affiliation with an HIV/AIDS organization. The study conducted with CCDT specifically included a cohort that has gone through a full disclosure process with family members, and received the appropriate medical treatment. Though initially there were uncertainties with illness disclosure, over time parents understood the importance of talking about the illness and included children in the discussions. The aim of our study was to focus on CCDT’s disclosure practices and learn about the process from parents’ and children’s perspective. In order to keep the study objective concise, there was no mention of the national level non-disclosure gaps.

The argument you have made in your letter is a sincere concern for India. Even though with the available resources and free treatment, non-disclosure is still a detrimental public health issue for the country. Families are in constant fear of the stigma associated with the illness. In order to address this further, more qualitative and quantitative data need to be generated. There are numerous HIV/AIDS NGOs working independently on this issue; yet there is a lack of collaboration on effective programming and valuable practices. Without stable infrastructure, laws are not sufficient to encourage disclosure. There is a great need of direct ground level efforts, and collaboration between NGOs and providers to tackle this challenge. This is feasible through stricter government policies, increased countrywide discussions to destigmatize the illness, and continuous efforts from health providers and NGOs to educate infected families on the importance of disclosure.

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Trisomy 8 Mosaicism in a Boy with Dysmorphic Features

Trisomy 8 mosaicism is a rare condition with prevalence estimates in the range of 1:25000-1:50,000 births. It is a rare genetic disorder and clinically heterogeneous condition associated with a spectrum of developmental abnormalities, including intellectual disability, congenital heart defects and agenesis of corpus callosum [1,2]. Approximately 50% of these patients present with renal abnormalities [3]. We report a boy with dysmorphic features and delayed development.

A 10-year-old boy with delayed development and dysmorphic features was referred to us for cytogenetic analysis. The proband was first born child to consanguineous parents. He had short stature (height 121 cm, US: LS=0.86), dolicocephaly, broad nose with anteverted nostrils, flat tip pinnae, bilateral limited extension of elbow, restricted joint movements, bilateral comptodactyly, bilateral radial head subluxation, bilateral femoral neck coxa valga, squint, tongue tie, webbed neck and agenesis of corpus callosum. He had vestibular hypersensitivity, and fear of swings, heights and climbing of ladder. Psychological examination showed moderate sub-normality in social functioning. Radiological examination showed generalized osteopenia; electroencephalography (EEG) and thyroid function tests were normal. The cytogenetic analysis using GTG banding revealed mosaic trisomy 8 in 25% of metaphases scored. Fluorescence in situ hybridization (FISH) analysis using Vysis centromeric probe for chromosome 8 showed 59% cells with trisomy 8, and the karyotype was determined as mosaic trisomy 8 (46,XY/46,XY+8).

Trisomy 8 mosaicism occurs due to non-disjunction of chromosome 8 during mitosis in the zygote phase of fetal development. This condition is clinically heterogeneous, and it is associated with wide range of clinical abnormalities [1-4]. Our patient had additional clinical features: restricted joint movements, bilateral comptodactyly, bilateral radial head subluxation, bilateral femoral neck coxa valga, squint, tongue tie and webbed neck. Correlation of genetic abnormalities with clinical phenotype is always important to establish the syndromic diagnosis. The follow-up of mosaic trisomy 8 is essential, as it is more commonly seen in patients with acute myeloid leukemia and myelodysplastic syndrome.

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REFERENCES
Predictors of Survival in Children with Methymalonic Acidemia with Homocystinuria

We read with interest the recent report by Qiliang, et al. [1] on the outcome of 45 children diagnosed with combined methymalonic academia (MMA) and homocystinemia. The authors report a 40% mortality in their cohort. Apart from mortality, it would be important to know the degree and pattern of neuromorbidity in the survivors. Combined MMA and homocystinemia is a potentially treatable inborn error of metabolism, and high mortality and possibly high morbidity in the reported cohort need a careful evaluation. The high mortality in the reported cohort can partly be explained by the inadequate parenteral B12 replacement given to the patients. Highlighting the importance of meticulous long-term treatment, we wish to point out certain important aspects of management of children with combined MMA and homocystinemia.

The critical component of the treatment of combined MMA and homocystinemia is parenteral B12. This therapy has to be given in the adequate doses daily and lifelong. Hydroxyl-cobalamine injections are the only form of B12 proven to be beneficial in patients with this disorder. It is recommended that hydroxyl-cobalamine be given daily intravenously, subcutaneously or intramuscularly. The recommended dose of parenteral hydroxyl-cobalamine is 0.3 mg/kg/day, once a day. The suggested targeted plasma B12 levels are ≥1,000,000 pg/mL [2]. Previous reports have shown that the progression of complications in patients with combined MMA and homocystinemia arise in part due to inadequate hydroxyl-cobalamine.

There are several reports of marked clinical and neurological deterioration in patients weaned from daily to less frequent dosing [3,4]. Hence, it is essential for all involved in the care of affected individuals to ensure daily administration of the injection. The monitoring parameters include serum MMA and total homocystine levels and normalization of plasma methionine and hematological parameters. The other co-factors recommended for use include oral Betaine (250 mg/kg/day in 3 divided doses), oral Folinic acid (5-15 mg/day in 2-3 Divided doses), and oral Levocarnitine (50-100 mg/kg/day in 3 divided doses).

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REFERENCES

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