

Table 2

Occurrence of extracutaneous symptoms as reported in 23 articles

<b>CNS</b>	Patients	References
Mental and motor retardation	55	(1, 4, 7, 9-16)
Cramps / seizures	41	(1, 4, 5, 7, 10-12, 15, 17-20)
Delayed motor and mental development	20	(2, 11, 13-22)
Hypotonia	15	(1, 5, 11, 17-20, 22, 23)
Microcephaly	12	(1, 7, 12)
Other	16	(5, 7, 11, 14, 15, 17-21)
<b>Musculo-skeletal</b>		
Cranio facial abnormalities	24	(4, 5, 12, 13, 17, 20, 20, 22)
Scoliosis	17	(1, 2, 6, 16, 17, 23)
Short stature	12	(1, 4)
Hemihypertrophy	10	(3, 7, 10, 13, 17, 19, 21)
Clinodactyly	9	(1-3, 9, 16)
Thorax-deformities	9	(1, 5, 13, 21)
Other	39	(1-6, 9, 11-14, 17-23)
<b>Ocular</b>		
Hypo-/Hypertelorism	9	(1, 2, 9, 14-17, 22)
Other	35	(1, 2, 5, 7, 11, 14, 15, 17, 20-23)
<b>Cardiac</b>		
Other	6	(1, 20, 22)