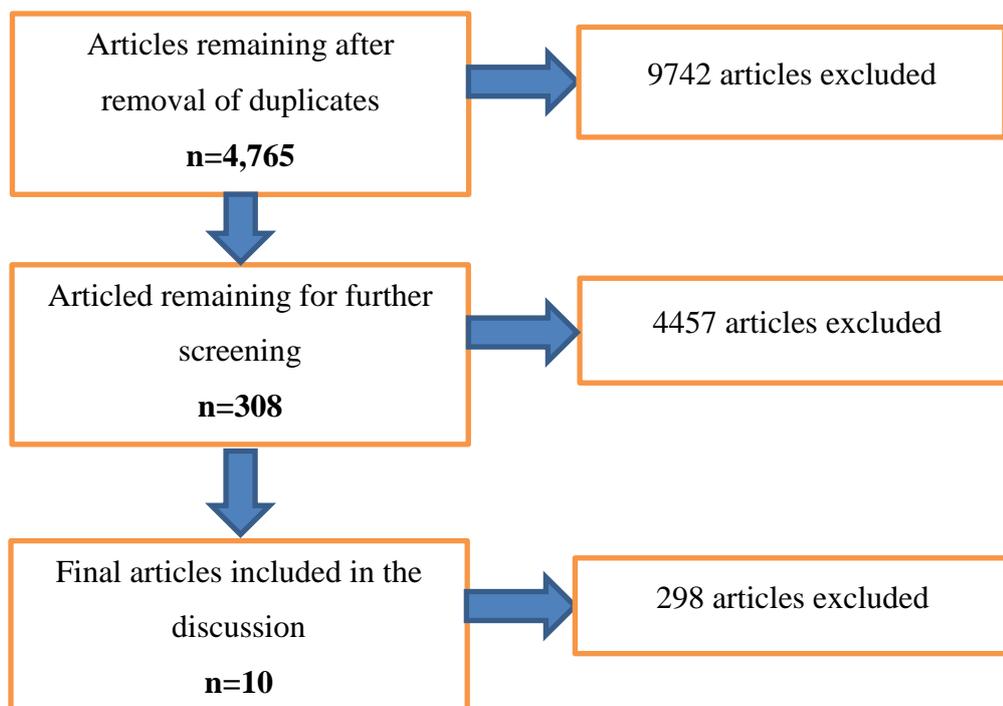
The Google Scholar search engine was used. Eleven electronic literature platforms and databases (i.e. ResearchGate, PubMed, BMC, Scopus, Science Direct, Web of Science, Sage, Embase, CINAHL, PsycInfo and Medline) were searched using the keywords: genetic disorders OR diseases OR conditions, children, parents, Africa OR African region. The inclusion criteria was that only original peer-reviewed qualitative research studies published in peer-reviewed journals and periodicals published in English language between 2018 and 2022 were included in this study. Thematic content analysis and synthesis was employed to analyze and present the findings.

RESULTS AND DISCUSSION

Results

A multistep selection process was carried out. Out of the 4765 identified articles, only 10 studies fully met the inclusion criteria. The inclusion criteria was: original peer-reviewed qualitative research studies, carried out in the African region, published in peer-reviewed journals and periodicals, and published in English language between 2018 and 2022.



**Figure 1: The flowchart****Table 2: The results**

Year of publication	Author(s) and title of article	Source/DOI
2018	Abbey, G. <i>Help-seeking behaviour of parents of children with autism spectrum disorder in Accra, Ghana.</i>	http://ugspace.ug.edu.gh/handle/123456789/27260
2018	Gedleh, A., Lee, S., Hill, J. A., Umukunda, Y., Qaiser, S., Kabiru, J., Kimani, K., Njambi, L., Kitonyi, G., & Dimaras, H. <i>“Where does it come from?” Experiences among survivors and parents of children with retinoblastoma in Kenya.</i>	<i>Journal of Genetic Counseling</i> , 27(3), 574–588. https://doi.org/10.1007/s10897-017-0174-8
2018	Zeleke, W. A., Hughes, T., & Chitiyo, M. <i>The path to an autism spectrum disorders diagnosis in Ethiopia: Parent perspective.</i>	<i>American Journal of Orthopsychiatry</i> , 88(3), 316-327. https://doi.org/10.1037/ort0000249
2019	Gardiner, S. A., Laing, N., Mall, S., & Wonkam, A. <i>Perceptions of parents of children with hearing loss of genetic origin in South Africa.</i>	<i>Journal of Community Genetics</i> , 10(3), 325–333. https://doi.org/10.1007/s12687-018-0396-y
2019	Wonkam, A. <i>Birth defects and genetic disease in Sub-Saharan Africa.</i>	<i>The Genetics of African Populations in Health and Disease</i> . 268–310. https://doi.org/10.1017/9781139680295.012
2020	Havugarurema, P. <i>Knowledge and experiences of parents with children affected by sickle cell disease.</i>	http://dr.ur.ac.rw/handle/123456789/1331
2020	Namisango, E., Bristowe, K., Murtagh, F. E., Downing, J., Powell, R. A., Abas, M., Lohfeld, L., Ali, Z., Atieno, M., Haufiku, D., Guma, S., Luyirika, E. B., Mwangi-Powell, F. N., Higginson,	<i>Palliative Medicine</i> , 34(3), 319–335. https://doi.org/10.1177/0269216319900137

	I. J., & Harding, R. <i>Towards person-centred quality care for children with life-limiting and life-threatening illness: Self-reported symptoms, concerns and priority outcomes from a multi-country qualitative study.</i>	
2021	Haw, T., & Henriques, S. <i>Exploring how mothers of a child with a genetic disorder experience their couple relationship in a low socio-economic setting.</i>	<i>Journal of Genetic Counseling</i> , 30(3), 885–899. https://doi.org/10.1002/jgc4.1391
2021	Kisanga, E., Mutagonda, R., Myemba, D. T., Njiro, B. J., Simon, F., Marealle, A. I., Mikomangwa, W. P., Kilonzi, M., Sambayi, G., & Bwire, G. M. <i>Premarital genetic screening and care of Tanzanian children with sickle cell disease: A qualitative study on parents' views and experiences.</i>	<i>Journal of Community Genetics</i> , 12(4), 515–523. https://doi.org/10.1007/s12687-021-00539-y
2022	Kilonzi, M., Mwakawanga, D. L., Felician, F. F., Mlyuka, H. J., Chirande, L., Myemba, D. T., Sambayi, G., Mutagonda, R. F., Mikomangwa, W. P., Ndunguru, J., Jonathan, A., Ruggajo, P., Minja, I. K., Balandya, E., Makani, J., & Sirili, N. <i>The effects of sickle cell disease on the quality of life: A focus on the untold experiences of parents in Tanzania.</i>	<i>International Journal of Environmental Research and Public Health</i> , 19(11), 6871. https://doi.org/10.3390/ijerph19116871

DISCUSSION

Genetic diseases are chronic in nature. Thus, when a child is diagnosed with a genetic disease, this calls for changes in the family because of the treatment (or lack of it) as well as increased care demands of the child. Genetic diseases impact not only the physical health, but also the psychological and social well-being of not just the patient, but their patient(s) and families as well. The challenges that were established in this study included low quality of life scores in the four quality of life domains. These four domains are physical, psychological, social and environmental health. The other challenges are lack of information, lack of specialized services, financial strain, negative or inappropriate coping mechanisms, as well as existential concerns⁹⁻²⁰.

A study carried out in South Africa showed that, as compared with parents with normal children, the parents of children with autism spectrum disorder had lower mean quality of life scores in the four quality of life domains. The study further showed that the domain where the discrepancy between groups was greatest was the physical domain¹⁰. Another study carried out in Rwanda showed that the respondents had persistent psychosocial and emotional distress and practical hardships¹⁴. Concerning support from spouses, a study done in South Africa, showed that mothers of a child with a genetic disorder frequently felt unsupported by their spouses and carried the responsibility of childcare alone. The majority of the

participants wanted more tangible and emotional support from their partners because without this support they felt isolated and alone¹⁵.

Namisango and colleagues, in their cross-sectional qualitative study in Kenya, Namibia, South Africa and Uganda also revealed physical and psycho-social concerns¹⁸. Another study carried out in Nigeria showed that four in every ten parent caregivers had emotional distress, which was predicted by frequent hospital attendance and disruptions in caregiver lifestyle, relationships, and interests¹¹. Other studies have shown similar results^{12,17,19,20}. Indeed, low quality of life scores cause great frustration and distress. Thus, the parents of a child with a genetic disorder should consult with a genetic counselor in addition to a medical geneticist to enable the provision of emotional support¹⁵. In addition to support from the partner and family, couples may also benefit from the support of others since genetic disorders often have local and national support groups. These organizations can help the affected parents to access resources that make life a little easier. The organizations also host events where families can meet with other families going through similar challenges.

Clinical genetic services are increasingly providing a more enlightening understanding of genetic disease diagnostics and future risk for patients. However, effectively conveying genetic information is essential for patients to make informed decisions. This is because, parental knowledge about the disease has a direct effect on reducing mortality and complications related to the disease. A study carried out in Rwanda showed that the majority of the parents had some knowledge of sickle cell disease but most of them had no knowledge of inheritance pattern of the disease. In fact, prior to the diagnosis being made, majority of the parents had never heard of the disease and they had misconceptions about the condition of their children^{13,14}.

Another study done in Ethiopia showed that the large majority of parents were unaware of the services provided to their children and indicated poor parent–agency coordination²⁰. In contrast, a study done in Dar es Salaam showed that most of the parents of children with sickle cell disease were knowledgeable about the disease as well as genetic testing and counselling¹⁷.

Children with genetic diseases need specialized services and treatment. To access the specialized services, parents in Africa need to travel to referral centers in larger cities, and sometimes those facilities lack proper organization of services. Some families experienced difficulties due to unprepared health professionals^{15,17,19,20}. A study carried out in Ghana showed that the major challenge parents faced was financial challenge mostly due to the cost of specialized treatment⁹. Another financial burden is due to nutrition for the affected child¹⁴. Due to the financial strain, the parents may be at a greater risk of poverty, depression and anxiety^{9,10,14-19}.

In our opinion, it is possible that caring for a child with a genetic disease could result in positive outcomes such as developing positive coping mechanisms like reading about the disease, meditating, listening to music, praying, watching television, and so forth. However, most of the times, the affected parents employ negative or inappropriate coping mechanisms. One study showed that the mothers used a positive coping mechanism of 'acceptance' as their coping strategy. In contrast, their partners used negative coping mechanism of escape-avoidance strategies such as abandonment, denying paternity, withdrawal, and partner-blame¹⁵. Another challenge faced by parents is existential concerns whereby the parents are constantly worried about the death of their child¹⁸. Indeed, all the studies concur that specialized services should be instituted through instituting national policies and programs to provide better screening and detection of genetic disorders, coordinate clinical management, facilitate training of medical genetics/genomics professionals, and stimulate research into genetic disorders.

CONCLUSION

The study concluded that parents of children with genetic diseases face many challenges such as: (i) low quality of life; (ii) lack of information; (iii) lack of specialized services; (iv) financial strain; (v) negative or inappropriate coping mechanisms; and (vi) existential concerns. These challenges are due to the chronic nature of genetic diseases, and has far reaching effects on the affected parents and their sick children, physically, psychologically and socially. The psychosocial and emotional distress is too much for the affected parents and their families. The personal and permanent nature of genetic disease can raise a range of emotions including guilt, fear, and helplessness.

The study recommends that: (i) counselling and social support for affected parents to help mitigate the impact of genetic diseases; (ii) raising awareness of genetic diseases among the affected parents and their families, as well as the public in general; (iii) international and national policies and programmes should be instituted to enhance screening and detection of genetic disorders, coordination of clinical management of genetic diseases, facilitation of training of medical genetics/genomics professionals. Specialists such as genetic counselors, social workers, and psychologists, as well as members of support groups, can be extremely helpful to patients and families as they deal with these difficult issues. It is therefore imperative that focusing on the child's overall well-being, not solely on the child's genetic condition at routine visits is helpful to the affected parents. It is important to talk about the newborn's personality, feeding patterns, and other personal traits. It is important to always remember that the newborn is an infant first and an infant with special needs second; stimulation of research in genetic diseases; (iv) ensure affordability of treatment and

healthcare services to parents with children with genetic diseases; (v) promote resilience and positive adaptive coping mechanisms; as well as (vi) provision of trauma counselling to the affected parents.

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