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ADVANCES IN PEDIATRIC RESEARCH

Silver-Russell syndrome with unusual clinical features: A case report

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Abstract

The Silver-Russell syndrome (SRS) is a rare heterogeneous genetic disorder whose pathogenesis remains controversial. The diagnosis is mainly based on the clinical association of characteristic signs, including intrauterine growth retardation, postnatal short stature, relative macrocephaly, triangular facies, clinodactyly of the fifth finger and asymmetry of the body. In this case report, we focused on a patient with SRS, who was spotted during a routine paediatric plastic surgery campaign in Ouagadougou, Burkina Faso (West Africa). The propositus is a young male with evident poor weight gain, relative macrocephaly, large and fibrous anterior fontanel, triangular facies, frontal bossing, hypertelorism, kypho-lordosis, clinodactyly and camptodactyly. The specific SRS features in this patient included flat feet and clubbing digit. In the subsequent follow-up, the patient revealed a few alterations in the craniofacial anomalies, but with heightened intellectual and psychological issues. The diagnosis, evolution and prognosis of this syndrome are discussed in the report, including the necessity for a clinical diagnostic score for SRS and other congenital malformations to aid clinicians in developing countries that are deprived of access to molecular or genetic diagnostics.

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Introduction

Silver–Russell syndrome (SRS) (OMIM #180860) [1] is defined as "a distinct syndromic growth disorder in which prenatal and postnatal growth failure are associated with other characteristic features, including relative macrocephaly at birth, protruding forehead in the early life, body asymmetry and substantial feeding difficulties. Almost all children with SRS are born small for gestational age (SGA). Postnatal catch up growth is uncommon in most of the children with SRS." [2]. This rare genetic disease ranks Q87.1 in the International Statistical Classification of Diseases and Related Health Problems' 10th Revision [3], and it is estimated to affect 1–30/100,000 individuals, with a prevalence of 1–9/1,000,000 live births. Globally, only around 400 cases have been reported until date [4]. The disease equally affects all races [5-7] and both the sexes [8].

Historically, in 1953, Silver et al. [9] first described this disease, followed by Russell in 1954 [10]. Silver et al. [9] described two children with congenital hemi-hypertrophy, low birth weight, short stature and increased urinary gonadotropins. Russell [10], however, reported five children with intrauterine growth retardation and anomalies of the skull and face. Even as both the authors believed they were describing two different conditions, the two phenotypes were later identified as a single entity and the disease was thus classified as SRS, using both their names [11].

From the perspective of heredity, transmission is observed to be sporadic or autosomal dominant,

although some alternate routes of inheritance have been reported [4].

At present, the two genotypic anomalies clearly recognisable in the initial stage of the disease include loss of methylation (LOM) on chromosome 11p15, imprinting the centre region 1 (ICR1) [12] and the maternal uniparental chromosome 7 disomy (upd(7)mat) [13]. The 11p15 ICR1 bears the genes responsible for the growth-regulating proteins, and the LOM in this region induces characteristic growth retardation in SRS. In fact, 44.6% (15-64%) of the SRS phenotypes show LOM, and the upd(7)mat accounts for 4.5% (2.3-11.4%) of the phenotypes. Besides SRS, there have been rather very rare reports of other duplications (1-2%) and chromosomal aberrations (1%) [14-16], which leaves around 50% of the SRS cases with unknown aetiology.

Great interest and numerous reasons are accorded to the understanding of this polymalformative syndrome and of this present case of SRS. First, SRS is relatively uncommon [4], with only very limited cases recorded in Africa [6,17], and the present instance is, to the best of our knowledge, the first case of SRS reported in Burkina Faso (West Africa). The clinical diagnosis of SRS is quite difficult, as several other syndromes show similar phenotypical characteristics [3,18-20]. The patient in this study reveals peculiarities not catalogued in the characteristics of SRS. Finally, although as a congenital affliction, there is no cure for SRS, prevention through genetic counselling is useful in familial cases of SRS.

Case Report

T.I. visited the Canadian Humanitarian Paediatric Surgery Mission "Sourires d'Afrique" (www.souriresdafrique.com) for consultation on 29 November, 2007. T.I was 11-year-old and manifested delayed closure of the anterior fontanel, spinal deformity and short stature. According to his medical history, T.I. was the outcome of a normal mono-foetal pregnancy without any complications that culminated in normal delivery at term.

At birth, his weight was 2480 g and body length 48 cm, with the head circumference (HC) of 32 cm and thoracic perimeter of 31 cm. Referring to the WHO child growth standards charts, the HC-for-age was comprised between 0 and +1 z-score, height-for-age was comprised between -1 and -2 z-score, weight-for-age equal to -2 z-score, weight-for-height was comprised between -2 and -3 z-score, and BMI-for-age is 10.77 kg/m^2 comprised between -2 and -3 z-score. The Apgar score was 6, 8 and 10, respectively, at 1, 5 and 10 min. The growth-monitoring chart showed a gradual shift in weight from 5 months of age, leaving the corridor of normal weight-for-age and entry into under nutrition area at 8 months of age without catching-up to the 21 months of age.

During the course of interrogation, it was revealed that the patient suffered from language and cognition development delay. His biological parents are not in a consanguineous marriage; his father is a trader aged 29 years, and the mother is a housewife of age 20 years, G4 P3, with a stillborn due to malaria.

T.I. is the eldest of the three siblings, with both the younger sisters in apparently good health. He is enrolled in the elementary school, grade two and has encountered learning difficulties.

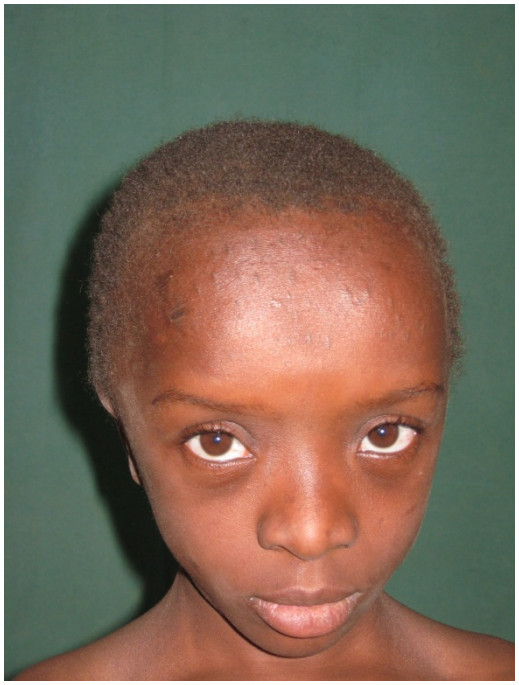


Figure 1. The Silver–Russell syndrome in the patient T. I., showing the triangular facies characteristic with relative macrocephaly, frontal bossing, mandibular hypoplasia and small chin (Photograph by B. Fougères).

On physical examination, the patient showed a general good condition, a weight of 18.5 kg, HC of 52 cm, height of 119 cm and a weight-to-height comprised between -1 and -2 z-score. His BMI of 13.1 kg/m² was comprised between -2 and -3 z-

score, indicating thinness. His skull and face examination revealed triangular facies with relative macrocephaly, prominent broad forehead with large bumps, mandibular hypoplasia and micrognathia (Figure 1). The anterior fontanel was broad, fibrous and non-pulsatile. Kypho-lordosis was evident, with the scapular spines showing detachment (Figure 2). The hands showed a clubbing digit, bilateral clinodactyly of the fifth finger and camptodactyly of the fifth right finger (Figure 3). Both his feet were flat. The rest of the examination was normal.

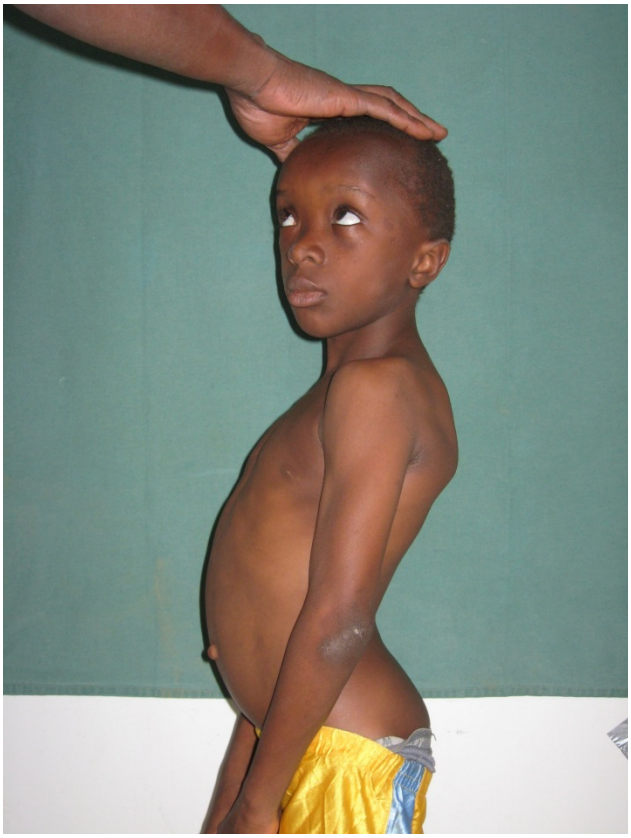


Figure 2. The Silver–Russell syndrome in the patient T. I. showing kypho-lordosis and scapular spines detachment (Photograph by B. Fougères).

No cytogenetic or molecular investigations were performed. The set of clinical signs observed facilitated the diagnosis of SRS. No growth hormone treatment was performed.

When the second examination was performed on 20 June, 2017, T.I. was of age 20 years and 6 months. His complaints, according to the mother, included his short stature and absence of weight gain despite having a good appetite. The young man also reported excess sweating.

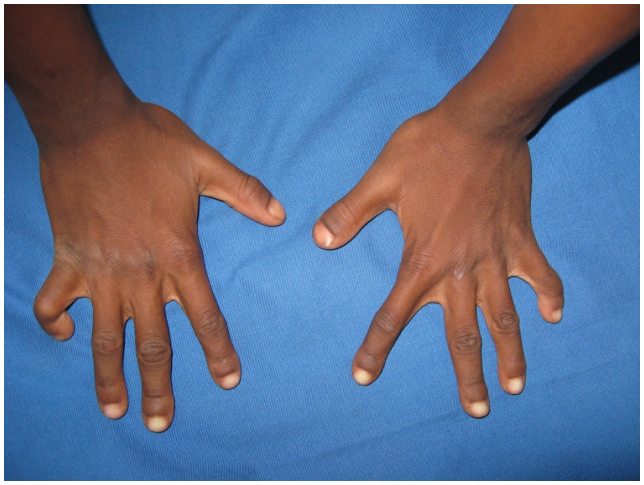


Figure 3. The Silver–Russell syndrome in the patient T. I. having clinodactyly, camptodactyly, and clubbing digit (Photograph by B. Fougères).

On examination, the patient weight was 45 kg, height 147.5 cm with a BMI of 20.7 kg/m², indicating a normal weight. The HC was 55 cm and the mid-parental height was 184 cm or 84.4% height percentile. The patient had strong muscles, the anterior fontanel was closed, but the craniofacial, skeletal, spinal anomalies (Figures 4 and 5) and extremities anomalies remain unchanged.

Figure 4. The Silver–Russell syndrome in the patient T. I. His craniofacial pattern at 20 and 6 months of age (Photograph by K. Nagalo).

A sort of claudication approach was evident; the pelvic right limb was 88-cm long, the left one being 91 cm, giving a difference of 3 cm in length. The palate was high arched and the teeth showed decay, with the pubertal development and language ability appearing normal. Intellectually, the patient

experienced memory lapses with academic delay; he is currently in secondary school, grade five and repeating each class. Psychologically, he feels ostracised by his classmates, which has given him a complex. He sometimes has outbursts of unjustified and excessive anger and uses both alcohol and tobacco. The count of blood cells, glycaemia, serum electrolytes (calcium and phosphorus) were normal. Table 1 summarises the phenotype presented by the patient, and Figure 6 shows the growth chart through two follow-up visits.

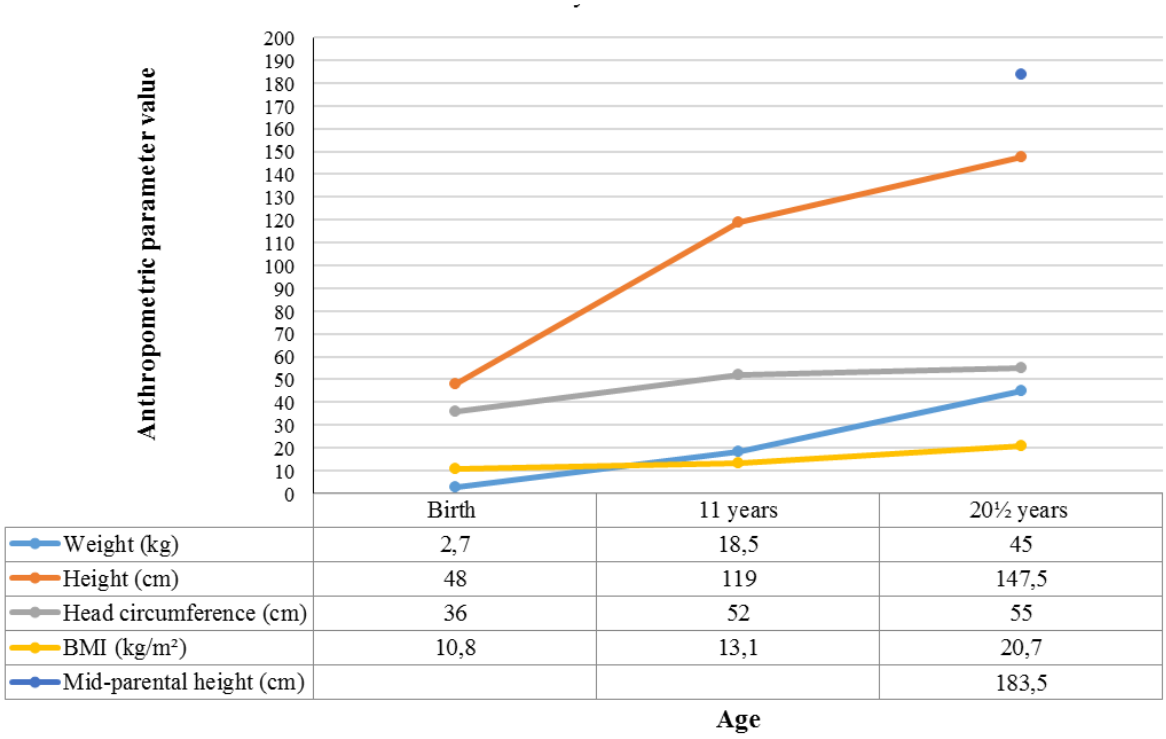
Figure 5. The Silver–Russell syndrome in the patient T. I. His craniofacial and spine anomalies at 20 and 6 months of age (Photograph by K. Nagalo).

Table 1. Summary of the phenotype of the Silver–Russell syndrome in the patient T.I.

Clinical signs*	**Patient**
Major characteristics	
Small for gestational age	+
Postnatal growth failure	+
Relative macrocephaly at birth	+
Protruding forehead	+
Body asymmetry	+
Feeding difficulties and/or low BMI	+
Associated signs	
Triangular face	+
Fifth finger clinodactyly	+
Shoulder dimples	+
Micrognathia	+
Low muscle mass	-
Excessive sweating	+
Low-set and/or posteriorly rotated ears	-
Down-turned mouth	-
High-pitched squeaky voice gold	-
Prominent heels	+
Delayed closure of fontanel	+
Male genital anomalies	-
Speech delay	+
Irregular or crowded teeth	+
Motor delay	-
Syndactyly of toes	-
Hypoglycaemia	-
Scoliosis and/or kyphosis	+
Peculiar signs	
Flat feet	+
Clubbing digit	+

*Adapted from Wakeling et al. 2017 [2]

+ present; - absent.



	Birth	11 years	20½ years
Weight (kg)	2,7	18,5	45
Height (cm)	48	119	147,5
Head circumference (cm)	36	52	55
BMI (kg/m²)	10,8	13,1	20,7
Mid-parental height (cm)			183,5

Figure 6. The follow-up of the growth of the patient T.I. presenting with the Silver-Russell syndrome.

Table 2. Frequency of the main clinical signs of the Silver-Russell syndrome.

Clinical sign	**Frequency**		
	Netchine, 2007 [22]	Kotzot, 2008 [23]	Wakeling, 2011 [24]
Growth			
Birth weight ≤ -2 SD	94	94	70-82
Height at examination ≤ -2 SD	85.7-96		57-65
Relative macrocephaly	84.6	68.4-92	70-90
Body asymmetry	64.1	53.2-77	30-68
Development			
Global retardation	23.1	20.5-43	20-6
Skull and face			
Triangular facies		76-97	59-90
Bulging forehead	92.3	68-88	60
Micro/retrognathia		44-73	35-64
Dental anomalies		28-64	36-45

Down-turned corners of the mouth		50-57.3	20-30
Low-set ears		40.3	36-75
Other clinical sign			
Clinodactyly 5th finger	64.1	69.9-82	45-75
Syndactyly 2/3 toes			20-23
Camptodactyly			16-25
Arthrogryposis			0-11
Café-au-lait spots	17.9		14-15
Other aspects			
Feeding difficulties	82		84-90
Sweating		67.4-75	64-75
Hypoglycaemia	15.4		24-29
Gastro-oesophageal reflux		12.5-15.2	10-14
Delayed closure of fontanel			36-43
Associated congenital malformations			10-36
Myoclonus-dystonia			0-15

Table 3. The diagnosis of the Silver-Russell syndrome in the patient T.I. according to the NH-CSS criteria.

NH-CSS criteria	Patient
Small for gestational age	+
Postnatal growth failure	+
Relative macrocephaly at birth	+
Protruding forehead	+
Body asymmetry	+
Feeding difficulties and/or low BMI	+
+ present; -absent	

Table 4. Differential diagnosis of the Silver-Russell syndrome.

Microcephaly	Bloom syndrome
	Nijmegen breakage syndrome
	MOPD II
	Meier-Gorlin syndrome
	IGF1R mutation or deletion
	IGF1 mutation
	Foetal alcohol syndrome
Normocephaly or macrocephaly	3-M syndrome
	Mulibrey nanism
	SHORT syndrome
	Floating-Harbor syndrome
	IMAGe syndrome
Other	Osteogenesis imperfect
	Rickets

Discussion

The place of congenital anomalies and SRS in developing countries

In the working conditions of a hospital in a developing country, we encountered several deformities, but the lack of amenities, of a geneticist and of a Human Genetics Unit made these anomalies remain underdiagnosed with accuracy. In the light of the high-impact conditions of neonatal morbidity and mortality, such as neonatal sepsis, perinatal asphyxia, and prematurity, congenital anomalies are largely neglected by both health authorities and health professionals. This is true even for SRS presented in this case study, the diagnosis of which was made fortuitously during a Canadian Humanitarian Mission for Paediatric Plastic Surgery in Burkina Faso.

The diagnosis of SRS

The historic cardinal clinical signs of SRS include intra-uterine growth retardation without postnatal catch-up, relative macrocephaly, a prominent forehead with large bumps and hypoplasia of the jaws that ends in a point by a micrognathia, which altogether give the face a triangular appearance [9,10].

Body asymmetry, a clinodactyly of the fifth finger and more recently feeding disorders have been associated with SRS [14,18,21]. The main clinical signs and their frequency in some studies are listed in Table 2.

Infrequent, uncommon, or rare anomalies may be associated with SRS, such as endocrine [25], urogenital, skin [6,26], heart [26–28], eye [19,25,29], neurological [25,30,31] and psychiatric [32] anomalies, besides the appearance of tumours [1,4].

To minimise the risk of over/underestimation of SRS cases and enable easier diagnosis, several clinical scores have been proposed [5,8,22,33,34]. At present, the diagnosis of SRS is based on the "Netchine–Harbinson clinical scoring system" (NH-CSS). When compared to other scores, NH-CSS offers the advantage of high sensitivity (97.9%), low specificity (36.4%) and high negative (88.9%) and positive (76.7%) predictive values in addition to being easy to use. The NH-CSS was proposed by Azzi et al. [35] and includes six criteria: (1) prenatal growth retardation; (2) postnatal growth retardation; (3) relative macrocephaly at birth; (4) protruding forehead; (5) body asymmetry; and (6) feeding difficulties and/or low BMI. Table 3 summarises the signs of SRS in patients with reference to the NH-CSS criteria.

The patient met all NH-CSS six criteria and was therefore considered as a clinical SRS. Even though none of the scores were perfect, they are definitely useful in diagnosing SRS for clinicians in developing countries, who are generally unfamiliar with SRS and lack access to specialised laboratories for genetic and molecular tests.

Molecular tests can confirm the diagnosis of SRS in around 60% of patients [22]. This means that a negative result does not exclude the diagnosis of SRS. Hence, the experts recommend that the diagnosis of SRS remains essentially clinical, as molecular tests are used for confirmation and classification of patients in the sub-groups (level A +++ recommendation); therefore, the NH CSS flow chart must be adopted for the diagnosis of SRS (level A ++ recommendation) [2].

Singular aspects observed in this patient include flat feet and clubbing digit. Even when these signs are minor clinical signs, our literature review did not reveal flat feet in SRS. Similarly, clubbing digit has not been reported among signs associated with SRS. After excluding obvious causes akin to cyanogen heart disease or chronic respiratory failure, clubbing has been related to SRS.

The diagnosis of SRS is difficult due to its phenotypic similarity with other syndromes [3,18-20] and the highly heterogeneous clinical forms that this disease presents. The diagnosis is more difficult in our situation; we discussed the other similar syndromes and withheld the diagnosis of clinical SRS according to the recommendations of the consensus statement on SRS [2]. The syndromes used to differentiate SRS in a child with intrauterine growth retardation, short stature and craniofacial anomalies are listed in Table 4.

Because of the lack of funding and essential facilities, we could not perform molecular tests. Burkina Faso is a developing country that lacks the availability of laboratories for molecular tests.

According to our experience, we considered sending the samples to advanced countries for analysis and accurate examination. However, even if such facilities were locally available, the costs incurred in doing so would have been exorbitant and out of limit of the considered patient's parents. The accessibility for such genetic tests must necessarily be made locally; given the protean presentation of SRS, clinicians will require performing confirmatory tests to facilitate the diagnosis when they encounter patients exhibiting signs that indicate this syndrome [2,24].

The correlation between SRS phenotype and the genetic anomaly responsible is well established. In LOM, patients have phenotypic characteristics of the SRS, although the signs are moderated in upd(7)mat [7,8,14,22,24,35]. In the present case, the dysmorphic features and other clinical signs exhibited by the patient direct to the hypothesis of 11p15 LOM. On the other hand, however, the patient manifested intellectual disorders that are especially described in upd(7)mat [7,24]. We unfortunately could not collect molecular test evidence to confirm the genotype of the SRS in this patient.

Evolution and prognosis of SRS

One of the distinct features of SRS is the attenuation of the signs over a period of time, which impedes the clinical diagnosis even more in older children and adults. If treatment with growth hormones is not administered, the final adult height gets reduced to 142.5–145 cm in males and 146.5 cm in females [30]. In the current case, the dysmorphic signs remain constant. A height of 147.5 cm is under mid-parental height, albeit quite close to the adult height reported for the disease. SRS, however, has a comparatively good prognosis. In fact, Russell described a patient in 1954 [10] who was doing satisfactorily well at 69 years of age despite having type 2 diabetes, hypercholesterolemia, hypogonadism and osteopenia [36]. Normally, while fatality is the exception, rather than the rule in SRS, Africa has recorded cases of "sudden" infant deaths [6]. These deaths, explained as "sudden" by the family members, most often carry the suspicion of infanticide, which is a common occurrence with infants having congenital malformations [37]. We propose that the main drawback with the present patient was that his intellectual deficit will continue throughout his lifetime. Although most children with SRS show normal intelligence level [5,18], 37–50% of them encounter intellectual difficulties [33,38], with some type of moderate, but significant decline in the intelligence quotient, reading performance, arithmetic ability and learning skill [15,39]. Therefore, these children require specific educational needs, like this young man who is at present facing academic failure. However, continuous education in a speciality centre may train him and improve his options of working toward successfully integrating into his society.

Conclusion

SRS ranks among the cosmopolitan polymalformative syndromes, and similar to other congenital malformations, continues to remain underdiagnosed in the developing countries. Therefore, in these countries, clinical diagnostic scores are highly beneficial for diagnosis. Hence, we have described uncommon clinical signs of SRS, which were distinguished by flat feet and clubbing digit.

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