

WEB TABLE I INDIVIDUAL PATIENT DETAILS AND SOFTWARE PREDICTIONS

No.	Age/Sex	Clinical gestalt diagnosis	Molecular/Cytogenetic diagnosis	Predicted as first diagnosis	Correct diagnosis in first ten diagnoses	Correct diagnosis after additional information
1.	4 y/M	Mucopolysaccharidosis	MPS II	Yes	Yes	-
2.	4 mo/F	Mowat Wilson syndrome	Mowat Wilson Syndrome	Yes	Yes	-
3.	4 y/M	Waardenburg syndrome	Waardenburg syndrome	No	No	No
4.	2 y/M	William syndrome	William syndrome	Yes	Yes	-
5.	5 y/F	Prader Willi syndrome	Angelman syndrome	Yes	Yes	-
6.	9 y/M	?Chromosomal	5p duplication	No	No	No
7.	12 y/F	Turner syndrome	Turner syndrome	No	No	No
8.	11 d/F	Down syndrome	Down syndrome	Yes	Yes	-
9.	4 y/M	?Chromosomal	MED12 related intellectual disability	No	No	No
10.	11 y/M	Cockayne syndrome	Cockayne syndrome	No	Yes	-
11.	1 y/M	Trisomy 18	Trisomy 18	No	Yes	-
12.	4 y/F	William syndrome	William syndrome	Yes	Yes	-
13.	12 y/M	Fragile X syndrome	Fragile X syndrome	Yes	Yes	-
14.	12 y/F	William syndrome	William Syndrome	Yes	Yes	-
15.	7 y/M	Fragile X syndrome	Fragile X syndrome	Yes	Yes	-
16.	1 y/M	Cockayne syndrome	Cockayne syndrome	Yes	Yes	-
17.	8 y/M	Cockayne syndrome	Cockayne syndrome	No	Yes	-
18.	6 y/M	Cockayne syndrome	Cockayne syndrome	No	No	Yes
19.	9 y/M	William syndrome	William syndrome	Yes	Yes	-
20.	8 y/F	Mucopolysaccharidoses	MPS III	No	No	No
21.	9 y/M	Smith Magenis syndrome	Smith Magenis syndrome	Yes	Yes	-
22.	5.5y/F	Mucopolysaccharidoses	MPS III	No	Yes	-
23.	2 y/F	Mucopolysaccharidoses	I cell disease	No	Yes	-
24.	2 y/F	Wolf-Hirschhorn syndrome	Wolf-Hirschhorn syndrome	No	No	No
25.	1 y/F	I cell disease	I cell disease	Yes	Yes	-
26.	13 y/F	William syndrome	William syndrome	No	No	No
27.	8 y/M	Lipodystrophy	Berardinelli Seip syndrome	No	No	No
28.	5 y/M	Syndromic intellectual disability	Cohen syndrome	No	Yes	-
29.	3 mo/F	Di George syndrome	Di George syndrome	No	Yes	-
30.	10 mo/M	I cell disease	I cell disease	No	No	No
31.	16 y/M	Noonan syndrome	Noonan syndrome	Yes	Yes	-
32.	7 mo/M	Geleophysic dysplasia	Geleophysic dysplasia	No	No	No
33.	1 y/M	MPS	Hurler syndrome	No	Yes	-
34.	7 y/M	MPSII	Hunter syndrome	No	Yes	-
35.	5 y/M	Achondroplasia	Achondroplasia	Yes	Yes	-
36.	3 y/M	Fragile X syndrome	Fragile X syndrome	Yes	Yes	-
37.	1 y/M	I cell disease	I cell disease	No	No	No
38.	5 y/F	Apert syndrome	Apert syndrome	No	No	No
39.	4 y/F	?Chromosomal	Winter Baraitser syndrome	Yes	Yes	-
40.	2 y/F	Cornelia De Lange syndrome	Cornelia De Lange syndrome	Yes	Yes	-

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<i>No.</i>	<i>Age/Sex</i>	<i>Clinical gestalt diagnosis</i>	<i>Molecular/Cytogenetic diagnosis</i>	<i>Predicted as first diagnosis</i>	<i>Correct diagnosis in first ten diagnoses</i>	<i>Correct diagnosis after additional information</i>
41.	13 y/F	Noonan syndrome	Noonan syndrome	Yes	Yes	-
42.	5 y/F	Mandibulo acral dysostosis	Mandibular Hypoplasia, Deafness, Progeroid features and Lipodystrophy syndrome	No	Yes (Progeria)	-
43.	12 Y/F	Noonan syndrome	Noonan syndrome	Yes	Yes	-
44.	9 y/M	Apert syndrome	Apert syndrome	Yes	Yes	-
45.	18 y/M	Noonan syndrome	Noonan syndrome	Yes	Yes	-
46.	1 y/F	Jacobson syndrome	Jacobson syndrome	No	No	No
47.	9 y/F	Noonan syndrome	Noonan syndrome	Yes	Yes	-
48.	15 y/M	Prader Willi syndrome	Prader Willi syndrome	No	No	No
49.	18 mo/F	Noonan syndrome	Noonan syndrome	Yes	Yes	-
50.	10mo/M	Down syndrome	Down syndrome	Yes	Yes	-
51.	9 mo/M	Achondroplasia	Achondroplasia	Yes	Yes	-