The Seckel syndrome: A case observed in the pediatric department of the University Hospital Center Sourou Sanou (Burkina Faso)

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Abstract

Seckel syndrome-1 or "bird-headed dwarfism", Online Mendelian Inheritance in Man number 210600, is a rare genetic disease with an autosomal recessive transmission. We report a female child of 56 months diagnosed with SCKL1 at the Pediatric department of the University Hospital Center Sourou Sanou, Burkina Faso. She showed the typical features including facial dysmorphism, dwarfism, microcephalus and mental retardation. Ophthalmic and dental anomaly and extremities were associated. Without a codified etiological treatment, a psychotherapist support, a genetic counseling, a regular pediatric follow-up, a quarterly odonto-stomatological and ophthalmological follow-up have been recommended.

Introduction

Seckel syndrome-1 or "bird-headed dwarfism", Online Mendelian Inheritance in Man number 210600 , is a rare genetic disease with autosomal recessive[1] inheritance caused by homozygous or compound heterozygous mutation in the ATR gene (Gene/locus Mendelian Inheritance in Man number 601215) on chromosome 3q23.[2] It is a morphological entity defined by four criteria combining intra-uterine growth retardation (IUGR) exceeding two standard deviations, microcephaly often correlated to a craniosynostosis, facial dysmorphism and mental retardation of variable degree.[3,4]

SCKL1 has been reported from different geographic areas worldwide, including Africa,[5] with a prevalence of 1/10.000.[6] We report the observation of Seckel syndrome in a child at the University Hospital Center Sourou Sanou (UHCSS) in Bobo-Dioulasso.

Case Report

D.P, a female child of 56 months, was referred by the Saint Leopold clinic of Bobo-Dioulasso to UHCSS in pediatric consultation on March 1st, 2017 for growth and mental retardation. She was born vaginally at the 43 week with an IUGR (birth weight 2120 g), from a consanguineous marriage (Figure 1). A retardation of psychomotor development was progressively documented with head control since 7 months, sitting since 8 months, standing and walking position acquired at 2 years, and markedly delayed language landmarks. On admission, the child showed a severe growth retardation: the weight at 11,80 kg (Weight for Age percentile = 0.1%; Weight for Height percentile = 0.8%), the height at 97,6 cm (Height for Age percentile =2.0%) and the head circumference at 39 cm (Head circumference for Age percentile =0.0%).[7] Her face was dysmorphic with a prominent beaked nose, facial asymmetry, low-set ears and a micrognathia; curly and brittle red hair; campodactyly and clinodactyly of the 5th finger (Figure 2). Ocular evaluation showed bilateral convergent strabism and retinal vascular tortuosity, with normal intra ocular pressure. Mental retardation was characterized by a delay in language and understanding, posture of the head, sitting, walking and standing. She also had emotional fragility with high dependence for activities including coloring, speech, performing games. Her IQ was estimated between 35-49. Bone age was 36 months. The child was prospectively followed for supportive care. At the age of 6 years, she is clinically stable, and her main limitation is due to the severe dwarfism and facial dysmorphism, which heavily limit her social activity. Furthermore, her eyesight may be progressively limited by retinal vasculopathy.

Discussion

In a review of literature, Kalay *et al.* reported a case of Seckel syndrome in South Africa;[6] to the best of our knowledge, this is the first case diagnosed in Burkina Faso. The failure to thrive was the cause of consultation in our patient, diagnosed at the age of 4 years and 8 months, as reported by Thapa *et al*[8]. The diagnosis has been reported at any age, even antenatal through medical imagery viewing[1,9] (echography in 3D, IRM).

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Ethics approval and consent to participate: This study was conducted in accordance with all relevant guidelines and procedures.

Informed consent: Informed consent has been obtained form the family.

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The observation of parental consanguinity is in keeping with an autosomal recessive disorder.[1,2]

The differential diagnosis is mainly with osteodysplastic dwarfism with type II microcephaly.[10]

In our medical and social setting with limited resources, the diagnosis of rare conditions may be based on clinical observation and recognition of the typical phenotype. Unfortunately, we do not have access to molecular studies which might confirm mutation in the associated gene. Faivre *et al*.[11] confirmed the heterogeneity of Seckel syndrome by excluding the previously mapped loci on chromosomes 3 and 18 in 5 consanguineous and 1 multiplex nonconsanguineous Seckel syndrome families.

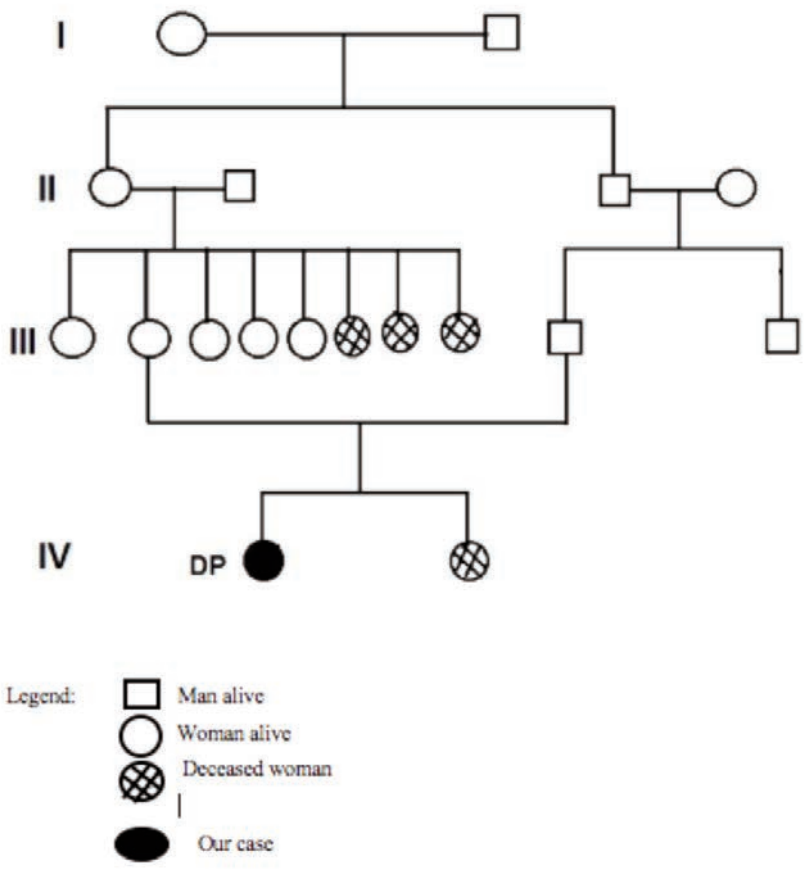

Figure 1. Family tree.

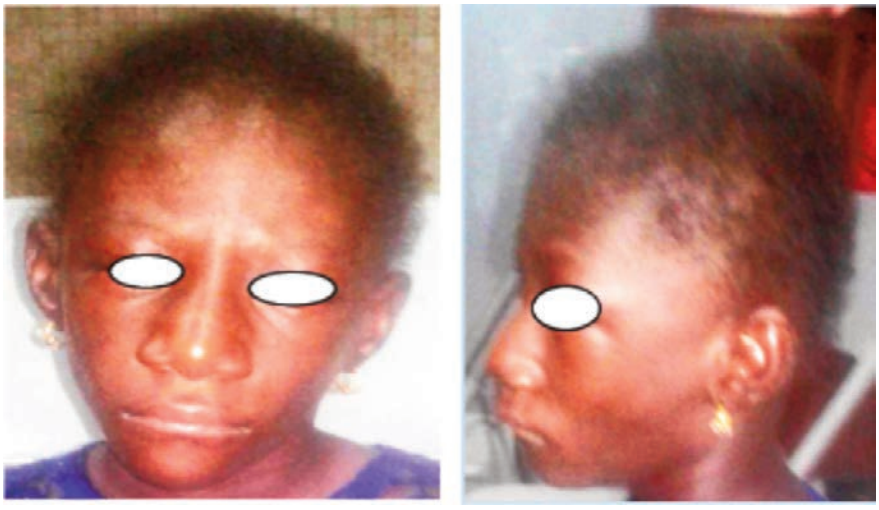

Figure 2. Characteristic facial dysmorphism with prominent beak-shaped nose, micrognathia, low-set ears.

Conclusions

This observation made it possible to highlight the limits in diagnosis confirmation namely in the carrying out of molecular biology.

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