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SUPPLEMENTARY INFORMATION

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Identifying facial phenotypes of genetic disorders using deep learning

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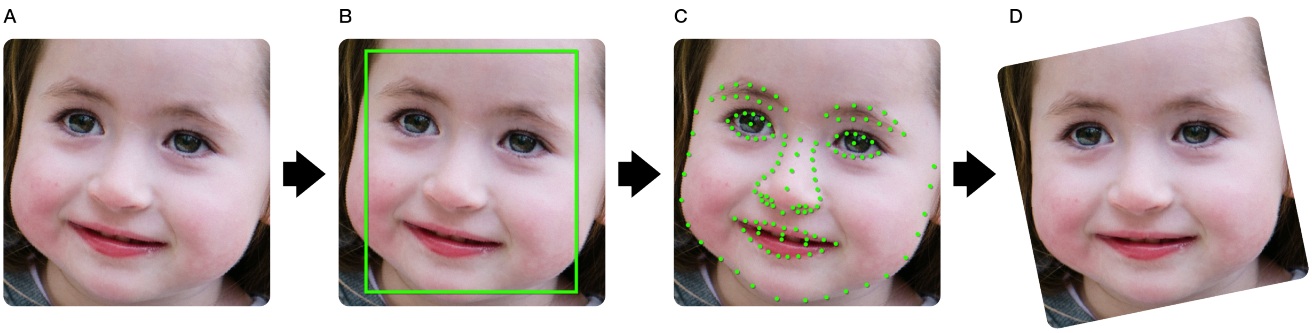
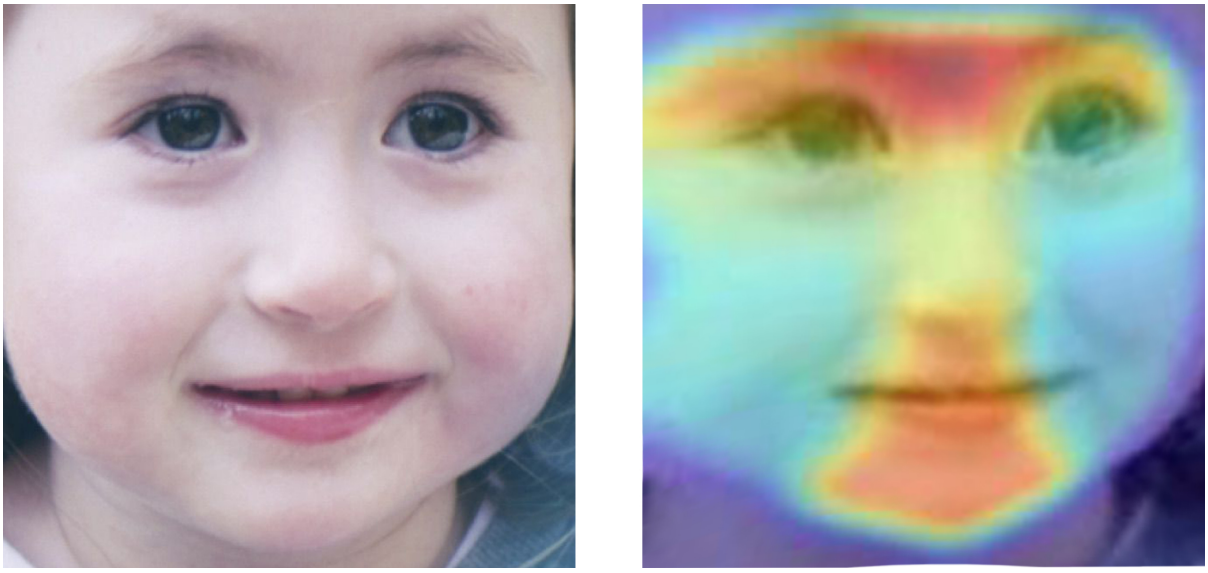


Supplementary Figure 1: Image Preprocessing Pipeline: (A) Input image. (B) Face detection rectangle output. (C) 130 facial landmarks output. (D) The detected face and landmarks are applied to geometrically normalize the patient facial image. Photo published with parental consent.



Supplementary Figure 2: Heatmap visualization showing the spatial correlation between the patient's facial photo and a specific selected output syndrome. We use the output score of a syndrome and back-propagate the error gradient through the Deep Convolutional Neural Network all the way to the input image. This process aggregates the impact of neurons on the input along the network and when reaching the input image, is able to show the contribution of each pixel on the network's decision on the score of the selected syndrome [1]. We use this property, with some prior knowledge on the facial areas, to visualize the facial correlation to a selected syndrome. Although the output can't be interpreted as specific facial features, it alludes to the features that contributed significantly to the syndrome's score. In the example of this heatmap we selected the molecularly diagnosed MRXSB Syndrome (OMIM #300986, https://omim.org/entry/300986?search=bain&highlight=bain). The typical features associated with MRXSB Syndrome, are about the inner and outer canthal regions, philtrum and mouth, exactly as highlighted by the red color of the heatmap. http://compbio.charite.de/hpoweb/showterm?id=HP:0000118#disease=OMIM:300986. Photo published with parental consent.

	Number of Genetic Disorders	Number of Training Samples (Syndromic)	Evaluation Method	Accuracy (top-1-accuracy)
Problem 1: Single syndrome vs. other population				
Saraydemir et al. [2]	1	15	3,4-Fold Cross-Validation	97.34%
Burccin et al. [3]	1	10	51 images in a test set	95.30%
Zhao et al. [4]	1	50	Leave-One-Out	96.70%
Basel-Vanagaite et al. [5]	1	134	7 images in test set	94%
Kruszka et al. [6]	1	129	Leave-One-Out	94.30%
Kruszka et al. [7]	1	156	Leave-One-Out	94.90%
Liehr et al. [8]	2	173	10-Fold Cross-Validation	100%
Shukla et al. [9]	6	1126	5-Fold Cross-Validation	94.93 (mAP)[1]
Ferry et al. [10]	8	1363	Leave-One-Out	94.90%
Problem 2: Syndromic vs. normal				
Zhao et al. [4]	14	24	Leave-One-Out	97%
Cerrolaza et al. [11]	15	73	Leave-One-Out	95%
Shukla et al. [9]	6	1126	5-Fold Cross-Validation	98.80%
Problem 3: Multiple syndromes classification				
Loos et al. [12]	5	55	Leave-One-Out	76%
Kuru et al. [13]	15	92	Leave-One-Out	53%
Boehringer et al. [14]	10	147	10-Fold Cross-Validation	75.70%
Boehringer et al. [15]	14	202	91 images in a test set	21%
Ferry et al. [10]	8	1363	Leave-One-Out	75.60%[2]
Shukla et al. [9]	6	1126	5-Fold Cross-Validation	48%[2]

Supplementary Table 1: Related Work Comparison. Comparing different studies in the field in terms of number of genetic disorders, number of training samples, the evaluation method used to test the model and accuracy results. The comparison is separated into the three main problems addressed in the literature. Problem 1 is single syndrome vs. other population - a binary classification problem of distinguishing subjects with a specific syndrome from subjects with other syndromes. Problem 2 is syndrome vs. normal - a binary classification problem of distinguishing subjects with any syndrome from normal (unaffected) subjects. Problem 3 is multiple syndromes classification - a multi-class problem of identifying the correct syndrome from multiple possible syndromes. Our work belongs to the third type, although we also demonstrate its capabilities on the first problem.

[1]Mean Average Precision (mAP) is reported as in the original paper

[2]Calculated by us from the confusion matrix in the original paper

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	Train Set		Clinical Test Set		Publication Test Set	
Age Group	**#Images**	**%**	**#Images**	**%**	**#Images**	**%**
0-1	2095	12.25	68	13.55	0	0.00
1-6	4085	23.88	106	21.12	244	74.16
6-12	1899	11.10	57	11.35	42	12.77
12-18	1430	8.36	49	9.76	43	13.07
18-above	1102	6.44	26	5.18	0	0.00
Not Reported	6495	37.97	196	39.04	0	0.00
Sex	**#Subjects**	**%**	**#Subjects**	**%**	**#Subjects**	**%**
Female	4308	39.33	122	32.53	100	31.25
Male	5483	50.06	147	39.20	220	68.75
Not Reported	1162	10.61	106	28.27	0	0.00
Diagnosis Type	**#Subjects**	**%**	**#Subjects**	**%**	**#Subjects**	**%**
Molecularly Confirmed	4608	42.07	248	66.13	0	0.00
Not Molecularly Confirmed	6345	57.93	127	33.87	320	100.00
Ethnicity	**#Subjects**	**%**	**#Subjects**	**%**	**#Subjects**	**%**
African	183	1.67	2	0.53	4	1.25
African American	303	2.77	0	0.00	1	0.31
Arab	132	1.21	6	1.60	0	0.00
Asian	666	6.08	5	1.33	6	1.88
Caucasian	4755	43.41	101	26.93	289	90.31
Chinese	3	0.03	0	0.00	0	0.00
Hispanic	382	3.49	16	4.27	1	0.31
Not Reported	4529	41.35	245	65.33	19	5.94

Supplementary Table 2: Multi-class Gestalt Model: demographic and clinical information of the train and test sets. The Age Groups are divided by year into the following sub groups: 0-1, 1-6, 6-12, 12-18, 18-above and Not Reported. Age Groups are set either by knowing the exact age of the patient in the image or by an estimation of the relevant age group. The Age Group distribution is obtained by counting the number of images annotated for each Age Group in each set.

The Sex, Diagnosis Type and Ethnicity distributions are obtained by counting the number of subjects annotated for each type.

	Train Set		Test Set	
Age Group	**#Images**	**%**	**#Images**	**%**
0-1	128	7.56	7	21.88
1-6	456	26.93	10	31.25
6-12	149	8.80	6	18.75
12-18	108	6.38	9	28.12
18-above	44	2.60	0	0.00
Not Reported	808	47.73	0	0.00
Sex	**#Subjects**	**%**	**#Subjects**	**%**
Female	389	39.02	13	40.62
Male	440	44.13	19	59.38
Not Reported	168	16.85	0	0.00
Diagnosis Type	**#Subjects**	**%**	**#Subjects**	**%**
Molecularly Confirmed	288	28.89	15	46.88
Not Molecularly Confirmed	709	71.11	17	53.12
Ethnicity	**#Subjects**	**%**	**#Subjects**	**%**
African	7	0.70	1	3.12
African American	18	1.81	1	3.12
Arab	7	0.70	0	0.00
Asian	48	4.81	0	0.00
Caucasian	296	29.69	28	87.50
Hispanic	13	1.30	0	0.00
Not Reported	608	60.98	2	6.25

Supplementary Table 3: Binary Gestalt Model - CdLS Experiment: demographic and clinical information of the train and test sets. The Age Groups are divided by year into the following sub groups: 0-1, 1-6, 6-12, 12-18, 18-above and Not Reported. Age Groups are set either by knowing the exact age of the patient in the image or by an estimation of the relevant age group. The Age Group distribution is obtained by counting the number of images annotated for each Age Group in each set.

The Sex, Diagnosis Type and Ethnicity distributions are obtained by counting the number of subjects annotated for each type.

	Train Set		Test Set	
Age Group	**#Images**	**%**	**#Images**	**%**
0-1	274	7.98	1	4.00
1-6	930	27.07	19	76.00
6-12	381	11.09	3	12.00
12-18	227	6.61	0	0.00
18-above	145	4.22	2	8.00
Not Reported	1478	43.03	0	0.00
Sex	**#Subjects**	**%**	**#Subjects**	**%**
Female	865	36.54	13	54.17
Male	1149	48.54	11	45.83
Not Reported	353	14.91	0	0.00
Diagnosis Type	**#Subjects**	**%**	**#Subjects**	**%**
Molecularly Confirmed	999	42.21	19	79.17
Not Molecularly Confirmed	1368	57.79	5	20.83
Ethnicity	**#Subjects**	**%**	**#Subjects**	**%**
African	8	0.34	0	0.00
African American	62	2.62	0	0.00
Arab	13	0.55	0	0.00
Asian	134	5.66	2	8.33
Caucasian	861	36.38	20	83.33
Hispanic	94	3.97	0	0.00
Not Reported	1195	50.49	2	8.33

Supplementary Table 4: Binary Gestalt Model - Angelman Experiment: demographic and clinical information of the train and test sets. The Age Groups are divided by year into the following sub groups: 0-1, 1-6, 6-12, 12-18, 18-above and Not Reported. Age Groups are set either by knowing the exact age of the patient in the image or by an estimation of the relevant age group. The Age Group distribution is obtained by counting the number of images annotated for each Age Group in each set.

The Sex, Diagnosis Type and Ethnicity distributions are obtained by counting the number of subjects annotated for each type.

	Test Set	
Age Group	**#Images**	**%**
0-1	0	0.00
1-6	21	84.00
6-12	4	16.00
12-18	0	0.00
18-above	0	0.00
Not Reported	0	0.00

Sex	**#Subjects**	**%**
Female	11	44.00
Male	14	56.00

Diagnosis Type	**#Subjects**	**%**
Molecularly Confirmed	24	96.00
Not Molecularly Confirmed	1	4.00

Ethnicity	**#Subjects**	**%**
Caucasian	23	92.00
Hispanic	2	8.00

Supplementary Table 5: Specialized Gestalt Model - Noonan Experiment: demographic and clinical information of the test set. The Age Groups are divided by year into the following sub groups: 0-1, 1-6, 6-12, 12-18, 18-above and Not Reported. Age Groups are set either by knowing the exact age of the patient in the image or by an estimation of the relevant age group. The Age Group distribution is obtained by counting the number of images annotated for each Age Group in the test set.

The Sex, Diagnosis Type and Ethnicity distributions are obtained by counting the number of subjects annotated for each type.

Supplementary Table 6: Publications Dataset.

Index	Syndrome Name	OMIM ID	Links	DeepGestalt Rank
1	Alagille Syndrome	PS118450	https://app.face2gene.com/dg/lmd/110/photo/345	28
2	Nijmegen Breakage Syndrome; NBS	251260	https://app.face2gene.com/dg/lmd/347/photo/4734	2
3	Nijmegen Breakage Syndrome; NBS	251260	https://app.face2gene.com/dg/lmd/347/photo/4730	2
4	Craniodiaphyseal Dysplasia; CDD	218300	https://app.face2gene.com/dg/lmd/373/photo/1088	1
5	Noonan Syndrome	PS163950	https://app.face2gene.com/dg/lmd/1946/photo/5976	19
6	Noonan Syndrome	PS163950	https://app.face2gene.com/dg/lmd/1946/photo/5985	11
7	Smith-Lemli-Opitz Syndrome; SLOS	270400	https://app.face2gene.com/dg/lmd/1608/photo/4893	1
8	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/2567	1
9	Focal Facial Dermal Dysplasia 3, Setleis Type; FFDD3	227260	https://app.face2gene.com/dg/lmd/1570/photo/4760	5
10	Burn-Mckeown Syndrome; BMKS	608572	https://app.face2gene.com/dg/lmd/2907/photo/6717	1
11	Burn-Mckeown Syndrome; BMKS	608572	https://app.face2gene.com/dg/lmd/2907/photo/6716	1
12	Burn-Mckeown Syndrome; BMKS	608572	https://app.face2gene.com/dg/lmd/2907/photo/6719	18
13	Burn-Mckeown Syndrome; BMKS	608572	https://app.face2gene.com/dg/lmd/2907/photo/6720	1
14	Meier-Gorlin Syndrome 1; MGORS1	224690	https://app.face2gene.com/dg/lmd/808/photo/2269	30
15	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/5615	1
16	Trichorhinophalangeal Syndrome, Type II; TRPS2	150230	https://app.face2gene.com/dg/lmd/971/photo/2778	1
17	Acrocallosal Syndrome; ACLS	200990	https://app.face2gene.com/dg/lmd/23/photo/80	7

18	Cohen Syndrome; COH1	216550	https://app.face2gene.com/dg/lmd/333/photo/1003	1
19	Schwartz-Jampel Syndrome, Type 1; SJS1	255800	https://app.face2gene.com/dg/lmd/1548/photo/4685	9
20	Treacher Collins Syndrome 1; TCS1	154500	https://app.face2gene.com/dg/lmd/1720/photo/5245	1
21	Trichorhinophalangeal Syndrome	PS190350	https://app.face2gene.com/dg/lmd/1730/photo/5290	1
22	Velocardiofacial Syndrome	192430	https://app.face2gene.com/dg/lmd/1762/photo/8285	3
23	Smith-Lemli-Opitz Syndrome; SLOS	270400	https://app.face2gene.com/dg/lmd/1608/photo/8342	1
24	Marfan Syndrome; MFS	154700	https://app.face2gene.com/dg/lmd/1057/photo/8255	1
25	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/5603	1
26	Mowat-Wilson Syndrome; MOWS	235730	https://app.face2gene.com/dg/lmd/4211/photo/13183	1
27	Mowat-Wilson Syndrome; MOWS	235730	https://app.face2gene.com/dg/lmd/4211/photo/13743	1
28	Mowat-Wilson Syndrome; MOWS	235730	https://app.face2gene.com/dg/lmd/4211/photo/13744	1
29	3MC Syndrome 3; 3MC3	248340	https://app.face2gene.com/dg/lmd/1048/photo/14730	2
30	Sotos Syndrome 2; SOTOS2	614753	https://app.face2gene.com/dg/lmd/1617/photo/15149	1
31	Sotos Syndrome 2; SOTOS2	614753	https://app.face2gene.com/dg/lmd/1617/photo/15152	1
32	Treacher Collins Syndrome 1; TCS1	154500	https://app.face2gene.com/dg/lmd/1720/photo/5227	1
33	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/958	1
34	Phelan-Mcdermid Syndrome; PHMDS	606232	https://app.face2gene.com/dg/lmd/4459/photo/16283	2
35	Mowat-Wilson Syndrome; MOWS	235730	https://app.face2gene.com/dg/lmd/4211/photo/16768	1
36	Donnai-Barrow Syndrome	222448	https://app.face2gene.com/dg/lmd/3473/photo/16950	49
37	Floating-Harbor Syndrome; FLHS	136140	https://app.face2gene.com/dg/lmd/596/photo/17148	17

38	Pitt-Hopkins Syndrome; PTHS	610954	https://app.face2gene.com/dg/lmd/8892/photo/17151	1
39	Marshall-Smith Syndrome; MRSHSS	602535	https://app.face2gene.com/dg/lmd/7513/photo/17204	1
40	3MC Syndrome 3; 3MC3	248340	https://app.face2gene.com/dg/lmd/1048/photo/17445	110
41	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/5608	1
42	Waardenburg Syndrome, Type 1; WS1	193500	https://app.face2gene.com/dg/lmd/1781/photo/5438	37
43	Nasopalpebral lipoma-coloboma syndrome; NPLCS	167730	https://app.face2gene.com/dg/lmd/1208/photo/8174	1
44	Sotos Syndrome 2; SOTOS2	614753	https://app.face2gene.com/dg/lmd/1617/photo/4945	1
45	Pitt-Hopkins Syndrome; PTHS	610954	https://app.face2gene.com/dg/lmd/8892/photo/17999	1
46	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/18077	1
47	Waardenburg Syndrome, Type 1; WS1	193500	https://app.face2gene.com/dg/lmd/1781/photo/8786	1
48	Waardenburg Syndrome, Type 1; WS1	193500	https://app.face2gene.com/dg/lmd/1781/photo/8794	25
49	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/9086	1
50	Holoprosencephaly	PS236100	https://app.face2gene.com/dg/lmd/787/photo/9105	10
51	Seckel Syndrome	PS210600	https://app.face2gene.com/dg/lmd/739/photo/9429	2
52	Rothmund-Thomson Syndrome; RTS	268400	https://app.face2gene.com/dg/lmd/1487/photo/9782	66
53	Aarskog-Scott Syndrome; AAS	305400	https://app.face2gene.com/dg/lmd/2/photo/9962	1
54	Coffin-Siris Syndrome1; CSS1	135900	https://app.face2gene.com/dg/lmd/329/photo/18535	15
55	Bohring-Opitz Syndrome; BOPS	605039	https://app.face2gene.com/dg/lmd/4337/photo/18830	1
56	SHORT syndrome	269880	https://app.face2gene.com/dg/lmd/1574/photo/4787	1

57	Schwartz-Jampel Syndrome, Type 1; SJS1	255800	https://app.face2gene.com/dg/lmd/1548/photo/4686	4
58	Hajdu-Cheney Syndrome; HJCYS	102500	https://app.face2gene.com/dg/lmd/722/photo/2067	1
59	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/967	1
60	Baraitser-Winter Syndrome 1; BRWS1	243310	https://app.face2gene.com/dg/lmd/149/photo/430	2
61	Beckwith-Wiedemann Syndrome; BWS	130650	https://app.face2gene.com/dg/lmd/173/photo/480	3
62	Baraitser-Winter Syndrome 1; BRWS1	243310	https://app.face2gene.com/dg/lmd/149/photo/432	2
63	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/279	1
64	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/268	1
65	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/7322/photo/276	1
66	Sotos Syndrome 2; SOTOS2	614753	https://app.face2gene.com/dg/lmd/1617/photo/4944	1
67	Cockayne Syndrome	PS216400	https://app.face2gene.com/dg/lmd/326/photo/946	1
68	Acromicric Dysplasia	102370	https://app.face2gene.com/dg/lmd/30/photo/145	24
69	Fetal Alcohol Syndrome; FAS	-	https://app.face2gene.com/dg/lmd/571/photo/1612	8
70	Ectodermal Dysplasia 1, Hypohidrotic, X-Linked; XHED	305100	https://app.face2gene.com/dg/lmd/508/photo/1407	1
71	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/960	6
72	Mucolipidosis II Alpha/beta	252500	https://app.face2gene.com/dg/lmd/838/photo/2386	5
73	Beckwith-Wiedemann Syndrome; BWS	130650	https://app.face2gene.com/dg/lmd/173/photo/484	6
74	Craniodiaphyseal Dysplasia; CDD	218300	https://app.face2gene.com/dg/lmd/373/photo/1093	2
75	Cockayne Syndrome	PS216400	https://app.face2gene.com/dg/lmd/326/photo/948	3
76	Floating-Harbor Syndrome; FLHS	136140	https://app.face2gene.com/dg/lmd/596/photo/1761	1

77	Rubinstein-Taybi Syndrome 1; RSTS1	180849	https://app.face2gene.com/dg/lmd/1490/photo/4474	1
78	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/970	1
79	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/971	1
80	Apert Syndrome	101200	https://app.face2gene.com/dg/lmd/99/photo/326	1
81	Branchiooculofacial Syndrome; BOFS	113620	https://app.face2gene.com/dg/lmd/633/photo/8345	1
82	Smith-Lemli-Opitz Syndrome; SLOS	270400	https://app.face2gene.com/dg/lmd/1608/photo/12105	1
83	Smith-Lemli-Opitz Syndrome; SLOS	270400	https://app.face2gene.com/dg/lmd/1608/photo/12105	1
84	Coffin-Siris Syndrome1; CSS1	135900	https://app.face2gene.com/dg/lmd/329/photo/12535	3
85	Coffin-Siris Syndrome1; CSS1	135900	https://app.face2gene.com/dg/lmd/329/photo/12535	2
86	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/13114	1
87	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/13116	1
88	Mowat-Wilson Syndrome; MOWS	235730	https://app.face2gene.com/dg/lmd/4211/photo/13734	1
89	Mowat-Wilson Syndrome; MOWS	235730	https://app.face2gene.com/dg/lmd/4211/photo/13736	58
90	Mowat-Wilson Syndrome; MOWS	235730	https://app.face2gene.com/dg/lmd/4211/photo/13742	1
91	Borjeson-Forssman-Lehmann Syndrome; BFLS	301900	https://app.face2gene.com/dg/lmd/216/photo/14759	56
92	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/14969	4
93	Aarskog-Scott Syndrome; AAS	305400	https://app.face2gene.com/dg/lmd/2/photo/15758	1
94	Aarskog-Scott Syndrome; AAS	305400	https://app.face2gene.com/dg/lmd/2/photo/15758	1

95	Opitz GBBB Syndrome, Type II; GBBB2	145410	https://app.face2gene.com/dg/lmd/640/photo/15940	9
96	Silver-Russell Syndrome; SRS	180860	https://app.face2gene.com/dg/lmd/1495/photo/16387	1
97	Phelan-Mcdermid Syndrome; PHMDS	606232	https://app.face2gene.com/dg/lmd/4459/photo/16512	1
98	Sotos Syndrome 2; SOTOS2	614753	https://app.face2gene.com/dg/lmd/1617/photo/17023	1
99	Sotos Syndrome 2; SOTOS2	614753	https://app.face2gene.com/dg/lmd/1617/photo/17024	10
100	CHARGE Syndrome	214800	https://app.face2gene.com/dg/lmd/301/photo/17094	2
101	Floating-Harbor Syndrome; FLHS	136140	https://app.face2gene.com/dg/lmd/596/photo/17124	1
102	Prader-Willi Syndrome; PWS	176270	https://app.face2gene.com/dg/lmd/1388/photo/4095	1
103	Craniometaphyseal Dysplasia	PS123000	https://app.face2gene.com/dg/lmd/371/photo/7667	22
104	Craniosynostosis, Adelaide Type; CRSA	600593	https://app.face2gene.com/dg/lmd/3793/photo/7990	101
105	Pitt-Hopkins Syndrome; PTHS	610954	https://app.face2gene.com/dg/lmd/8892/photo/18000	1
106	Baraitser-Winter Syndrome 1; BRWS1	243310	https://app.face2gene.com/dg/lmd/149/photo/10154	10
107	Baraitser-Winter Syndrome 1; BRWS1	243310	https://app.face2gene.com/dg/lmd/149/photo/10156	41
108	Baraitser-Winter Syndrome 1; BRWS1	243310	https://app.face2gene.com/dg/lmd/149/photo/10156	41
109	3MC Syndrome 3; 3MC3	248340	https://app.face2gene.com/dg/lmd/1048/photo/10305	2
110	Potocki-Shaffer Syndrome	601224	https://app.face2gene.com/dg/lmd/3929/photo/10549	2
111	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/18744	1
112	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/18745	4
113	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/18747	1

114	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/18749	1
115	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/263	2
116	Pierre Robin Sequence with Cleft Mandible and Limb Anomalies	268305	https://app.face2gene.com/dg/lmd/2127/photo/6384	3
117	Pierre Robin Sequence with Cleft Mandible and Limb Anomalies	268305	https://app.face2gene.com/dg/lmd/2127/photo/6385	1
118	Crouzon Syndrome	123500	https://app.face2gene.com/dg/lmd/383/photo/1133	1
119	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/265	3
120	Beckwith-Wiedemann Syndrome; BWS	130650	https://app.face2gene.com/dg/lmd/173/photo/41779	1
121	Otopalatodigital Syndrome	PS311300	https://app.face2gene.com/dg/lmd/1266/photo/41780	1
122	Waardenburg Syndrome, Type 1; WS1	193500	https://app.face2gene.com/dg/lmd/1781/photo/41781	1
123	Rubinstein-Taybi Syndrome	PS180849	https://app.face2gene.com/dg/lmd/1490/photo/41785	1
124	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41799	1
125	Cornelia De Lange Syndrome	PS122470	https://app.face2gene.com/dg/lmd/423/photo/41811	1
126	Cornelia De Lange Syndrome	PS122470	https://app.face2gene.com/dg/lmd/423/photo/41812	1
127	Cornelia De Lange Syndrome	PS122470	https://app.face2gene.com/dg/lmd/423/photo/41813	1
128	Cornelia De Lange Syndrome	PS122470	https://app.face2gene.com/dg/lmd/423/photo/41814	1
129	Sotos Syndrome	PS117550	https://app.face2gene.com/dg/lmd/1617/photo/41817	1
130	Ectodermal Dysplasia 1, Hypohidrotic, X-Linked; XHED	305100	https://app.face2gene.com/dg/lmd/508/photo/41823	2
131	Down Syndrome	190685	https://app.face2gene.com/dg/lmd/6905/photo/41830	1
132	Wolf-Hirschhorn Syndrome; WHS	194190	https://app.face2gene.com/dg/lmd/4706/photo/41832	1

133	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41840	1
134	Hutchinson-Gilford Progeria Syndrome; HGPS	176670	https://app.face2gene.com/dg/lmd/1396/photo/41849	1
135	Cri-Du-Chat Syndrome	123450	https://app.face2gene.com/dg/lmd/13213/photo/41863	1
136	Hurler Syndrome	607014	https://app.face2gene.com/dg/lmd/13858/photo/41873	1
137	Hurler Syndrome	607014	https://app.face2gene.com/dg/lmd/13858/photo/41874	1
138	Mucopolysaccharidosis, Type II; MPS2	309900	https://app.face2gene.com/dg/lmd/801/photo/41875	1
139	Mucopolysaccharidosis, Type II; MPS2	309900	https://app.face2gene.com/dg/lmd/801/photo/41876	1
140	Mucopolysaccharidosis, Type II; MPS2	309900	https://app.face2gene.com/dg/lmd/801/photo/41878	1
141	Mucopolysaccharidosis, Type II; MPS2	309900	https://app.face2gene.com/dg/lmd/801/photo/41879	1
142	Mucolipidosis II Alpha/beta	252500	https://app.face2gene.com/dg/lmd/838/photo/41883	1
143	Cleidocranial Dysplasia; CCD	119600	https://app.face2gene.com/dg/lmd/13331/photo/41890	5
144	Cleidocranial Dysplasia; CCD	119600	https://app.face2gene.com/dg/lmd/13331/photo/41891	1
145	Seckel Syndrome	PS210600	https://app.face2gene.com/dg/lmd/739/photo/41904	12
146	Silver-Russell Syndrome; SRS	180860	https://app.face2gene.com/dg/lmd/1495/photo/41913	1
147	Crouzon Syndrome	123500	https://app.face2gene.com/dg/lmd/383/photo/41914	2
148	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41923	1
149	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41924	1
150	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41925	1
151	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41926	1
152	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41927	1

153	Sotos Syndrome	PS117550	https://app.face2gene.com/dg/lmd/1617/photo/41940	1
154	Sotos Syndrome	PS117550	https://app.face2gene.com/dg/lmd/1617/photo/41941	1
155	Silver-Russell Syndrome; SRS	180860	https://app.face2gene.com/dg/lmd/1495/photo/41945	2
156	Hallermann-Streiff Syndrome; HSS	234100	https://app.face2gene.com/dg/lmd/730/photo/41946	1
157	Cleidocranial Dysplasia; CCD	119600	https://app.face2gene.com/dg/lmd/13331/photo/41947	1
158	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41956	4
159	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41958	1
160	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41964	1
161	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41965	1
162	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41966	1
163	Fragile X Mental Retardation Syndrome	300624	https://app.face2gene.com/dg/lmd/13243/photo/41973	1
164	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/41983	1
165	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/15947	1
166	Pierpont Syndrome; PRPTS	602342	https://app.face2gene.com/dg/lmd/4198/photo/41638	3
167	Pierpont Syndrome; PRPTS	602342	https://app.face2gene.com/dg/lmd/4198/photo/41640	1
168	SHORT syndrome	269880	https://app.face2gene.com/dg/lmd/1574/photo/4780	1
169	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/2566	1
170	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/2560	1
171	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/2562	8
172	Otopalatodigital Syndrome	PS311300	https://app.face2gene.com/dg/lmd/1266/photo/3719	1
173	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/977	1
174	Ohdo Syndrome, SBBYS Variant; SBBYSS	603736	https://app.face2gene.com/dg/lmd/1255/photo/8343	1
175	Rubinstein-Taybi Syndrome 1; RSTS1	180849	https://app.face2gene.com/dg/lmd/358/photo/11625	2

176	Auriculocondylar Syndrome 1; ARCND1	602483	https://app.face2gene.com/dg/lmd/4192/photo/12354	1
177	Coffin-Siris Syndrome1; CSS1	135900	https://app.face2gene.com/dg/lmd/329/photo/12530	1
178	Urofacial Syndrome 1; UFS1	236730	https://app.face2gene.com/dg/lmd/1236/photo/13084	3
179	3MC Syndrome 3; 3MC3	248340	https://app.face2gene.com/dg/lmd/1048/photo/13477	1
180	Smith-Lemli-Opitz Syndrome; SLOS	270400	https://app.face2gene.com/dg/lmd/1608/photo/14313	1
181	3MC Syndrome 3; 3MC3	248340	https://app.face2gene.com/dg/lmd/1048/photo/14727	1
182	Phelan-Mcdermid Syndrome; PHMDS	606232	https://app.face2gene.com/dg/lmd/4459/photo/16282	5
183	Lig4 Syndrome	606593	https://app.face2gene.com/dg/lmd/4654/photo/17338	16
184	Craniometaphyseal Dysplasia	PS123000	https://app.face2gene.com/dg/lmd/371/photo/1078	1
185	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/284	1
186	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/274	1
187	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/264	1
188	Craniofrontonasal Syndrome; CFNS	304110	https://app.face2gene.com/dg/lmd/376/photo/1113	1
189	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/269	1
190	Floating-Harbor Syndrome; FLHS	136140	https://app.face2gene.com/dg/lmd/596/photo/1756	1
191	Rubinstein-Taybi Syndrome 1; RSTS1	180849	https://app.face2gene.com/dg/lmd/1490/photo/4488	1
192	Coffin-Siris Syndrome1; CSS1	135900	https://app.face2gene.com/dg/lmd/329/photo/981	1
193	Rubinstein-Taybi Syndrome 1; RSTS1	180849	https://app.face2gene.com/dg/lmd/1490/photo/4489	1
194	Noonan Syndrome	PS163950	https://app.face2gene.com/dg/lmd/1229/photo/3585	1
195	Floating-Harbor Syndrome; FLHS	136140	https://app.face2gene.com/dg/lmd/596/photo/8231	1
196	Smith-Lemli-Opitz Syndrome; SLOS	270400	https://app.face2gene.com/dg/lmd/1608/photo/12105	1

197	Smith-Lemli-Opitz Syndrome; SLOS	270400	https://app.face2gene.com/dg/lmd/1608/photo/12105	2
198	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/13113	1
199	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/13113	1
200	Sotos Syndrome 2; SOTOS2	614753	https://app.face2gene.com/dg/lmd/1617/photo/15150	1
201	Phelan-Mcdermid Syndrome; PHMDS	606232	https://app.face2gene.com/dg/lmd/4459/photo/16512	1
202	Phelan-Mcdermid Syndrome; PHMDS	606232	https://app.face2gene.com/dg/lmd/4459/photo/16512	1
203	Sotos Syndrome 2; SOTOS2	614753	https://app.face2gene.com/dg/lmd/1617/photo/17025	1
204	Potocki-Shaffer Syndrome	601224	https://app.face2gene.com/dg/lmd/3929/photo/10549	9
205	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/18745	1
206	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/18745	1
207	Birk-Barel Mental Retardation Dysmorphism Syndrome	612292	https://app.face2gene.com/dg/lmd/5569/photo/19317	12
208	Blepharophimosis, Ptosis, and Epicanthus Inversus; BPES	110100	https://app.face2gene.com/dg/lmd/202/photo/580	81
209	Focal Dermal Hypoplasia; FDH	305600	https://app.face2gene.com/dg/lmd/13389/photo/41764	1
210	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41783	1
211	Williams-Beuren Syndrome; WBS	194050	https://app.face2gene.com/dg/lmd/1829/photo/41784	1
212	Moebius Syndrome; MBS	157900	https://app.face2gene.com/dg/lmd/1150/photo/41804	1
213	Moebius Syndrome; MBS	157900	https://app.face2gene.com/dg/lmd/1150/photo/41805	1
214	Cornelia De Lange Syndrome	PS122470	https://app.face2gene.com/dg/lmd/423/photo/41808	1
215	Cornelia De Lange Syndrome	PS122470	https://app.face2gene.com/dg/lmd/423/photo/41810	1
216	Sotos Syndrome	PS117550	https://app.face2gene.com/dg/lmd/1617/photo/41818	1

217	Moebius Syndrome; MBS	157900	https://app.face2gene.com/dg/lmd/1150/photo/41821	1
218	Focal Dermal Hypoplasia; FDH	305600	https://app.face2gene.com/dg/lmd/13389/photo/41822	6
219	Mucopolysaccharidosis, Type II; MPS2	309900	https://app.face2gene.com/dg/lmd/801/photo/41824	3
220	Prader-Willi Syndrome; PWS	176270	https://app.face2gene.com/dg/lmd/1388/photo/41837	1
221	Crouzon Syndrome	123500	https://app.face2gene.com/dg/lmd/383/photo/41842	1
222	Rubinstein-Taybi Syndrome	PS180849	https://app.face2gene.com/dg/lmd/1490/photo/41844	1
223	Hutchinson-Gilford Progeria Syndrome; HGPS	176670	https://app.face2gene.com/dg/lmd/1396/photo/41845	1
224	Hutchinson-Gilford Progeria Syndrome; HGPS	176670	https://app.face2gene.com/dg/lmd/1396/photo/41846	1
225	Hutchinson-Gilford Progeria Syndrome; HGPS	176670	https://app.face2gene.com/dg/lmd/1396/photo/41847	1
226	Hutchinson-Gilford Progeria Syndrome; HGPS	176670	https://app.face2gene.com/dg/lmd/1396/photo/41848	1
227	Hutchinson-Gilford Progeria Syndrome; HGPS	176670	https://app.face2gene.com/dg/lmd/1396/photo/41850	1
228	Noonan Syndrome	PS163950	https://app.face2gene.com/dg/lmd/1229/photo/41851	1
229	Down Syndrome	190685	https://app.face2gene.com/dg/lmd/6905/photo/41853	1
230	Down Syndrome	190685	https://app.face2gene.com/dg/lmd/6905/photo/41854	1
231	Rubinstein-Taybi Syndrome	PS180849	https://app.face2gene.com/dg/lmd/1490/photo/41859	1
232	Achondroplasia; ACH	100800	https://app.face2gene.com/dg/lmd/17/photo/41871	9
233	Hurler Syndrome	607014	https://app.face2gene.com/dg/lmd/13858/photo/41872	1
234	Cleidocranial Dysplasia; CCD	119600	https://app.face2gene.com/dg/lmd/13331/photo/41899	1
235	Silver-Russell Syndrome; SRS	180860	https://app.face2gene.com/dg/lmd/1495/photo/41908	1
236	Saethre-Chotzen Syndrome; SCS	101400	https://app.face2gene.com/dg/lmd/1504/photo/41909	1
237	Cleidocranial Dysplasia; CCD	119600	https://app.face2gene.com/dg/lmd/13331/photo/41948	7
238	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41951	1

239	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41952	1
240	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41954	1
241	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41959	1
242	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41960	1
243	Fragile X Mental Retardation Syndrome	300624	https://app.face2gene.com/dg/lmd/13243/photo/41982	1
244	Pitt-Hopkins Syndrome; PTHS	610954	https://app.face2gene.com/dg/lmd/8892/photo/18003	1
245	Nijmegen Breakage Syndrome; NBS	251260	https://app.face2gene.com/dg/lmd/347/photo/4723	1
246	Nijmegen Breakage Syndrome; NBS	251260	https://app.face2gene.com/dg/lmd/347/photo/4736	13
247	Noonan Syndrome	163950	https://app.face2gene.com/dg/lmd/1946/photo/5983	2
248	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/2565	1
249	Craniodiaphyseal Dysplasia; CDD	218300	https://app.face2gene.com/dg/lmd/373/photo/1091	1
250	Ectodermal Dysplasia 1, Hypohidrotic, X-Linked; XHED	305100	https://app.face2gene.com/dg/lmd/508/photo/1400	1
251	Mowat-Wilson Syndrome; MOWS	235730	https://app.face2gene.com/dg/lmd/4211/photo/13735	1
252	3MC Syndrome 3; 3MC3	248340	https://app.face2gene.com/dg/lmd/1048/photo/13467	1
253	Auriculocondylar Syndrome 1; ARCND1	602483	https://app.face2gene.com/dg/lmd/4192/photo/14125	2
254	Nicolaides-Baraitser Syndrome; NCBRS	601358	https://app.face2gene.com/dg/lmd/3394/photo/14255	1
255	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/14454	1
256	Nicolaides-Baraitser Syndrome; NCBRS	601358	https://app.face2gene.com/dg/lmd/3394/photo/14700	1
257	Noonan Syndrome	163950	https://app.face2gene.com/dg/lmd/1229/photo/3581	1
258	KBG Syndrome; KBGS	148050	https://app.face2gene.com/dg/lmd/912/photo/15604	1
259	Angelman Syndrome; AS	105830	https://app.face2gene.com/dg/lmd/81/photo/272	1

260	Aarskog-Scott Syndrome; AAS	305400	https://app.face2gene.com/dg/lmd/2/photo/9965	1
261	Beckwith-Wiedemann Syndrome; BWS	130650	https://app.face2gene.com/dg/lmd/173/photo/481	6
262	Craniofrontonasal Syndrome; CFNS	304110	https://app.face2gene.com/dg/lmd/376/photo/1115	1
263	Floating-Harbor Syndrome; FLHS	136140	https://app.face2gene.com/dg/lmd/596/photo/1760	1
264	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/974	1
265	Sotos Syndrome 2; SOTOS2	614753	https://app.face2gene.com/dg/lmd/1617/photo/15146	1
266	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/15950	1
267	Phelan-Mcdermid Syndrome; PHMDS	606232	https://app.face2gene.com/dg/lmd/4459/photo/16512	5
268	Lateral Meningocele Syndrome; LMNS	130720	https://app.face2gene.com/dg/lmd/983/photo/10028	3
269	KBG Syndrome; KBGS	148050	https://app.face2gene.com/dg/lmd/912/photo/10163	1
270	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/18749	4
271	Aarskog Syndrome	PS305400	https://app.face2gene.com/dg/lmd/2/photo/41775	1
272	Bardet-Biedl Syndrome	PS209900	https://app.face2gene.com/dg/lmd/13278/photo/41796	1
273	Cornelia De Lange Syndrome	PS122470	https://app.face2gene.com/dg/lmd/423/photo/41807	1
274	Cri-Du-Chat Syndrome	123450	https://app.face2gene.com/dg/lmd/13213/photo/41826	1
275	Crouzon Syndrome	123500	https://app.face2gene.com/dg/lmd/383/photo/41828	1
276	Prader-Willi Syndrome; PWS	176270	https://app.face2gene.com/dg/lmd/1388/photo/41911	1
277	Rubinstein-Taybi Syndrome	PS180849	https://app.face2gene.com/dg/lmd/1490/photo/41858	1
278	Rubinstein-Taybi Syndrome	PS180849	https://app.face2gene.com/dg/lmd/1490/photo/41860	1
279	Mucopolysaccharidosis, Type II; MPS2	309900	https://app.face2gene.com/dg/lmd/801/photo/41877	1
280	Trichorhinophalangeal Syndrome	PS190350	https://app.face2gene.com/dg/lmd/1730/photo/41886	1

281	Trichorhinophalangeal Syndrome	PS190350	https://app.face2gene.com/dg/lmd/1730/photo/41887	1
282	Cleidocranial Dysplasia; CCD	119600	https://app.face2gene.com/dg/lmd/13331/photo/41900	32
283	Saethre-Chotzen Syndrome; SCS	101400	https://app.face2gene.com/dg/lmd/1504/photo/41910	42
284	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41961	1
285	Sifrim-Hitz-Weiss syndrome; SIHIWES	617159	https://app.face2gene.com/dg/lmd/13594/photo/41701	6
286	Frontometaphyseal Dysplasia; FMD	305620	https://app.face2gene.com/dg/lmd/622/photo/41685	1
287	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/2569	1
288	Nijmegen Breakage Syndrome; NBS	251260	https://app.face2gene.com/dg/lmd/347/photo/4728	7
289	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/978	1
290	Hutchinson-Gilford Progeria Syndrome; HGPS	176670	https://app.face2gene.com/dg/lmd/1396/photo/4126	1
291	Costello Syndrome; CSTLO	218040	https://app.face2gene.com/dg/lmd/2014/photo/6166	2
292	Kabuki Syndrome	PS147920	https://app.face2gene.com/dg/lmd/893/photo/13115	1
293	Noonan Syndrome	PS163950	https://app.face2gene.com/dg/lmd/4716/photo/14088	7
294	Noonan Syndrome	PS163950	https://app.face2gene.com/dg/lmd/4716/photo/14090	3
295	Noonan Syndrome	PS163950	https://app.face2gene.com/dg/lmd/4716/photo/14091	2
296	Noonan Syndrome	PS163950	https://app.face2gene.com/dg/lmd/4716/photo/14092	1
297	KBG Syndrome; KBGS	148050	https://app.face2gene.com/dg/lmd/912/photo/15603	1
298	Opitz GBBB Syndrome, Type II; GBBB2	145410	https://app.face2gene.com/dg/lmd/640/photo/15945	3
299	Gapo Syndrome	230740	https://app.face2gene.com/dg/lmd/642/photo/16435	3
300	Velocardiofacial Syndrome	192430	https://app.face2gene.com/dg/lmd/1762/photo/19049	13
301	Velocardiofacial Syndrome	192430	https://app.face2gene.com/dg/lmd/1762/photo/5395	21
302	Trichorhinophalangeal Syndrome	PS190350	https://app.face2gene.com/dg/lmd/1730/photo/5284	1

303	Blepharophimosis, Ptosis, and Epicanthus Inversus; BPES	110100	https://app.face2gene.com/dg/lmd/202/photo/581	42
304	Noonan Syndrome	PS163950	https://app.face2gene.com/dg/lmd/1229/photo/3582	26
305	Craniofrontonasal Syndrome; CFNS	304110	https://app.face2gene.com/dg/lmd/376/photo/1114	1
306	Trichorhinophalangeal Syndrome	PS190350	https://app.face2gene.com/dg/lmd/1730/photo/5282	1
307	Alagille Syndrome	PS118450	https://app.face2gene.com/dg/lmd/110/photo/349	1
308	Smith-Lemli-Opitz Syndrome; SLOS	270400	https://app.face2gene.com/dg/lmd/1608/photo/12105	2
309	Opitz GBBB Syndrome, Type II; GBBB2	145410	https://app.face2gene.com/dg/lmd/640/photo/15943	1
310	Lubs X-Linked Mental Retardation Syndrome; MRXSL	300260	https://app.face2gene.com/dg/lmd/5107/photo/16174	19
311	Phelan-Mcdermid Syndrome; PHMDS	606232	https://app.face2gene.com/dg/lmd/4459/photo/16512	1
312	Cornelia de Lange Syndrome 1; CDLS1	122470	https://app.face2gene.com/dg/lmd/423/photo/17342	1
313	Craniometaphyseal Dysplasia	PS123000	https://app.face2gene.com/dg/lmd/371/photo/7667	5
314	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/17870	1
315	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/17868	6
316	Coffin-Lowry Syndrome; CLS	303600	https://app.face2gene.com/dg/lmd/328/photo/17868	60
317	Branchiooculofacial Syndrome; BOFS	113620	https://app.face2gene.com/dg/lmd/633/photo/9227	17
318	Smith-Magenis Syndrome; SMS	182290	https://app.face2gene.com/dg/lmd/3562/photo/18747	2
319	Oculodentodigital Dysplasia	PS164200	https://app.face2gene.com/dg/lmd/1246/photo/19085	8

320	Birk-Barel Mental Retardation Dysmorphism Syndrome	612292	https://app.face2gene.com/dg/lmd/5569/photo/19317	15
321	Blepharophimosis, Ptosis, and Epicanthus Inversus; BPES	110100	https://app.face2gene.com/dg/lmd/202/photo/580	65
322	Craniometaphyseal Dysplasia	PS123000	https://app.face2gene.com/dg/lmd/371/photo/41763	1
323	Rubinstein-Taybi Syndrome	PS180849	https://app.face2gene.com/dg/lmd/1490/photo/41843	1
324	Seckel Syndrome	PS210600	https://app.face2gene.com/dg/lmd/739/photo/41855	1
325	Prader-Willi Syndrome; PWS	176270	https://app.face2gene.com/dg/lmd/1388/photo/41911	1
326	Crouzon Syndrome	123500	https://app.face2gene.com/dg/lmd/383/photo/41942	1
327	Treacher Collins Syndrome	PS154500	https://app.face2gene.com/dg/lmd/1720/photo/41957	1
328	Fountain Syndrome	229120	https://app.face2gene.com/dg/lmd/602/photo/9695	1
329	Frontometaphyseal Dysplasia; FMD	305620	https://app.face2gene.com/dg/lmd/622/photo/41569	1

Supplementary Table 6: Description of the Public Dataset in terms of syndrome name, OMIM id, the relevant link in Face2Gene Library and DeepGestalt rank for every image, indicating the placing of the diagnosed syndrome within the short list of syndromes predicted by DeepGestalt for the reported model. In order to download the images, you should first login to Face2Gene and then follow the links in the table. We can provide this table in a .csv file format upon request.

For any questions or requests, please contact the corresponding author.