Growth Pattern in children affected of Lowe Syndrome

Descriptive multicenter international study: Preliminary data

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Background

Lowe Syndrome or oculocerebrorenal syndrome is a very rare condition (1:50000) caused by mutations in the OCRL1 gene. It is an X-linked disorder characterized by congenital cataracts, renal tubular dysfunction, neurological defects (generalized hypotonia and mental retardation) and growth disorders. Growth pattern in Lowe Syndrome has not been described in population-based studies so far.

Objective

Descriptive multicenter international study aimed to describe growth pattern in Lowe Syndrome affected subjects.

Method

47 subjects have been identified around the world. Auxologic data of 14 subjects (age 3-24 years) with Lowe Syndrome from Spain (n=8), Italy (n=5) and Argentina (n=1) were reported through a web-based questionnaire.

Results

Length/height is expressed as followed in media ± standard deviations. Weight is expressed as followed in media ± standard deviations. Only one subject older than 13 years was identified with the following height measurements: height at the age of 13, 15, 16 and 23 were 130, 135, 149 and 158 cm respectively.

Age (month)	n	Media (Kg)	SD
3	8	5,729	0,76
6	11	7,127	1,01
10	8	7,970	1,18
12	8	8,100	1,51
18	8	9,430	1,31
24	10	10,000	1,74
36	9	11,350	2,39

Weight 0 to 36 months

Age (month)	n	Media (cm)	SD
3	11	61,2	2,75
6	10	65,8	1,94
9	10	69,7	2,39
12	9	71,3	4,97
18	9	75,4	3,37
24	9	78,9	7,13
36	6	84,6	7,43

Length / Height 0 to 36 months

Age (years)	n	Media (Kg)	SD
4	9	12,470	2,59
5	6	14,700	3,26
6	6	15,130	3,44
7	6	17,100	3,73
8	5	18,300	3,18
9	5	18,300	3,18
10	3	25,700	1,76
12	2	28,000	5,66

Weight 2 to 12 years

Age (years)	n	Media (cm)	SD
4	6	86,8	6,38
5	4	90,4	7,82
6	4	96,8	5,19
7	3	102,0	5,29
8	4	106,2	4,54
9	4	111,3	4,92
10	3	118,3	1,53
12	2	124,3	0,35

Height 2 to 12 years

Conclusion

Data regarding growth patterns of children affected of Lowe-Syndrome would enhance to design specific growth charts for children with this condition.

Lowe Syndrome as well as other rare conditions requires joint efforts and advocacy of both healthcare members and patient´s families to go further in the knowledge of the disease.

"Rare Commons" online platform is a collaborative project network (including families and doctors) to gain knowledge in rare diseases.

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Growth.

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