

# **Prioritization of Exome Data by Image Analysis**

## **Supplementary Material**

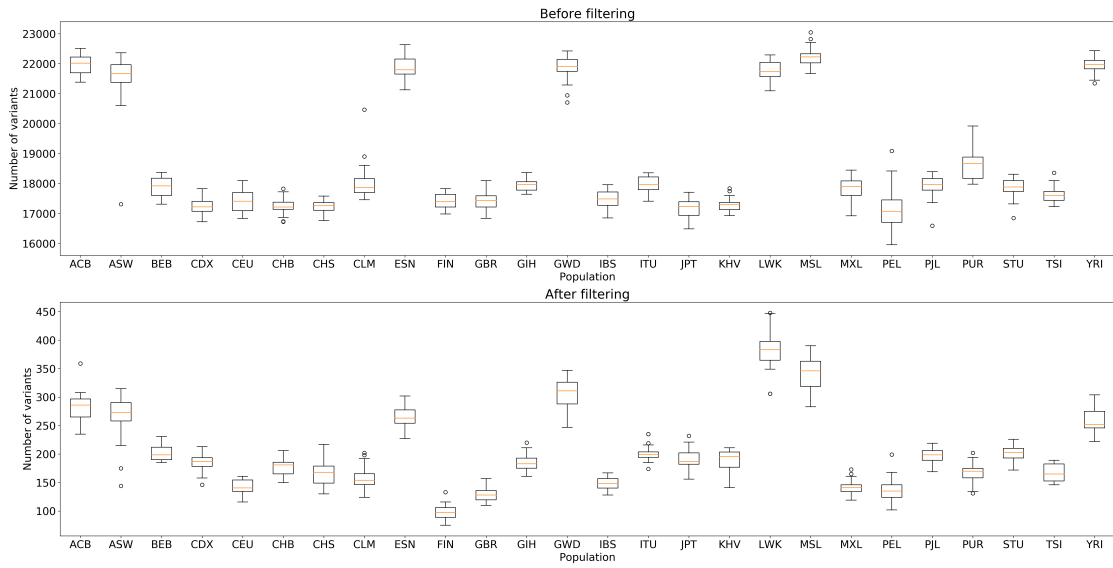
Hsieh, Tzung-Chien <sup>\*1, 2</sup>, Mensah, Martin-Atta <sup>\*2, 3</sup>, Pantel, Jean Tori<sup>2, 3</sup>, PEDIA consortium<sup>1</sup>,  
and Krawitz, Peter<sup>1</sup>

<sup>1</sup>Institute of Genomic Statistics and Bioinformatics, University of Bonn, Bonn, Germany

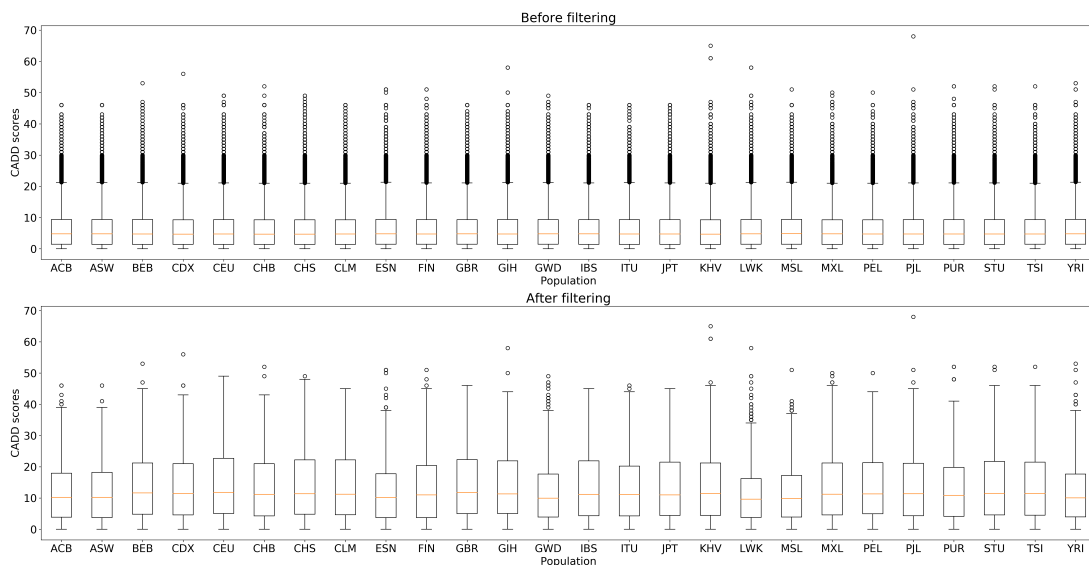
<sup>2</sup>Charit Universittsmedizin Berlin, corporate member of Freie Universitt Berlin, Humboldt-Universitt zu Berlin, and Berlin  
Institute of Health, Institute of Medical Genetics and Human Genetics, Berlin, Germany

<sup>3</sup>Berlin Institute of Health (BIH), Anna-Louisa-Karsch 2, 10178 Berlin, German

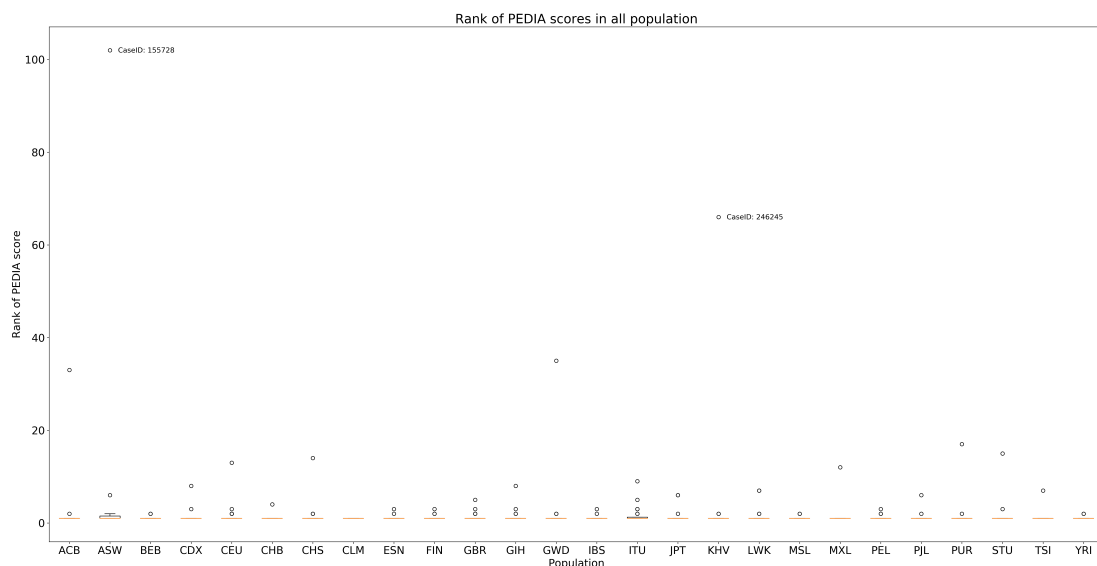
The influence of the ethnic background on the performance of the PEDIA approach was analyzed by spiking in the disease-causing mutations into the different populations of the 1000 genomes project (ACB, African Caribbeans in Barbados; ASW Americans of African Ancestry in SW USA; BEB, Bengali from Bangladesh; CDX, Chinese Dai in Xishuangbanna, China; CEU Western European Ancestry; CHB Han Chinese in Beijing, China; CHS, Southern Han Chinese; CLM, Colombians from Medellin, Colombia; ESN, Esan in Nigeria; FIN, Finnish in Finland; GBR, British in England Scotland; GIH, Gujarati Indian from Houston Texas; GWD, Gambian in Western Divisions in the Gambia; IBS, Iberian Population in Spain; ITU, Indian Telugu from the UK; JPT, Japanese in Tokyo, Japan; KHV, Kinh in Ho Chi Minh City, Vietnam; LWK, Luhya in Webuye, Kenya; MSL, Mende in Sierra Leone; MXL, Mexican Ancestry from Los Angeles USA; PEL, Peruvians from Lima, Peru; PJL, Punjabi from Lahore, Pakistan; PUR, Puerto Ricans from Puerto Rico; STU, Sri Lankan Tamil from the UK; TSI, Toscani in Italia; YRI, Yoruba in Ibadan, Nigeria).



**Supplementary Figure 1:** The number of variants that are called in reference-guided exome sequencing varies depending on the ethnic background. Filtering according to Wright, et al., results in 100 to 400 rare variants, with the smallest numbers observed in individuals from Finland and the largest in individuals from Kenya.



**Supplementary Figure 2:** The distribution of CADD scores is less dependent on the ethnic background. In all populations there is a shift in the median of the CADD scores after filtering for rare variants.



**Supplementary Figure 3:** The disease-causing mutations of the PEDIA cohort have been spiked into randomly selected samples from the 1KGP project. Shown are the results of one such simulation. The mean rank that the disease-causing gene achieves, does not depend on the ethnic background. The phenotypic and molecular data of poorly performing examples, e.g. cases 155728 and 246245 (see Supplementary Table 1 for details) yield lowest ranks in all population.

**Supplementary Table 2:** Comparison results of using different scores for gene prioritization. First column indicates the scores used in each row. The second and third columns are the Top-1 and Top-10 accuracy and confidence interval. For genes with the same PEDIA score, the highest (worst) rank was used for all of them, as this represents best the associated time for the workup. (F, Feature match score. C, CADD score. G, Gestalt score. P, Phenomizer score. B, Boqa score).

	Top-1 (%)	Top-10 (%)
<b>Photo + Exome + Feature</b>		
F,C,G,B,P	89.4 [81.4 - 97.4]	98.7 [95.7 - 100]
C,G,B,P	89.0 [80.0 - 98.0]	98.7 [95.1 - 100]
F,C,G,P	89.1 [79.9 - 98.3]	98.5 [95.1 - 100]
F,C,G,B	87.9 [78.7 - 97.1]	98.1 [94.7 - 100]
C,G,P	88.5 [79.3 - 97.7]	98.2 [93.8 - 100]
C,G,B	86.0 [75.4 - 96.6]	97.8 [95.8 - 99.8]
F,C,G	87.8 [76.4 - 99.2]	97.9 [94.1 - 100]
<b>Photo + Exome</b>		
C,G	82.6 [67.6 - 97.6]	97.5 [95.1 - 99.9]
<b>Photo + Feature</b>		
F,G,B,P	25.1 [1.7 - 48.5]	69.2 [35.8 - 100]
G,B,P	24.0 [3.0 - 45.0]	65.0 [32.4 - 97.6]
F,G,P	20.5 [1.7 - 39.3]	67.2 [33.0 - 100]
F,G,B	23.1 [1.5 - 44.7]	65.9 [36.1 - 95.7]
G,P	18.6 [2.6 - 34.6]	61.3 [24.9 - 97.7]
G,B	22.0 [4.6 - 39.4]	58.6 [31.4 - 85.8]
F,G	16.8 [0 - 34.4]	59.2 [29.8 - 88.6]
<b>Exome + Feature</b>		
F,C,B,P	74.4 [60.0 - 88.8]	93.7 [88.5 - 98.9]
C,B,P	61.4 [43.2 - 79.6]	89.0 [84.2 - 93.8]
F,C,P	74.1 [64.1 - 84.1]	93.8 [87.2 - 100]
F,C,B	65.8 [47.8 - 83.8]	91.3 [82.1 - 100]
C,P	57.9 [39.1 - 76.7]	88.7 [81.3 - 96.1]
C,B	35.5 [12.1 - 58.9]	62.8 [40.4 - 85.2]
F,C	65.1 [49.1 - 81.1]	90.9 [80.5 - 100]
<b>Photo</b>		
G	14.3 [0.3 - 28.3]	48.6 [23.4 - 73.8]
<b>Exome</b>		
C	14.1 [0 - 31.1]	44.6 [13.6 - 75.6]
<b>Feature</b>		
F,B,P	16.4 [0 - 34.2]	54.4 [25.4 - 83.4]
B,P	16.2 [0.8 - 31.6]	48.9 [21.9 - 75.9]
F,P	16.7 [0 - 36.9]	53.8 [27.0 - 80.6]
F,B	16.4 [0 - 34.4]	53.8 [24.6 - 83.0]
P	1.8 [0 - 4.6]	16.4 [0 - 33.2]
B	11.2 [0 - 28.8]	46.4 [22.0 - 70.8]
F	13.4 [0 - 29.4]	50.7 [25.5 - 75.9]

**Supplementary Table 3:** Comparison of results by using DeepGestalt and PEDIA on DeepGestalt publication test set. The PEDIA rank is the average rank.

Index	Syndrome Name	Photo Link	DeepGestalt	PEDIA
1	Alagille Syndrome	<a href="https://app.face2gene.com/dg/lmd/110/photo/345">https://app.face2gene.com/dg/lmd/110/photo/345</a>	28	1.9
2	Nijmegen Breakage Syndrome; NBS	<a href="https://app.face2gene.com/dg/lmd/347/photo/4734">https://app.face2gene.com/dg/lmd/347/photo/4734</a>	2	1.0
3	Nijmegen Breakage Syndrome; NBS	<a href="https://app.face2gene.com/dg/lmd/347/photo/4730">https://app.face2gene.com/dg/lmd/347/photo/4730</a>	2	1.0
4	Craniodiaphyseal Dysplasia; CDD	<a href="https://app.face2gene.com/dg/lmd/373/photo/1088">https://app.face2gene.com/dg/lmd/373/photo/1088</a>	1	1.0
5	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/1946/photo/5976">https://app.face2gene.com/dg/lmd/1946/photo/5976</a>	19	1.4
6	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/1946/photo/5985">https://app.face2gene.com/dg/lmd/1946/photo/5985</a>	11	1.1
7	Smith-Lemli-Opitz Syndrome; SLOS	<a href="https://app.face2gene.com/dg/lmd/1608/photo/4893">https://app.face2gene.com/dg/lmd/1608/photo/4893</a>	1	1.0
8	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/2567">https://app.face2gene.com/dg/lmd/893/photo/2567</a>	1	1.0
9	Focal Facial Dermal Dysplasia 3, Setleis Type; FFDD3	<a href="https://app.face2gene.com/dg/lmd/1570/photo/4760">https://app.face2gene.com/dg/lmd/1570/photo/4760</a>	5	1.0
10	Burn-Mckeown Syndrome; BMKS	<a href="https://app.face2gene.com/dg/lmd/2907/photo/6717">https://app.face2gene.com/dg/lmd/2907/photo/6717</a>	1	1.0
11	Burn-Mckeown Syndrome; BMKS	<a href="https://app.face2gene.com/dg/lmd/2907/photo/6716">https://app.face2gene.com/dg/lmd/2907/photo/6716</a>	1	1.0
12	Burn-Mckeown Syndrome; BMKS	<a href="https://app.face2gene.com/dg/lmd/2907/photo/6719">https://app.face2gene.com/dg/lmd/2907/photo/6719</a>	18	2.8
13	Burn-Mckeown Syndrome; BMKS	<a href="https://app.face2gene.com/dg/lmd/2907/photo/6720">https://app.face2gene.com/dg/lmd/2907/photo/6720</a>	1	1.2
14	Meier-Gorlin Syndrome 1; MGORS1	<a href="https://app.face2gene.com/dg/lmd/808/photo/2269">https://app.face2gene.com/dg/lmd/808/photo/2269</a>	30	1.2
16	Trichorhinophalangeal Syndrome, Type II; TRPS2	<a href="https://app.face2gene.com/dg/lmd/971/photo/2778">https://app.face2gene.com/dg/lmd/971/photo/2778</a>	1	1.0
17	Acrocallosal Syndrome; ACLS	<a href="https://app.face2gene.com/dg/lmd/23/photo/80">https://app.face2gene.com/dg/lmd/23/photo/80</a>	7	1.6
18	Cohen Syndrome; COH1	<a href="https://app.face2gene.com/dg/lmd/333/photo/1003">https://app.face2gene.com/dg/lmd/333/photo/1003</a>	1	1.0
19	Schwartz-Jampel Syndrome, Type 1; SJS1	<a href="https://app.face2gene.com/dg/lmd/1548/photo/4685">https://app.face2gene.com/dg/lmd/1548/photo/4685</a>	9	1.1
20	Treacher Collins Syndrome 1; TCS1	<a href="https://app.face2gene.com/dg/lmd/1720/photo/5245">https://app.face2gene.com/dg/lmd/1720/photo/5245</a>	1	1.0
21	Trichorhinophalangeal Syndrome	<a href="https://app.face2gene.com/dg/lmd/1730/photo/5290">https://app.face2gene.com/dg/lmd/1730/photo/5290</a>	1	1.0
22	Velocardiofacial Syndrome	<a href="https://app.face2gene.com/dg/lmd/1762/photo/8285">https://app.face2gene.com/dg/lmd/1762/photo/8285</a>	3	1.0
23	Smith-Lemli-Opitz Syndrome; SLOS	<a href="https://app.face2gene.com/dg/lmd/1608/photo/8342">https://app.face2gene.com/dg/lmd/1608/photo/8342</a>	1	1.0
24	Marfan Syndrome; MFS	<a href="https://app.face2gene.com/dg/lmd/1057/photo/8255">https://app.face2gene.com/dg/lmd/1057/photo/8255</a>	1	1.0
26	Mowat-Wilson Syndrome; MOWS	<a href="https://app.face2gene.com/dg/lmd/4211/photo/13183">https://app.face2gene.com/dg/lmd/4211/photo/13183</a>	1	1.0
27	Mowat-Wilson Syndrome; MOWS	<a href="https://app.face2gene.com/dg/lmd/4211/photo/13743">https://app.face2gene.com/dg/lmd/4211/photo/13743</a>	1	1.0
28	Mowat-Wilson Syndrome; MOWS	<a href="https://app.face2gene.com/dg/lmd/4211/photo/13744">https://app.face2gene.com/dg/lmd/4211/photo/13744</a>	1	1.0
29	3MC Syndrome 3; 3MC3	<a href="https://app.face2gene.com/dg/lmd/1048/photo/14730">https://app.face2gene.com/dg/lmd/1048/photo/14730</a>	2	1.0
30	Sotos Syndrome 2; SOTOS2	<a href="https://app.face2gene.com/dg/lmd/1617/photo/15149">https://app.face2gene.com/dg/lmd/1617/photo/15149</a>	1	1.1
31	Sotos Syndrome 2; SOTOS2	<a href="https://app.face2gene.com/dg/lmd/1617/photo/15152">https://app.face2gene.com/dg/lmd/1617/photo/15152</a>	1	1.0

32	Treacher Collins Syndrome 1; TCS1	<a href="https://app.face2gene.com/dg/lmd/1720/photo/5227">https://app.face2gene.com/dg/lmd/1720/photo/5227</a>	1	1.0
33	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/958">https://app.face2gene.com/dg/lmd/328/photo/958</a>	1	1.0
35	Mowat-Wilson Syndrome; MOWS	<a href="https://app.face2gene.com/dg/lmd/4211/photo/16768">https://app.face2gene.com/dg/lmd/4211/photo/16768</a>	1	1.0
36	Donnai-Barrow Syndrome	<a href="https://app.face2gene.com/dg/lmd/3473/photo/16950">https://app.face2gene.com/dg/lmd/3473/photo/16950</a>	49	1.2
37	Floating-Harbor Syndrome; FLHS	<a href="https://app.face2gene.com/dg/lmd/596/photo/17148">https://app.face2gene.com/dg/lmd/596/photo/17148</a>	17	1.3
38	Pitt-Hopkins Syndrome; PTHS	<a href="https://app.face2gene.com/dg/lmd/8892/photo/17151">https://app.face2gene.com/dg/lmd/8892/photo/17151</a>	1	1.0
39	Marshall-Smith Syndrome; MRSHSS	<a href="https://app.face2gene.com/dg/lmd/7513/photo/17204">https://app.face2gene.com/dg/lmd/7513/photo/17204</a>	1	1.0
40	3MC Syndrome 3; 3MC3	<a href="https://app.face2gene.com/dg/lmd/1048/photo/17445">https://app.face2gene.com/dg/lmd/1048/photo/17445</a>	110	1.7
42	Waardenburg Syndrome, Type 1; WS1	<a href="https://app.face2gene.com/dg/lmd/1781/photo/5438">https://app.face2gene.com/dg/lmd/1781/photo/5438</a>	37	1.0
44	Sotos Syndrome 2; SOTOS2	<a href="https://app.face2gene.com/dg/lmd/1617/photo/4945">https://app.face2gene.com/dg/lmd/1617/photo/4945</a>	1	1.0
45	Pitt-Hopkins Syndrome; PTHS	<a href="https://app.face2gene.com/dg/lmd/8892/photo/17999">https://app.face2gene.com/dg/lmd/8892/photo/17999</a>	1	1.0
46	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/18077">https://app.face2gene.com/dg/lmd/893/photo/18077</a>	1	1.0
47	Waardenburg Syndrome, Type 1; WS1	<a href="https://app.face2gene.com/dg/lmd/1781/photo/8786">https://app.face2gene.com/dg/lmd/1781/photo/8786</a>	1	1.0
48	Waardenburg Syndrome, Type 1; WS1	<a href="https://app.face2gene.com/dg/lmd/1781/photo/8794">https://app.face2gene.com/dg/lmd/1781/photo/8794</a>	25	1.0
49	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/9086">https://app.face2gene.com/dg/lmd/3562/photo/9086</a>	1	1.0
51	Seckel Syndrome	<a href="https://app.face2gene.com/dg/lmd/739/photo/9429">https://app.face2gene.com/dg/lmd/739/photo/9429</a>	2	1.0
52	Rothmund-Thomson Syndrome; RTS	<a href="https://app.face2gene.com/dg/lmd/1487/photo/9782">https://app.face2gene.com/dg/lmd/1487/photo/9782</a>	66	1.5
54	Coffin-Siris Syndrome1; CSS1	<a href="https://app.face2gene.com/dg/lmd/329/photo/18535">https://app.face2gene.com/dg/lmd/329/photo/18535</a>	15	1.0
55	Bohring-Opitz Syndrome; BOPS	<a href="https://app.face2gene.com/dg/lmd/4337/photo/18830">https://app.face2gene.com/dg/lmd/4337/photo/18830</a>	1	1.0
56	SHORT syndrome	<a href="https://app.face2gene.com/dg/lmd/1574/photo/4787">https://app.face2gene.com/dg/lmd/1574/photo/4787</a>	1	1.0
57	Schwartz-Jampel Syndrome, Type 1; SJS1	<a href="https://app.face2gene.com/dg/lmd/1548/photo/4686">https://app.face2gene.com/dg/lmd/1548/photo/4686</a>	4	1.0
58	Hajdu-Cheney Syndrome; HJCYS	<a href="https://app.face2gene.com/dg/lmd/722/photo/2067">https://app.face2gene.com/dg/lmd/722/photo/2067</a>	1	1.0
59	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/967">https://app.face2gene.com/dg/lmd/328/photo/967</a>	1	1.0
60	Baraitser-Winter Syndrome 1; BRWS1	<a href="https://app.face2gene.com/dg/lmd/149/photo/430">https://app.face2gene.com/dg/lmd/149/photo/430</a>	2	1.0
61	Beckwith-Wiedemann Syndrome; BWS	<a href="https://app.face2gene.com/dg/lmd/173/photo/480">https://app.face2gene.com/dg/lmd/173/photo/480</a>	3	1.0
62	Baraitser-Winter Syndrome 1; BRWS1	<a href="https://app.face2gene.com/dg/lmd/149/photo/432">https://app.face2gene.com/dg/lmd/149/photo/432</a>	2	1.0
66	Sotos Syndrome 2; SOTOS2	<a href="https://app.face2gene.com/dg/lmd/1617/photo/4944">https://app.face2gene.com/dg/lmd/1617/photo/4944</a>	1	1.0
67	Cockayne Syndrome	<a href="https://app.face2gene.com/dg/lmd/326/photo/946">https://app.face2gene.com/dg/lmd/326/photo/946</a>	1	1.0
68	Acromicric Dysplasia	<a href="https://app.face2gene.com/dg/lmd/30/photo/145">https://app.face2gene.com/dg/lmd/30/photo/145</a>	24	1.0
70	Ectodermal Dysplasia 1, Hypohidrotic, X-Linked; XHED	<a href="https://app.face2gene.com/dg/lmd/508/photo/1407">https://app.face2gene.com/dg/lmd/508/photo/1407</a>	1	1.0
71	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/960">https://app.face2gene.com/dg/lmd/328/photo/960</a>	6	1.0
72	Mucopolipidosis II Alpha/beta	<a href="https://app.face2gene.com/dg/lmd/838/photo/2386">https://app.face2gene.com/dg/lmd/838/photo/2386</a>	5	1.0

73	Beckwith-Wiedemann Syndrome; BWS	<a href="https://app.face2gene.com/dg/lmd/173/photo/484">https://app.face2gene.com/dg/lmd/173/photo/484</a>	6	1.0
74	Craniodiaphyseal Dysplasia; CDD	<a href="https://app.face2gene.com/dg/lmd/373/photo/1093">https://app.face2gene.com/dg/lmd/373/photo/1093</a>	2	1.0
75	Cockayne Syndrome	<a href="https://app.face2gene.com/dg/lmd/326/photo/948">https://app.face2gene.com/dg/lmd/326/photo/948</a>	3	1.1
76	Floating-Harbor Syndrome; FLHS	<a href="https://app.face2gene.com/dg/lmd/596/photo/1761">https://app.face2gene.com/dg/lmd/596/photo/1761</a>	1	1.0
77	Rubinstein-Taybi Syndrome 1; RSTS1	<a href="https://app.face2gene.com/dg/lmd/1490/photo/4474">https://app.face2gene.com/dg/lmd/1490/photo/4474</a>	1	1.0
78	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/970">https://app.face2gene.com/dg/lmd/328/photo/970</a>	1	1.0
79	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/971">https://app.face2gene.com/dg/lmd/328/photo/971</a>	1	1.0
80	Apert Syndrome	<a href="https://app.face2gene.com/dg/lmd/99/photo/326">https://app.face2gene.com/dg/lmd/99/photo/326</a>	1	1.0
81	Branchiooculofacial Syndrome; BOFS	<a href="https://app.face2gene.com/dg/lmd/633/photo/8345">https://app.face2gene.com/dg/lmd/633/photo/8345</a>	1	1.0
82	Smith-Lemli-Opitz Syndrome; SLOS	<a href="https://app.face2gene.com/dg/lmd/1608/photo/12105">https://app.face2gene.com/dg/lmd/1608/photo/12105</a>	1	1.0
83	Smith-Lemli-Opitz Syndrome; SLOS	<a href="https://app.face2gene.com/dg/lmd/1608/photo/12105">https://app.face2gene.com/dg/lmd/1608/photo/12105</a>	1	1.0
84	Coffin-Siris Syndrome1; CSS1	<a href="https://app.face2gene.com/dg/lmd/329/photo/12535">https://app.face2gene.com/dg/lmd/329/photo/12535</a>	3	1.0
85	Coffin-Siris Syndrome1; CSS1	<a href="https://app.face2gene.com/dg/lmd/329/photo/12535">https://app.face2gene.com/dg/lmd/329/photo/12535</a>	2	1.0
86	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/13114">https://app.face2gene.com/dg/lmd/893/photo/13114</a>	1	1.0
87	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/13116">https://app.face2gene.com/dg/lmd/893/photo/13116</a>	1	1.0
88	Mowat-Wilson Syndrome; MOWS	<a href="https://app.face2gene.com/dg/lmd/4211/photo/13734">https://app.face2gene.com/dg/lmd/4211/photo/13734</a>	1	1.0
89	Mowat-Wilson Syndrome; MOWS	<a href="https://app.face2gene.com/dg/lmd/4211/photo/13736">https://app.face2gene.com/dg/lmd/4211/photo/13736</a>	58	1.5
90	Mowat-Wilson Syndrome; MOWS	<a href="https://app.face2gene.com/dg/lmd/4211/photo/13742">https://app.face2gene.com/dg/lmd/4211/photo/13742</a>	1	1.0
91	Borjeson-Forssman-Lehmann Syndrome; BFLS	<a href="https://app.face2gene.com/dg/lmd/216/photo/14759">https://app.face2gene.com/dg/lmd/216/photo/14759</a>	56	2.8
92	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/14969">https://app.face2gene.com/dg/lmd/3562/photo/14969</a>	4	1.2
95	Opitz GBBB Syndrome, Type II; GBBB2	<a href="https://app.face2gene.com/dg/lmd/640/photo/15940">https://app.face2gene.com/dg/lmd/640/photo/15940</a>	9	1.0
98	Sotos Syndrome 2; SOTOS2	<a href="https://app.face2gene.com/dg/lmd/1617/photo/17023">https://app.face2gene.com/dg/lmd/1617/photo/17023</a>	1	1.0
99	Sotos Syndrome 2; SOTOS2	<a href="https://app.face2gene.com/dg/lmd/1617/photo/17024">https://app.face2gene.com/dg/lmd/1617/photo/17024</a>	10	1.3
100	CHARGE Syndrome	<a href="https://app.face2gene.com/dg/lmd/301/photo/17094">https://app.face2gene.com/dg/lmd/301/photo/17094</a>	2	1.0
101	Floating-Harbor Syndrome; FLHS	<a href="https://app.face2gene.com/dg/lmd/596/photo/17124">https://app.face2gene.com/dg/lmd/596/photo/17124</a>	1	1.0
103	Craniometaphyseal Dysplasia	<a href="https://app.face2gene.com/dg/lmd/371/photo/7667">https://app.face2gene.com/dg/lmd/371/photo/7667</a>	22	1.0
105	Pitt-Hopkins Syndrome; PTHS	<a href="https://app.face2gene.com/dg/lmd/8892/photo/18000">https://app.face2gene.com/dg/lmd/8892/photo/18000</a>	1	1.0
106	Baraitser-Winter Syndrome 1; BRWS1	<a href="https://app.face2gene.com/dg/lmd/149/photo/10154">https://app.face2gene.com/dg/lmd/149/photo/10154</a>	10	1.4
107	Baraitser-Winter Syndrome 1; BRWS1	<a href="https://app.face2gene.com/dg/lmd/149/photo/10156">https://app.face2gene.com/dg/lmd/149/photo/10156</a>	41	1.8
108	Baraitser-Winter Syndrome 1; BRWS1	<a href="https://app.face2gene.com/dg/lmd/149/photo/10156">https://app.face2gene.com/dg/lmd/149/photo/10156</a>	41	1.8
109	3MC Syndrome 3; 3MC3	<a href="https://app.face2gene.com/dg/lmd/1048/photo/10305">https://app.face2gene.com/dg/lmd/1048/photo/10305</a>	2	1.0
111	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/18744">https://app.face2gene.com/dg/lmd/3562/photo/18744</a>	1	1.0

112	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/18745">https://app.face2gene.com/dg/lmd/3562/photo/18745</a>	4	1.0
113	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/18747">https://app.face2gene.com/dg/lmd/3562/photo/18747</a>	1	1.5
114	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/18749">https://app.face2gene.com/dg/lmd/3562/photo/18749</a>	1	1.1
118	Crouzon Syndrome	<a href="https://app.face2gene.com/dg/lmd/383/photo/1133">https://app.face2gene.com/dg/lmd/383/photo/1133</a>	1	1.0
120	Beckwith-Wiedemann Syndrome; BWS	<a href="https://app.face2gene.com/dg/lmd/173/photo/41779">https://app.face2gene.com/dg/lmd/173/photo/41779</a>	1	1.0
121	Otopalatodigital Syndrome	<a href="https://app.face2gene.com/dg/lmd/1266/photo/41780">https://app.face2gene.com/dg/lmd/1266/photo/41780</a>	1	1.0
122	Waardenburg Syndrome, Type 1; WS1	<a href="https://app.face2gene.com/dg/lmd/1781/photo/41781">https://app.face2gene.com/dg/lmd/1781/photo/41781</a>	1	1.0
123	Rubinstein-Taybi Syndrome	<a href="https://app.face2gene.com/dg/lmd/1490/photo/41785">https://app.face2gene.com/dg/lmd/1490/photo/41785</a>	1	1.0
124	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41799">https://app.face2gene.com/dg/lmd/1720/photo/41799</a>	1	1.0
125	Cornelia De Lange Syndrome	<a href="https://app.face2gene.com/dg/lmd/423/photo/41811">https://app.face2gene.com/dg/lmd/423/photo/41811</a>	1	1.5
126	Cornelia De Lange Syndrome	<a href="https://app.face2gene.com/dg/lmd/423/photo/41812">https://app.face2gene.com/dg/lmd/423/photo/41812</a>	1	1.5
127	Cornelia De Lange Syndrome	<a href="https://app.face2gene.com/dg/lmd/423/photo/41813">https://app.face2gene.com/dg/lmd/423/photo/41813</a>	1	1.5
128	Cornelia De Lange Syndrome	<a href="https://app.face2gene.com/dg/lmd/423/photo/41814">https://app.face2gene.com/dg/lmd/423/photo/41814</a>	1	2.0
129	Sotos Syndrome	<a href="https://app.face2gene.com/dg/lmd/1617/photo/41817">https://app.face2gene.com/dg/lmd/1617/photo/41817</a>	1	1.1
130	Ectodermal Dysplasia 1, Hypohidrotic, X-Linked; XHED	<a href="https://app.face2gene.com/dg/lmd/508/photo/41823">https://app.face2gene.com/dg/lmd/508/photo/41823</a>	2	1.0
134	Hutchinson-Gilford Progeria Syndrome; HGPS	<a href="https://app.face2gene.com/dg/lmd/1396/photo/41849">https://app.face2gene.com/dg/lmd/1396/photo/41849</a>	1	1.0
136	Hurler Syndrome	<a href="https://app.face2gene.com/dg/lmd/13858/photo/41873">https://app.face2gene.com/dg/lmd/13858/photo/41873</a>	1	1.1
137	Hurler Syndrome	<a href="https://app.face2gene.com/dg/lmd/13858/photo/41874">https://app.face2gene.com/dg/lmd/13858/photo/41874</a>	1	1.1
138	Mucopolysaccharidosis, Type II; MPS2	<a href="https://app.face2gene.com/dg/lmd/801/photo/41875">https://app.face2gene.com/dg/lmd/801/photo/41875</a>	1	1.1
139	Mucopolysaccharidosis, Type II; MPS2	<a href="https://app.face2gene.com/dg/lmd/801/photo/41876">https://app.face2gene.com/dg/lmd/801/photo/41876</a>	1	1.0
140	Mucopolysaccharidosis, Type II; MPS2	<a href="https://app.face2gene.com/dg/lmd/801/photo/41878">https://app.face2gene.com/dg/lmd/801/photo/41878</a>	1	1.1
141	Mucopolysaccharidosis, Type II; MPS2	<a href="https://app.face2gene.com/dg/lmd/801/photo/41879">https://app.face2gene.com/dg/lmd/801/photo/41879</a>	1	1.1
142	Mucopolysaccharidosis II Alpha/beta	<a href="https://app.face2gene.com/dg/lmd/838/photo/41883">https://app.face2gene.com/dg/lmd/838/photo/41883</a>	1	1.0
143	Cleidocranial Dysplasia; CCD	<a href="https://app.face2gene.com/dg/lmd/13331/photo/41890">https://app.face2gene.com/dg/lmd/13331/photo/41890</a>	5	1.4
144	Cleidocranial Dysplasia; CCD	<a href="https://app.face2gene.com/dg/lmd/13331/photo/41891">https://app.face2gene.com/dg/lmd/13331/photo/41891</a>	1	1.0
145	Seckel Syndrome	<a href="https://app.face2gene.com/dg/lmd/739/photo/41904">https://app.face2gene.com/dg/lmd/739/photo/41904</a>	12	1.1
147	Crouzon Syndrome	<a href="https://app.face2gene.com/dg/lmd/383/photo/41914">https://app.face2gene.com/dg/lmd/383/photo/41914</a>	2	1.0
153	Sotos Syndrome	<a href="https://app.face2gene.com/dg/lmd/1617/photo/41940">https://app.face2gene.com/dg/lmd/1617/photo/41940</a>	1	1.0
154	Sotos Syndrome	<a href="https://app.face2gene.com/dg/lmd/1617/photo/41941">https://app.face2gene.com/dg/lmd/1617/photo/41941</a>	1	1.2
157	Cleidocranial Dysplasia; CCD	<a href="https://app.face2gene.com/dg/lmd/13331/photo/41947">https://app.face2gene.com/dg/lmd/13331/photo/41947</a>	1	1.0
158	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41956">https://app.face2gene.com/dg/lmd/1720/photo/41956</a>	4	1.0
159	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41958">https://app.face2gene.com/dg/lmd/1720/photo/41958</a>	1	1.0



163	Fragile X Mental Retardation Syndrome	<a href="https://app.face2gene.com/dg/lmd/13243/photo/41973">https://app.face2gene.com/dg/lmd/13243/photo/41973</a>	1	2.1
165	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/15947">https://app.face2gene.com/dg/lmd/3562/photo/15947</a>	1	1.2
166	Pierpont Syndrome; PRPTS	<a href="https://app.face2gene.com/dg/lmd/4198/photo/41638">https://app.face2gene.com/dg/lmd/4198/photo/41638</a>	3	1.0
167	Pierpont Syndrome; PRPTS	<a href="https://app.face2gene.com/dg/lmd/4198/photo/41640">https://app.face2gene.com/dg/lmd/4198/photo/41640</a>	1	1.0
168	SHORT syndrome	<a href="https://app.face2gene.com/dg/lmd/1574/photo/4780">https://app.face2gene.com/dg/lmd/1574/photo/4780</a>	1	1.0
169	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/2566">https://app.face2gene.com/dg/lmd/893/photo/2566</a>	1	1.1
170	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/2560">https://app.face2gene.com/dg/lmd/893/photo/2560</a>	1	1.0
171	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/2562">https://app.face2gene.com/dg/lmd/893/photo/2562</a>	8	1.8
172	Otopalatodigital Syndrome	<a href="https://app.face2gene.com/dg/lmd/1266/photo/3719">https://app.face2gene.com/dg/lmd/1266/photo/3719</a>	1	1.0
173	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/977">https://app.face2gene.com/dg/lmd/328/photo/977</a>	1	1.0
174	Ohdo Syndrome, SBBYS Variant; SBBYSS	<a href="https://app.face2gene.com/dg/lmd/1255/photo/8343">https://app.face2gene.com/dg/lmd/1255/photo/8343</a>	1	1.1
175	Rubinstein-Taybi Syndrome 1; RSTS1	<a href="https://app.face2gene.com/dg/lmd/358/photo/11625">https://app.face2gene.com/dg/lmd/358/photo/11625</a>	2	1.1
176	Auriculocondylar Syndrome 1; ARCND1	<a href="https://app.face2gene.com/dg/lmd/4192/photo/12354">https://app.face2gene.com/dg/lmd/4192/photo/12354</a>	1	1.0
177	Coffin-Siris Syndrome1; CSS1	<a href="https://app.face2gene.com/dg/lmd/329/photo/12530">https://app.face2gene.com/dg/lmd/329/photo/12530</a>	1	1.0
178	Urofacial Syndrome 1; UFS1	<a href="https://app.face2gene.com/dg/lmd/1236/photo/13084">https://app.face2gene.com/dg/lmd/1236/photo/13084</a>	3	1.0
179	3MC Syndrome 3; 3MC3	<a href="https://app.face2gene.com/dg/lmd/1048/photo/13477">https://app.face2gene.com/dg/lmd/1048/photo/13477</a>	1	1.0
180	Smith-Lemli-Opitz Syndrome; SLOS	<a href="https://app.face2gene.com/dg/lmd/1608/photo/14313">https://app.face2gene.com/dg/lmd/1608/photo/14313</a>	1	1.0
181	3MC Syndrome 3; 3MC3	<a href="https://app.face2gene.com/dg/lmd/1048/photo/14727">https://app.face2gene.com/dg/lmd/1048/photo/14727</a>	1	1.0
183	Lig4 Syndrome	<a href="https://app.face2gene.com/dg/lmd/4654/photo/17338">https://app.face2gene.com/dg/lmd/4654/photo/17338</a>	16	1.0
184	Cranio metaphyseal Dysplasia	<a href="https://app.face2gene.com/dg/lmd/371/photo/1078">https://app.face2gene.com/dg/lmd/371/photo/1078</a>	1	1.0
188	Craniofrontonasal Syndrome; CFNS	<a href="https://app.face2gene.com/dg/lmd/376/photo/1113">https://app.face2gene.com/dg/lmd/376/photo/1113</a>	1	1.7
190	Floating-Harbor Syndrome; FLHS	<a href="https://app.face2gene.com/dg/lmd/596/photo/1756">https://app.face2gene.com/dg/lmd/596/photo/1756</a>	1	1.0
191	Rubinstein-Taybi Syndrome 1; RSTS1	<a href="https://app.face2gene.com/dg/lmd/1490/photo/4488">https://app.face2gene.com/dg/lmd/1490/photo/4488</a>	1	1.0
192	Coffin-Siris Syndrome1; CSS1	<a href="https://app.face2gene.com/dg/lmd/329/photo/981">https://app.face2gene.com/dg/lmd/329/photo/981</a>	1	1.0
193	Rubinstein-Taybi Syndrome 1; RSTS1	<a href="https://app.face2gene.com/dg/lmd/1490/photo/4489">https://app.face2gene.com/dg/lmd/1490/photo/4489</a>	1	1.1
194	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/1229/photo/3585">https://app.face2gene.com/dg/lmd/1229/photo/3585</a>	1	1.1
195	Floating-Harbor Syndrome; FLHS	<a href="https://app.face2gene.com/dg/lmd/596/photo/8231">https://app.face2gene.com/dg/lmd/596/photo/8231</a>	1	1.0
196	Smith-Lemli-Opitz Syndrome; SLOS	<a href="https://app.face2gene.com/dg/lmd/1608/photo/12105">https://app.face2gene.com/dg/lmd/1608/photo/12105</a>	1	1.0
197	Smith-Lemli-Opitz Syndrome; SLOS	<a href="https://app.face2gene.com/dg/lmd/1608/photo/12105">https://app.face2gene.com/dg/lmd/1608/photo/12105</a>	2	1.0
198	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/13113">https://app.face2gene.com/dg/lmd/893/photo/13113</a>	1	1.0
199	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/13113">https://app.face2gene.com/dg/lmd/893/photo/13113</a>	1	1.1
200	Sotos Syndrome 2; SOTOS2	<a href="https://app.face2gene.com/dg/lmd/1617/photo/15150">https://app.face2gene.com/dg/lmd/1617/photo/15150</a>	1	1.1
203	Sotos Syndrome 2; SOTOS2	<a href="https://app.face2gene.com/dg/lmd/1617/photo/17025">https://app.face2gene.com/dg/lmd/1617/photo/17025</a>	1	1.1
205	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/18745">https://app.face2gene.com/dg/lmd/3562/photo/18745</a>	1	1.3

206	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/18745">https://app.face2gene.com/dg/lmd/3562/photo/18745</a>	1	1.1
207	Birk-Barel Mental Retardation Dysmorphism Syndrome	<a href="https://app.face2gene.com/dg/lmd/5569/photo/19317">https://app.face2gene.com/dg/lmd/5569/photo/19317</a>	12	1.4
209	Focal Dermal Hypoplasia; FDH	<a href="https://app.face2gene.com/dg/lmd/13389/photo/41764">https://app.face2gene.com/dg/lmd/13389/photo/41764</a>	1	1.0
214	Cornelia De Lange Syndrome	<a href="https://app.face2gene.com/dg/lmd/423/photo/41808">https://app.face2gene.com/dg/lmd/423/photo/41808</a>	1	1.0
215	Cornelia De Lange Syndrome	<a href="https://app.face2gene.com/dg/lmd/423/photo/41810">https://app.face2gene.com/dg/lmd/423/photo/41810</a>	1	1.0
216	Sotos Syndrome	<a href="https://app.face2gene.com/dg/lmd/1617/photo/41818">https://app.face2gene.com/dg/lmd/1617/photo/41818</a>	1	1.2
218	Focal Dermal Hypoplasia; FDH	<a href="https://app.face2gene.com/dg/lmd/13389/photo/41822">https://app.face2gene.com/dg/lmd/13389/photo/41822</a>	6	1.0
219	Mucopolysaccharidosis, Type II; MPS2	<a href="https://app.face2gene.com/dg/lmd/801/photo/41824">https://app.face2gene.com/dg/lmd/801/photo/41824</a>	3	1.2
221	Crouzon Syndrome	<a href="https://app.face2gene.com/dg/lmd/383/photo/41842">https://app.face2gene.com/dg/lmd/383/photo/41842</a>	1	1.0
222	Rubinstein-Taybi Syndrome	<a href="https://app.face2gene.com/dg/lmd/1490/photo/41844">https://app.face2gene.com/dg/lmd/1490/photo/41844</a>	1	1.0
223	Hutchinson-Gilford Progeria Syndrome; HGPS	<a href="https://app.face2gene.com/dg/lmd/1396/photo/41845">https://app.face2gene.com/dg/lmd/1396/photo/41845</a>	1	1.0
224	Hutchinson-Gilford Progeria Syndrome; HGPS	<a href="https://app.face2gene.com/dg/lmd/1396/photo/41846">https://app.face2gene.com/dg/lmd/1396/photo/41846</a>	1	1.0
225	Hutchinson-Gilford Progeria Syndrome; HGPS	<a href="https://app.face2gene.com/dg/lmd/1396/photo/41847">https://app.face2gene.com/dg/lmd/1396/photo/41847</a>	1	1.0
226	Hutchinson-Gilford Progeria Syndrome; HGPS	<a href="https://app.face2gene.com/dg/lmd/1396/photo/41848">https://app.face2gene.com/dg/lmd/1396/photo/41848</a>	1	1.0
227	Hutchinson-Gilford Progeria Syndrome; HGPS	<a href="https://app.face2gene.com/dg/lmd/1396/photo/41850">https://app.face2gene.com/dg/lmd/1396/photo/41850</a>	1	1.0
228	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/1229/photo/41851">https://app.face2gene.com/dg/lmd/1229/photo/41851</a>	1	1.0
231	Rubinstein-Taybi Syndrome	<a href="https://app.face2gene.com/dg/lmd/1490/photo/41859">https://app.face2gene.com/dg/lmd/1490/photo/41859</a>	1	1.0
232	Achondroplasia; ACH	<a href="https://app.face2gene.com/dg/lmd/17/photo/41871">https://app.face2gene.com/dg/lmd/17/photo/41871</a>	9	1.0
233	Hurler Syndrome	<a href="https://app.face2gene.com/dg/lmd/13858/photo/41872">https://app.face2gene.com/dg/lmd/13858/photo/41872</a>	1	1.4
234	Cleidocranial Dysplasia; CCD	<a href="https://app.face2gene.com/dg/lmd/13331/photo/41899">https://app.face2gene.com/dg/lmd/13331/photo/41899</a>	1	1.0
236	Saethre-Chotzen Syndrome; SCS	<a href="https://app.face2gene.com/dg/lmd/1504/photo/41909">https://app.face2gene.com/dg/lmd/1504/photo/41909</a>	1	1.0
237	Cleidocranial Dysplasia; CCD	<a href="https://app.face2gene.com/dg/lmd/13331/photo/41948">https://app.face2gene.com/dg/lmd/13331/photo/41948</a>	7	2.5
238	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41951">https://app.face2gene.com/dg/lmd/1720/photo/41951</a>	1	1.0
239	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41952">https://app.face2gene.com/dg/lmd/1720/photo/41952</a>	1	1.0
240	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41954">https://app.face2gene.com/dg/lmd/1720/photo/41954</a>	1	1.0
241	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41959">https://app.face2gene.com/dg/lmd/1720/photo/41959</a>	1	1.0
242	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41960">https://app.face2gene.com/dg/lmd/1720/photo/41960</a>	1	1.0
243	Fragile X Mental Retardation Syndrome	<a href="https://app.face2gene.com/dg/lmd/13243/photo/41982">https://app.face2gene.com/dg/lmd/13243/photo/41982</a>	1	12.3
244	Pitt-Hopkins Syndrome; PTHS	<a href="https://app.face2gene.com/dg/lmd/8892/photo/18003">https://app.face2gene.com/dg/lmd/8892/photo/18003</a>	1	1.0

245	Nijmegen Breakage Syndrome; NBS	<a href="https://app.face2gene.com/dg/lmd/347/photo/4723">https://app.face2gene.com/dg/lmd/347/photo/4723</a>	1	1.0
246	Nijmegen Breakage Syndrome; NBS	<a href="https://app.face2gene.com/dg/lmd/347/photo/4736">https://app.face2gene.com/dg/lmd/347/photo/4736</a>	13	1.0
247	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/1946/photo/5983">https://app.face2gene.com/dg/lmd/1946/photo/5983</a>	2	1.0
248	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/2565">https://app.face2gene.com/dg/lmd/893/photo/2565</a>	1	1.0
249	Craniodiaphyseal Dysplasia; CDD	<a href="https://app.face2gene.com/dg/lmd/373/photo/1091">https://app.face2gene.com/dg/lmd/373/photo/1091</a>	1	1.0
250	Ectodermal Dysplasia 1, Hypohidrotic, X-Linked; XHED	<a href="https://app.face2gene.com/dg/lmd/508/photo/1400">https://app.face2gene.com/dg/lmd/508/photo/1400</a>	1	1.0
251	Mowat-Wilson Syndrome; MOWS	<a href="https://app.face2gene.com/dg/lmd/4211/photo/13735">https://app.face2gene.com/dg/lmd/4211/photo/13735</a>	1	1.0
252	3MC Syndrome 3; 3MC3	<a href="https://app.face2gene.com/dg/lmd/1048/photo/13467">https://app.face2gene.com/dg/lmd/1048/photo/13467</a>	1	1.0
253	Auriculocondylar Syndrome 1; ARCND1	<a href="https://app.face2gene.com/dg/lmd/4192/photo/14125">https://app.face2gene.com/dg/lmd/4192/photo/14125</a>	2	1.0
254	Nicolaides-Baraitser Syndrome; NCBRS	<a href="https://app.face2gene.com/dg/lmd/3394/photo/14255">https://app.face2gene.com/dg/lmd/3394/photo/14255</a>	1	1.0
255	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/14454">https://app.face2gene.com/dg/lmd/3562/photo/14454</a>	1	1.1
256	Nicolaides-Baraitser Syndrome; NCBRS	<a href="https://app.face2gene.com/dg/lmd/3394/photo/14700">https://app.face2gene.com/dg/lmd/3394/photo/14700</a>	1	1.0
257	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/1229/photo/3581">https://app.face2gene.com/dg/lmd/1229/photo/3581</a>	1	1.0
258	KBG Syndrome; KBGS	<a href="https://app.face2gene.com/dg/lmd/912/photo/15604">https://app.face2gene.com/dg/lmd/912/photo/15604</a>	1	1.0
261	Beckwith-Wiedemann Syndrome; BWS	<a href="https://app.face2gene.com/dg/lmd/173/photo/481">https://app.face2gene.com/dg/lmd/173/photo/481</a>	6	1.0
262	Craniofrontonasal Syndrome; CFNS	<a href="https://app.face2gene.com/dg/lmd/376/photo/1115">https://app.face2gene.com/dg/lmd/376/photo/1115</a>	1	1.0
263	Floating-Harbor Syndrome; FLHS	<a href="https://app.face2gene.com/dg/lmd/596/photo/1760">https://app.face2gene.com/dg/lmd/596/photo/1760</a>	1	1.0
264	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/974">https://app.face2gene.com/dg/lmd/328/photo/974</a>	1	1.0
265	Sotos Syndrome 2; SOTOS2	<a href="https://app.face2gene.com/dg/lmd/1617/photo/15146">https://app.face2gene.com/dg/lmd/1617/photo/15146</a>	1	1.0
266	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/15950">https://app.face2gene.com/dg/lmd/3562/photo/15950</a>	1	2.7
268	Lateral Meningocele Syndrome; LMNS	<a href="https://app.face2gene.com/dg/lmd/983/photo/10028">https://app.face2gene.com/dg/lmd/983/photo/10028</a>	3	1.0
269	KBG Syndrome; KBGS	<a href="https://app.face2gene.com/dg/lmd/912/photo/10163">https://app.face2gene.com/dg/lmd/912/photo/10163</a>	1	1.0
270	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/18749">https://app.face2gene.com/dg/lmd/3562/photo/18749</a>	4	20.8
272	Bardet-Biedl Syndrome	<a href="https://app.face2gene.com/dg/lmd/13278/photo/41796">https://app.face2gene.com/dg/lmd/13278/photo/41796</a>	1	1.0
273	Cornelia De Lange Syndrome	<a href="https://app.face2gene.com/dg/lmd/423/photo/41807">https://app.face2gene.com/dg/lmd/423/photo/41807</a>	1	1.5
275	Crouzon Syndrome	<a href="https://app.face2gene.com/dg/lmd/383/photo/41828">https://app.face2gene.com/dg/lmd/383/photo/41828</a>	1	1.0
277	Rubinstein-Taybi Syndrome	<a href="https://app.face2gene.com/dg/lmd/1490/photo/41858">https://app.face2gene.com/dg/lmd/1490/photo/41858</a>	1	1.0
278	Rubinstein-Taybi Syndrome	<a href="https://app.face2gene.com/dg/lmd/1490/photo/41860">https://app.face2gene.com/dg/lmd/1490/photo/41860</a>	1	1.0
279	Mucopolysaccharidosis, Type II; MPS2	<a href="https://app.face2gene.com/dg/lmd/801/photo/41877">https://app.face2gene.com/dg/lmd/801/photo/41877</a>	1	1.0
280	Trichorhinophalangeal Syndrome	<a href="https://app.face2gene.com/dg/lmd/1730/photo/41886">https://app.face2gene.com/dg/lmd/1730/photo/41886</a>	1	1.0
281	Trichorhinophalangeal Syndrome	<a href="https://app.face2gene.com/dg/lmd/1730/photo/41887">https://app.face2gene.com/dg/lmd/1730/photo/41887</a>	1	1.0
282	Cleidocranial Dysplasia; CCD	<a href="https://app.face2gene.com/dg/lmd/13331/photo/41900">https://app.face2gene.com/dg/lmd/13331/photo/41900</a>	32	3.1
283	Saethre-Chotzen Syndrome; SCS	<a href="https://app.face2gene.com/dg/lmd/1504/photo/41910">https://app.face2gene.com/dg/lmd/1504/photo/41910</a>	42	1.0

284	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41961">https://app.face2gene.com/dg/lmd/1720/photo/41961</a>	1	1.0
286	Frontometaphyseal Dysplasia; FMD	<a href="https://app.face2gene.com/dg/lmd/622/photo/41685">https://app.face2gene.com/dg/lmd/622/photo/41685</a>	1	1.0
287	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/2569">https://app.face2gene.com/dg/lmd/893/photo/2569</a>	1	1.0
288	Nijmegen Breakage Syndrome; NBS	<a href="https://app.face2gene.com/dg/lmd/347/photo/4728">https://app.face2gene.com/dg/lmd/347/photo/4728</a>	7	1.0
289	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/978">https://app.face2gene.com/dg/lmd/328/photo/978</a>	1	1.0
290	Hutchinson-Gilford Progeria Syndrome; HGPS	<a href="https://app.face2gene.com/dg/lmd/1396/photo/4126">https://app.face2gene.com/dg/lmd/1396/photo/4126</a>	1	1.0
291	Costello Syndrome; CSTLO	<a href="https://app.face2gene.com/dg/lmd/2014/photo/6166">https://app.face2gene.com/dg/lmd/2014/photo/6166</a>	2	1.0
292	Kabuki Syndrome	<a href="https://app.face2gene.com/dg/lmd/893/photo/13115">https://app.face2gene.com/dg/lmd/893/photo/13115</a>	1	1.0
293	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/4716/photo/14088">https://app.face2gene.com/dg/lmd/4716/photo/14088</a>	7	2.4
294	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/4716/photo/14090">https://app.face2gene.com/dg/lmd/4716/photo/14090</a>	3	1.2
295	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/4716/photo/14091">https://app.face2gene.com/dg/lmd/4716/photo/14091</a>	2	1.0
296	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/4716/photo/14092">https://app.face2gene.com/dg/lmd/4716/photo/14092</a>	1	1.0
297	KBG Syndrome; KBGS	<a href="https://app.face2gene.com/dg/lmd/912/photo/15603">https://app.face2gene.com/dg/lmd/912/photo/15603</a>	1	1.0
298	Opitz GBBB Syndrome, Type II; GBBB2	<a href="https://app.face2gene.com/dg/lmd/640/photo/15945">https://app.face2gene.com/dg/lmd/640/photo/15945</a>	3	1.0
299	Gapo Syndrome	<a href="https://app.face2gene.com/dg/lmd/642/photo/16435">https://app.face2gene.com/dg/lmd/642/photo/16435</a>	3	1.0
300	Velocardiofacial Syndrome	<a href="https://app.face2gene.com/dg/lmd/1762/photo/19049">https://app.face2gene.com/dg/lmd/1762/photo/19049</a>	13	1.0
301	Velocardiofacial Syndrome	<a href="https://app.face2gene.com/dg/lmd/1762/photo/5395">https://app.face2gene.com/dg/lmd/1762/photo/5395</a>	21	1.0
302	Trichorhinophalangeal Syndrome	<a href="https://app.face2gene.com/dg/lmd/1730/photo/5284">https://app.face2gene.com/dg/lmd/1730/photo/5284</a>	1	1.0
304	Noonan Syndrome	<a href="https://app.face2gene.com/dg/lmd/1229/photo/3582">https://app.face2gene.com/dg/lmd/1229/photo/3582</a>	26	2.3
305	Craniofrontonasal Syndrome; CFNS	<a href="https://app.face2gene.com/dg/lmd/376/photo/1114">https://app.face2gene.com/dg/lmd/376/photo/1114</a>	1	1.0
306	Trichorhinophalangeal Syndrome	<a href="https://app.face2gene.com/dg/lmd/1730/photo/5282">https://app.face2gene.com/dg/lmd/1730/photo/5282</a>	1	1.0
307	Alagille Syndrome	<a href="https://app.face2gene.com/dg/lmd/110/photo/349">https://app.face2gene.com/dg/lmd/110/photo/349</a>	1	1.0
308	Smith-Lemli-Opitz Syndrome; SLOS	<a href="https://app.face2gene.com/dg/lmd/1608/photo/12105">https://app.face2gene.com/dg/lmd/1608/photo/12105</a>	2	1.0
309	Opitz GBBB Syndrome, Type II; GBBB2	<a href="https://app.face2gene.com/dg/lmd/640/photo/15943">https://app.face2gene.com/dg/lmd/640/photo/15943</a>	1	1.0
312	Cornelia de Lange Syndrome 1; CDLS1	<a href="https://app.face2gene.com/dg/lmd/423/photo/17342">https://app.face2gene.com/dg/lmd/423/photo/17342</a>	1	2.0
313	Cranioetaphyseal Dysplasia	<a href="https://app.face2gene.com/dg/lmd/371/photo/7667">https://app.face2gene.com/dg/lmd/371/photo/7667</a>	5	1.0
314	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/17870">https://app.face2gene.com/dg/lmd/328/photo/17870</a>	1	1.0
315	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/17868">https://app.face2gene.com/dg/lmd/328/photo/17868</a>	6	1.0
316	Coffin-Lowry Syndrome; CLS	<a href="https://app.face2gene.com/dg/lmd/328/photo/17868">https://app.face2gene.com/dg/lmd/328/photo/17868</a>	60	3.9
317	Branchiooculofacial Syndrome; BOFS	<a href="https://app.face2gene.com/dg/lmd/633/photo/9227">https://app.face2gene.com/dg/lmd/633/photo/9227</a>	17	1.1
318	Smith-Magenis Syndrome; SMS	<a href="https://app.face2gene.com/dg/lmd/3562/photo/18747">https://app.face2gene.com/dg/lmd/3562/photo/18747</a>	2	4.0
319	Oculodentodigital Dysplasia	<a href="https://app.face2gene.com/dg/lmd/1246/photo/19085">https://app.face2gene.com/dg/lmd/1246/photo/19085</a>	8	1.0
320	Birk-Barel Mental Retardation Dysmorphism Syndrome	<a href="https://app.face2gene.com/dg/lmd/5569/photo/19317">https://app.face2gene.com/dg/lmd/5569/photo/19317</a>	15	1.0
322	Cranioetaphyseal Dysplasia	<a href="https://app.face2gene.com/dg/lmd/371/photo/41763">https://app.face2gene.com/dg/lmd/371/photo/41763</a>	1	1.0

323	Rubinstein-Taybi Syndrome	<a href="https://app.face2gene.com/dg/lmd/1490/photo/41843">https://app.face2gene.com/dg/lmd/1490/photo/41843</a>	1	1.0
324	Seckel Syndrome	<a href="https://app.face2gene.com/dg/lmd/739/photo/41855">https://app.face2gene.com/dg/lmd/739/photo/41855</a>	1	1.0
326	Crouzon Syndrome	<a href="https://app.face2gene.com/dg/lmd/383/photo/41942">https://app.face2gene.com/dg/lmd/383/photo/41942</a>	1	1.0
327	Treacher Collins Syndrome	<a href="https://app.face2gene.com/dg/lmd/1720/photo/41957">https://app.face2gene.com/dg/lmd/1720/photo/41957</a>	1	1.0
329	Frontometaphyseal Dysplasia; FMD	<a href="https://app.face2gene.com/dg/lmd/622/photo/41569">https://app.face2gene.com/dg/lmd/622/photo/41569</a>	1	1.0