

APPENDIX I SUPPLEMENTARY MULTIMEDIA

Additional visualizations of 3D faces generated by the NF model are provided in the following multimedia files:

`Ageing_Modes.gif`: Maximally probable faces produced by the proposed NF model for different syndrome (rows) and sex (columns) conditions. The age condition is increasing with time.

`Ageing_Samples.gif`: Random samples from the proposed NF model with syndrome condition fixed to unaffected. The faces on the left half are female and those on the right half are male. The age condition is increasing with time. Samples were drawn from the latent prior with a temperature of 0.8.

APPENDIX II DEMOGRAPHICS

Figure 8 shows a detailed demographic breakdown of the 3D facial scan data.

APPENDIX III CONFUSION ANALYSIS

Table III shows, for each syndrome, the most common erroneous MAP diagnosis and its frequency from the cross validated syndrome inference experiment. In general, most confusion occurs among different syndrome classes. Unaffected patients are rarely confused for syndromic and syndromic patients are rarely confused for unaffected.

TABLE III

FOR EACH SYNDROME, THE MOST COMMON ERRONEOUS SYNDROME DIAGNOSIS AND ITS FREQUENCY FROM THE CROSS VALIDATED SYNDROME INFERENCE EXPERIMENT.

Syndrome	Most Frequent Error	Error Rate (%)
1p36 Del	Rett	11.5
22q 11 2 Del	Ehlers Danlos	9.2
4p Del Wolff-Hirschhorn	1p36 Del	8.9
5p Del Cri du Chat	Ehlers Danlos	4.7
Achondroplasia	Neurofibromatosis	4.3
Angelman	Rett	12.9
CHARGE	Neurofibromatosis	5.4
Cardiofaciocutaneous	Neurofibromatosis	14.3
Cleft Lip Palate	22q 11 2 Del	10.5
Cockayne	Russell Silver	5.0
Coffin Siris	Turner	8.3
Cohen	Rett	9.9
Cornelia de Lange	Pierre Robin Sequence	6.1
Costello	Cardiofaciocutaneous	15.6
Crouzon	Marfan	22.3
Down	Noonan	6.6
Ehlers Danlos	Neurofibromatosis	13.3
Fragile X	Stickler	12.2
Goldenhar	Marfan	8.3
Jacobsen	Phelan McDermid	9.1
Joubert	Cleft Lip Palate	7.6
Kabuki	Turner	11.8
Klinefelter	Fragile X	8.2
Loeys Dietz	Ehlers Danlos	17.0
Marfan	Ehlers Danlos	18.5
Moebius	Cleft Lip Palate	13.3
Mucopolysaccharidosis	Fragile X	7.2
Neurofibromatosis	Klinefelter	5.4
Noonan	Neurofibromatosis	16.1
Osteogenesis Imperfecta	Ehlers Danlos	25.3
Pallister Killian	Jacobsen	6.1
Phelan McDermid	Ehlers Danlos	7.4
Pierre Robin Sequence	22q 11 2 Del	12.1
Pitt Hopkins	22q 11 2 Del	11.8
Prader-Willi	Cleft Lip Palate	11.6
Rett	Ehlers Danlos	7.9
Rhizomelic Chondro Punctata	Noonan	5.9
Rubinstein Taybi	Cohen	6.2
Russell Silver	Turner	17.4
Smith Lemli Opitz	Coffin Siris	12.5
Sotos	Marfan	8.8
Stickler	Ehlers Danlos	13.5
Treacher Collins	Neurofibromatosis	9.2
Trisomy 18	CHARGE	11.1
Turner	22q 11 2 Del	4.9
Unaffected	Ehlers Danlos	2.5
Williams	Neurofibromatosis	3.9
X Linked-Hypohidrotic Ectodermal	Noonan	10.3

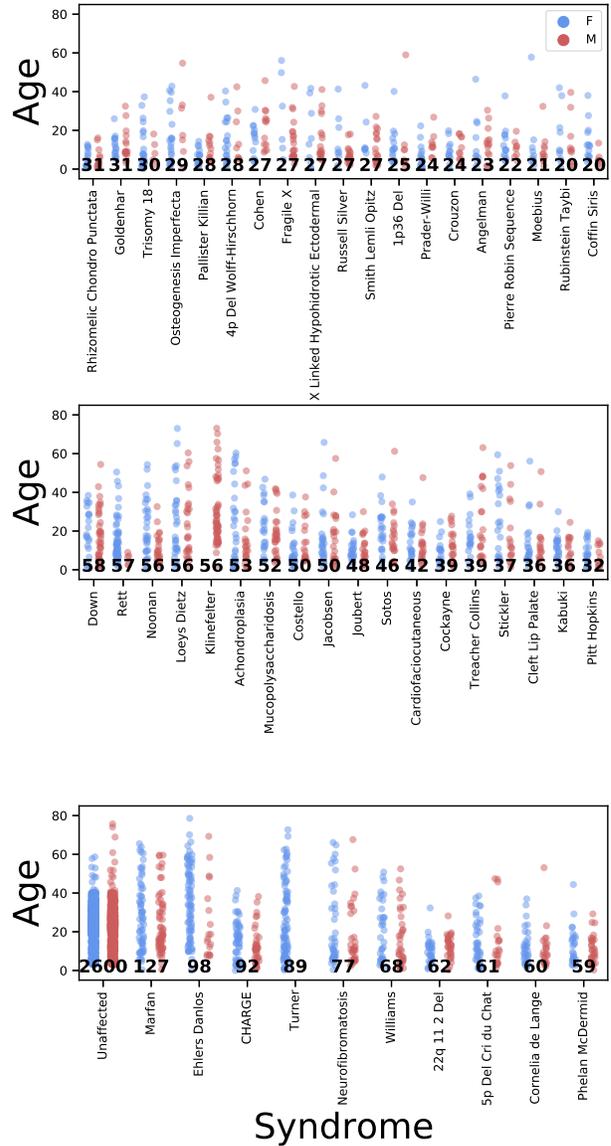


Fig. 8. The joint distribution of patient age, sex, and genetic syndrome in the 3D facial scan data.